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## O01

### Anti-CD19 CAR-T cell therapy for the treatment of severe/refractory pediatric autoimmune diseases: a single-center case series

R. Nicolai[1,*], M. Becilli[2], E. Marasco[1], P. Merli[2], V. Messia[1], F. Del Bufalo[2], M. I. Petrone[1], M. G. Cefalo[2], A. Aquilani[1], C. Rosignoli[2], G. Olivieri[1], M. Algeri[2], F. De Benedetti[2], F. Locatelli[2,3], C. Bracaglia[1]
[1]Division of Rheumatology; [2]Department of Hematology/Oncology, Cell and Gene Therapy, IRCCS Ospedale Pediatrico Bambino Gesù; [3]Catholic University of the Sacred Heart, Rome, Italy
**Correspondence:** R. Nicolai
*Pediatric Rheumatology 2026*, **24(S1):**O01

**Introduction**
CD19 CAR T-cell therapy represents an innovative and promising treatment option in patients with severe and/or refractory autoimmune diseases (AD). The experience in children and adolescents is currently limited.
**Objectives**
To report on the safety and efficacy of anti-CD19 CAR T-cell therapy in 6 patients with severe and refractory AD: 3 with Juvenile Dermatomyositis (JDM), 2 with childhood Systemic Lupus Erythematosus (cSLE) and 1 with juvenile Systemic Sclerosis (jSSc).
**Methods**
All 6 patients received a single infusion of fresh autologous anti-CD19 CAR T cells (second-generation lentiviral vector, $1\times10^6$ CAR T cells/kg), after lymphodepletion with cyclophosphamide and fludarabine.
**Results**
The median follow-up after CAR T-cell therapy is 17 months (range 1-27). Two out of 3 JDM patients had a history of severe chronically active disease with extensive calcinosis and ulcerations; the third patient presented interstitial lung disease (ILD) and persistently active muscle and skin disease with ulcerations. Both cSLE patients had ILD (one with pulmonary hypertension and one with diffuse alveolar hemorrhages) and class V nephritis. One patient had also optic neuritis. The patient with jSSc had diffuse skin involvement, vascular manifestations, ILD, gastrointestinal and cardiac involvement. All patients were refractory to multiple immunosuppressive treatment lines and presented significant side effects from chronic glucocorticoid treatment. In vivo, CAR T cells rapidly expanded, reaching a peak with a median number of 42.35 cells per microliter (range 12.86-535.02) after a median of 11 days (range 7-28). CD19+ B cells became undetectable in peripheral blood after a mean of 7 days (median 7, range 5-12). B-cell reconstitution started after a mean of 78 days (median 84, range 42-112). All patients with JDM (follow-up of 27, 12, 9 months respectively) achieved and maintained an ACR–EULAR major clinical response using the Total Improvement Score. Both cSLE patients (follow-up of 27 and 22 months) presented resolution of all signs and symptoms: SLEDAI-2K score (22 at baseline in pt 1; 38 in pt 2) normalized at month 3 (SLEDAI-2K=0). All JDM and cSLE patients are off glucocorticoids and immunosuppressant. The jSSc patient presented reduction in modified Rodnan skin score (from 26 at baseline to 12 at month 6) with stable ILD and left ventricular ejection fraction. Four patients (2 JDM and 2 cSLE) presented a grade 1 cytokine release syndrome (G1), not requiring treatment; one patient with JDM presented a mild (G1) immune effector cell–associated neurotoxicity syndrome (ICANS), characterized by confusion and transient EEG abnormalities, resolving spontaneously after 72 h. Transient anemia (range G2-G3) and neutropenia (range G3-G4) were recorded in all patients, but were limited to the first weeks after infusion. No severe infections were observed.
**Conclusion**
These data provide evidence of the short- and medium-term efficacy and safety of CD19 CAR T-cell therapy in pediatric patients with refractory AD.

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**Disclosure of interest**
R. Nicolai: None declared, M. Becilli: None declared, E. Marasco: None declared, P. Merli Consultant with: SOBI, V. Messia: None declared, F. Del Bufalo: None declared, M. Petrone: None declared, M. Cefalo: None declared, A. Aquilani: None declared, C. Rosignoli: None declared, G. Olivieri: None declared, M. Algeri: None declared, F. De Benedetti Consultant with: Abbvie, SOBI, Novimmune, Novartis, Roche, Pfizer, F. Locatelli Consultant with: Amgen, Novartis, Sanofi, Vertex, Speaker Bureau with: Miltenyi Biomedicine, Amgen, Novartis, Bristol Myers Squibb, Gilead, SOBI, C. Bracaglia Consultant with: SOBI, Novartis.

## O02

### Immunometabolic changes associated with sustained remission in patients with SLE and JDM treated with anti-CD19 CAR T cells

E. Marasco[1,*], A. Ariolli[1], N. De Franco[1], R. Nicolai[1], C. Bracaglia[1], M. Becilli[2], P. Merli[2], M. I. Petrone[1], F. Del Bufalo[2], M. Algeri[2], M. G. Cefalo[2], C. Quintarelli[2], F. Locatelli[2], F. De Benedetti[1]
[1]Rheumatology Unit; [2]Hematology Department, Ospedale Pediatrico Bambino Gesù, Rome, Italy
**Correspondence:** E. Marasco
*Pediatric Rheumatology 2026*, **24(S1):**O02

**Introduction**
CD19-CAR T cell therapy is an innovative strategy to induce profound B cell depletion in patients with refractory autoimmune diseases (ADs). Its effects on immune phenotypes remain largely unknown.

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**Objectives**
To characterize changes in immune cell subsets and transcriptomic profiles in 3 patients with juvenile dermatomyositis (JDM) and 2 patients with childhood-onset systemic lupus erythematosus (cSLE) treated with anti-CD19 CAR T cell therapy.
**Methods**
Patients received an infusion of autologous anti-CD19 CAR T cells. Immune cell phenotyping in peripheral blood (PB) and bone marrow (BM) samples was performed according to SOPs before and up to one year after CAR T cell infusion. RNA sequencing was performed on whole blood RNA.
**Results**
All patients achieved and maintained clinical remission off medication following CAR T cell therapy. In BM, B cell reconstitution started from B progenitors followed by immature, transitional and naïve B cells. In PB, the first B cells to emerge had a transitional phenotype, followed by naïve B cells. Unswitched memory B cells started to reconstitute 6 months after treatment, whereas switched memory B cells were absent up to 12 months after treatment. The fraction of IL-10 producing B cells assessed before therapy was reduced compared to controls and progressively increased following treatment. The ratio of CD4:CD8 T cells decreased after treatment and remained low throughout follow-up. Expansion of terminally differentiated CCR7-CD28-CD57+ CD8 T cells and reduction in activated CD69+ CD8 T cells was observed. In all patients, transcriptomic analysis revealed a consistent downregulation of interferon signatures, together with an upregulation of metabolic pathways following treatment. Specific enrichment of ribosomal pathways in cSLE patients and pathways related to growth factors, nuclear organization, and immune regulation in JDM patients were observed.
**Conclusion**
CD19-CAR T cell therapy emerges as a powerful disease-modifying approach, capable of achieving deep, durable, and drug-free remission in children with refractory ADs. CAR T cell therapy allowed the reconstitution of a naïve B cell repertoire and restored the regulatory functions of B cells. Terminally differentiated CD8 T cells expanded and activated CD8 T cells decreased after treatment. Transcriptomic analysis revealed a modulation of the interferon module and of metabolic pathways that could be associated with remission.
**Disclosure of interest**
E. Marasco Consultant with: Novartis, BMS, A. Ariolli: None declared, N. De Franco: None declared, R. Nicolai: None declared, C. Bracaglia: None declared, M. Becilli: None declared, P. Merli: None declared, M. Petrone: None declared, F. Del Bufalo: None declared, M. Algeri: None declared, M. Cefalo: None declared, C. Quintarelli: None declared, F. Locatelli: None declared, F. De Benedetti Consultant with: Novartis, SOBI.

## O03

### CLUSTERA: a multicentric data-driven phenotypic cluster analysis in enthesitis-related arthritis

Ö. Özenli Yağcı[1,*], B. Küçükali[1], S. Atamyıldız Uçar[2], E. Tunce[2], C. Dinler Yaraşır[3], P. Prencuva Akyürek[4], D. Gezgin Yıldırım[1], N. Aktay Ayaz[4], S. Elmas[3], B. Sözeri[2], S. A. Bakkaloğlu[1]
[1]Department of Pediatric Rheumatology, Gazi University Faculty of Medicine, Ankara; [2]Department of Pediatric Rheumatology, Ümraniye Training and Research Hospital, İstanbul; [3]Department of Pediatric Rheumatology, Ankara Etlik City Hospital, Ankara; [4]Department of Pediatric Rheumatology, İstanbul University İstanbul Faculty of Medicine, İstanbul, Türkiye
**Correspondence:** Ö. Özenli Yağcı
*Pediatric Rheumatology 2026*, **24(S1):**O03

**Introduction**
Enthesitis-related arthritis (ERA) is a heterogeneous subtype of JIA with considerable variability in clinical presentation, laboratory findings, and disease course, suggesting the presence of distinct underlying disease phenotypes (1). However, phenotypic stratification in ERA remains poorly defined, and data-driven approaches are limited.
**Objectives**
The study aims to identify distinct disease phenotypes in pediatric patients with ERA using data-driven cluster analysis based on demographic, clinical, laboratory, and MRI findings, alongside disease activity and damage scores. Furthermore, we sought to investigate the association between these phenotypic clusters and their response patterns to conventional and biologic therapies.
**Methods**
This multicenter retrospective study included pediatric patients diagnosed with ERA according to the ILAR JIA classification, from four tertiary pediatric rheumatology centers in Türkiye. Data were collected from medical records. Disease activity was assessed using the Wallace criteria for remission. Two-step cluster analysis was performed and the clustering solution quality was fair, with a silhouette coefficient in the range of 0.3–0.5.
**Results**
The study included 165 pediatric patients with ERA. Two-step cluster analysis identified two distinct phenotypic clusters: Cluster 1 ($n$=65) and Cluster 2 ($n$=100). Cluster 1 demonstrated a higher inflammatory burden, with elevated CRP (6.30 [1.26–23.43] vs. 1.20 [0.30–3.70], $p$<0.001) and ESR (28.00 [14.25–47.25] vs. 12.00 [5.00–21.00], $p$<0.001), and younger age at symptom onset (11.50 [9.00–13.00] vs. 13.00 [11.00–15.00], $p$=0.01), compared to Cluster 2. Cluster 1 was characterized by higher frequencies of peripheral arthritis (100% vs. 63%, $p$=0.001) and HLA-B27 positivity (100% vs. 18%, $p$<0.001). In contrast, axial involvement was more frequent in Cluster 2 (69% vs. 45%, $p$=0.003), while structural damage was observed exclusively in Cluster 2 (27% vs. 0%, $p$<0.001). Despite similar rates of csDMARD response and anti-TNF exposure, active disease at 6 months was more frequent in Cluster 2 (43% vs. 32%, $p$=0.032).
**Conclusion**
Data-driven cluster analysis identified two clinically distinct ERA phenotypes: an inflammatory peripheral phenotype and an axial-predominant phenotype with structural damage similar to adult studies. Despite comparable anti-TNF treatment exposure, clusters demonstrated meaningfully different outcomes, with Cluster 2 exhibiting greater structural damage and more persistent disease activity. These findings support the need for phenotype-based risk stratification in pediatric ERA, and suggest that patients with an axial-predominant phenotype may benefit from earlier treatment escalation and closer radiological surveillance to prevent structural progression.

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**Disclosure of interest**
None declared.

## O04

### Longitudinal relevance of synovial effusion in transitional juvenile idiopathic arthritis: implications for OMERACT pediatric and adult-inspired ultrasound definitions

A. Pilato[1], G. Tarantino[2], M. I. Petrone[2], A. Aquilani[2], E. Marasco[2], R. Nicolai[2], L. Navarini[1], R. Giacomelli[1], F. De Benedetti[2], S. Magni Manzoni[2,*]
[1]Rheumatology and Clinical Immunology, University of Rome Campus Biomedico; [2]Rheumatology Unit, "Bambino Gesù" Children Hospital IRCSS, Rome, Italy
**Correspondence:** S. Magni Manzoni
*Pediatric Rheumatology 2026*, **24(S1):**O04

**Introduction**
Musculoskeletal ultrasound (US) is widely used to assess synovitis in juvenile idiopathic arthritis (JIA). However, pediatric and adult OMERACT US definitions differ in the role assigned to synovial effusion. The clinical relevance of this discrepancy remains unclear in transitional JIA.
**Objectives**
To compare pediatric and adult inspired OMERACT US synovitis definitions and assess whether isolated effusion at baseline predicts follow-up inflammatory US activity in non-systemic JIA in transition.
**Methods**
We performed a longitudinal US analysis in non-systemic transitioning JIA patients. US was performed at baseline and at a follow-up visit, on clinically selected joints. Effusion, hypertrophy and Power Doppler (PD) signal were recorded as binary variables. OMERACT Pediatric-inspired synovitis was defined as effusion and/or hypertrophy, while adult-inspired synovitis required hypertrophy. Baseline US patterns were classified into four mutually exclusive groups: PD-positive, hypertrophy (no PD), isolated effusion, and no US lesion. The primary outcome was PD positivity at follow-up. Multiple sensitivity analyses were performed to address event rarity and within-patient clustering.

**Results**
The overall cohort included 210 patients, with 3,239 ultrasound-assessed joints across T0 and T1. Longitudinal analyses were restricted to the 902 joints assessed at both time points. OMERACT Pediatric-inspired synovitis decreased from 128/902 joints (14.2%) at baseline to 69/902 (7.6%) at follow-up; adult-inspired synovitis decreased from 34/902 (3.8%) to 25/902 (2.8%). At baseline, 772 joints showed no US abnormalities, 60 isolated effusion, 16 hypertrophy without PD, and 54 PD positivity. PD at follow-up occurred in 9/60 joints with isolated effusion versus 6/772 joints without baseline US abnormalities (15.0% vs. 0.8%). Isolated effusion was associated with T1 PD positivity in Firth regression (OR 21.75, 95% CI 7.68–61.61, $p<0.001$) and modified Poisson (RR 19.30, 95% CI 5.38–69.24, $p<0.001$). At patient level, 5/27 (18.5%) patients with ≥1 isolated-effusion joint had PD at follow-up versus 1/50 (2.0%) with US-negative joints only (RR 9.26, 95% CI 1.14–75.27).
**Conclusion**
In transitioning JIA patients, OMERACT pediatric and adult-inspired US definitions of synovitis showed substantial differences in classification and longitudinal behavior, largely driven by the role assigned to synovial effusion. Isolated synovial effusion was associated with T1 PD activity across joint-level, rare-event, cluster-adjusted, and patient-level sensitivity analyses. Our results support caution when applying adult-inspired US definitions to transitional JIA. Pediatric-inspired definition, by retaining effusion as a defining component of synovitis criterion, may capture prognostically relevant abnormalities that could be overlooked by adult criteria.
**Disclosure of interest**
None declared.

## O05

### Obesity, diagnostic delay and disease severity in juvenile psoriatic arthritis: an international cohort study

E. Broden-Barbareau[1,*], A. Cox[2], H. Lythgoe[3], C. Ciurtin[4], M. Batthish[5], J. Guzman[6], K. Hyrich[2,7], F. McErlane[8], S. Shoop-Worrall[2] on behalf of CAPS PIs
[1]Children and Young Person's Rheumatology Research Programme, Centre for Musculoskeletal Research, The University of Manchester, Manchester; [2]Children and Young Person's Rheumatology Research Programme, Centre for Musculoskeletal Research, The University of Manchester; [3]Paediatric Rheumatology and Adolescent Rheumatology, Royal Manchester Children's Hospital, Manchester; [4]Department of Ageing, Rheumatology and Regenerative Medicine, Division of Medicine, University College London, London, UK; [5]Division of Rheumatology, Department of Paediatrics, McMaster University, Ontario; [6]University of British Columbia, Division of Rheumatology, BC Children's Hospital, Vancouver, Canada; [7]NIHR Manchester Biomedical Research Centre, Manchester Foundation NHS Trust, Manchester Academic Health Science Centre, Manchester; [8]Paediatric Rheumatology, Newcastle Hospitals, Newcastle, UK
**Correspondence:** E. Broden-Barbareau
*Pediatric Rheumatology 2026*, **24(S1):**O05

**Introduction**
Obesity is associated with increased inflammation and poorer outcomes in adult psoriatic arthritis, but its impact in JPsA remains unclear.
**Objectives**
To investigate whether obesity is associated with delayed diagnosis and more severe disease presentation in children and young people (CYP) with JPsA.
**Methods**
CYP were recruited from three prospective inception cohorts: CAPS (UK, 2001–2019), ReaCCh Out (Canada, 2005–2010), and the CAPRI JIA Registry (Canada, from 2019). Participants had a physician diagnosis of JPsA within one year of presentation to rheumatology. Baseline demographic, clinical, and patient reported outcomes were collected. BMI z-scores (SD from age- and sex-adjusted WHO reference BMI) were calculated at presentation. Baseline clinical features were compared between BMI groups using Kruskal-Wallis tests and chi-squared tests.
**Results**
A total of 190 CYP were included: 112 (59.0%) had a healthy BMI, 41 (21.6%) were overweight, and 37 (19.5%) were obese. CYP with obesity were older at symptom onset than those in the healthy-weight group (11.1 years, IQR 10.2-14.0 vs. 9.6 years, IQR 4.2-12.6; $p=0.02$). Time from symptom onset to diagnosis was also longer in the CYP with obesity than in the healthy-weight group (6.2 months, IQR 2.6-19.1 vs. 5.0 months, IQR 2.3-11; $p=0.31$). Erythrocyte sedimentation rate was higher in CYP with obesity compared with the healthy-weight group (25.0, IQR 15.0-45.0 vs. 13.0, IQR 6.0-28.0; $p=0.008$). They also showed a greater prevalence of psoriasis (69.7% vs. 43.58%; $p=0.005$). Although not significant, CYP with obesity demonstrated numerically higher active joint counts (4.0, IQR 1.0-11.5 vs. 2.0, IQR 1.0-6.0), increased limited joint counts (2.0, IQR 1.0-8.0 vs. 1.0, IQR 0.0-4.0), while physician global disease activity scores were similar between groups (2.5, IQR 1.2-4.5 vs. 2.4, IQR 1.0-4.0). Patient-reported outcomes were poorer in CYP with obesity compared with those in the healthy-weight group, with higher scores indicating worse wellbeing, pain, and functional impairment. (3.7, IQR 2.1-6.0 vs. 1.9, IQR 0.3-4.2; $p=0.005$) and greater pain scores (4.9/10 cm, IQR 2.0-6.8 vs. 2.7/10 cm, IQR 1.0-5.0; $p=0.046$). CHAQ functional ability scores were also higher in the CYP with obesity, although this difference did not reach statistical significance (0.69, IQR 0.3-1.5 vs. 0.38, IQR 0.13-0.88).
**Conclusion**
In this international cohort, CYP with obesity showed a more severe baseline presentation, with greater inflammatory burden and poorer patient-reported outcomes. These findings also suggest a trend towards delayed diagnosis in CYP with JPsA and obesity.
**Disclosure of interest**
None declared.

## O06

### FATP2 is the receptor for fatty acids on CD4 +T cells in JIA at the site of inflammation

S. Hebbar Subramanyam[1,*], K. Ohl[1,2], A. Rajendiran[1], R. de Jong[1], J. Turyne Hriczko[1], C. T. Neullens[1], M.-F. Mashreghi[3,4], J. van Loosdregt[5], B. Vastert[5], K. Tenbrock[1,6]
[1]Department of Pediatrics, Uniklinik RWTH Aachen, Aachen; [2]Faculty of Food and Nutrition Sciences, University of Applied Sciences, Hochschule Niederrhein, Moenchengladbach; [3]Leibniz-Institute, German Rheumatology Research Center (DRFZ); [4]German Center for Child and Adolescent Health (DZKJ), Berlin, Germany; [5]Pediatric Rheumatology and Immunology, UMC Utrecht, Utrecht, Netherlands; [6]Department of Pediatrics, Inselspital, University of Bern, Bern, Switzerland
**Correspondence:** S. Hebbar Subramanyam
*Pediatric Rheumatology 2026*, **24(S1):**O06

**Introduction**
Juvenile idiopathic arthritis (JIA) is a chronic inflammatory joint disorder affecting children, in which T cells function within a microenvironment characterized by low glucose and elevated fatty acid levels. The metabolic mechanisms that allow T cells to adapt and remain active under these nutrient-limited conditions are still not well understood.
**Objectives**
To identify metabolic adaptations that enable CD4⁺ T cells to function within the inflammatory, fatty acid–rich microenvironment of juvenile idiopathic arthritis (JIA), and to characterize the role of newly identified pathways in regulating T cell function and inflammatory responses.
**Methods**
We examined T cells from both the peripheral blood (PB) and Synovial fluid (SF) of JIA patients and conducted bulk gene expression analysis and single-cell RNA sequencing. These analyses revealed increased expression of Fatty Acid Transport Protein 2 (FATP2), which was confirmed by flow cytometry. We then investigated the effects of inhibiting FATP2 using the compound lipofermata in vitro through flow cytometry and RNA sequencing and in vivo using a collagen-induced arthritis mouse model.
**Results**
T cells from SF showed significantly higher fatty acid uptake and FATP2 expression compared to PB T cells, particularly in activated and memory T cell subsets. Inhibition of FATP2 with lipofermata reduced fatty acid uptake, T cell proliferation, and interferon-γ production through pAKT3/mTOR-dependent pathway. However, lipofermata treatment did not alleviate arthritis symptoms in mice, likely because FATP2 is not similarly regulated in mouse memory T cells. Analysis of open-source sequencing data indicates FATP2 is also upregulated in psoriatic and rheumatoid arthritis at sites of inflammation. A humanized psoriatic arthritis mouse model is planned.
**Conclusion**
These findings indicate that T cells in JIA adapt to a fatty acid–rich environment by increasing FATP2 expression. Disrupting this metabolic

adaptation through FATP2 inhibition may represent a potential therapeutic strategy for treating JIA and other autoimmune inflammatory diseases.

**Disclosure of interest**
None declared.

## O07

### An extrafollicular response characterizes patients with oligoarticular and polyarticular juvenile idiopathic arthritis at disese onset

A. Ariolli[1,*], N. De Franco[1], A. Aquilani[1], G. Tarantino[1], M. I. Petrone[1], R. Nicolai[1], I. Caiello[1], S. Magni Manzoni[1], F. De Benedetti[1], E. Marasco[1]
[1]Rheumatology Unit, Ospedale Pediatrico Bambino Gesù, Rome, Italy
**Correspondence:** A. Ariolli
*Pediatric Rheumatology 2026*, **24(S1):**O07

**Introduction**
An hyperactivation of B cells characterizes patients with oligoarticular- and polyarticular-JIA (1) and switched memory B cells are expanded in these group of patients (2). Our preliminary data showed changes in the immunoglobulin repertoire of JIA patients with reduced somatic hypermutation, suggesting extrafollicular activation of autoreactive B cells.
**Objectives**
We aimed to investigate the phenotype of B cells in patients with JIA.
**Methods**
A total of 64 JIA patients and 25 pediatric controls were enrolled, including 15 patients in the inception cohort and 49 patients in the validation cohort. Paired mononuclear cell samples isolated from peripheral blood (PBMC) and synovial fluid (SF) of JIA patients ($n$ = 15) were retrospectively retrieved from our biobank. B- and T-cell immunophenotypes were characterized by multiparametric flow cytometry. Patients were followed up for a minimum of three years after diagnosis. BAFF and CXCL13 levels were evaluated by ELISA. To assess TLR9 activation, PBMCs were labeled with CFSE and stimulated with CpG as previously described (3).
**Results**
We enrolled 15 patients with a diagnosis of JIA according to the ILAR criteria. 67% patients had a diagnosis of oligoarticular-JIA, 33% RF negative poly-JIA; 74% were female. 34% patients were on methotrexate, 60% patients were on methotrexate + TNF inhibitor, one patient was on TNF inhibitor alone. The IgG+ to IgA+ memory B-cell ratio was increased in JIA patients versus controls, reflecting an expansion of IgG+ memory B cells, which represented the predominant B-cell subset in SF. IgG+ memory B cells were enriched for a CD21−CXCR5−CD11c+ phenotype, particularly in SF, suggesting an extrafollicular origin. Tbet+ B cells and activated ICOS+ PD1+ peripheral helper T cells (Tph) were expanded in SF compared to PB of patients and controls. Our validation cohort consisted of 49 children with JIA: 85% had oligo-JIA and 15% had RF- poly-JIA; 68% were female. The expansion of CD21-CXCR5-CD11c+IgG+ B cells in patients compared to controls was confirmed in the validation cohort. Patients at diagnosis showed higher levels of BAFF and CXCL13 and an expansion of Tph cells. We then separated patients into two groups based on the follow up data: patients who were treated only with joint injections (no DMARDs) and patients who had to start methotrexate or a TNF inhibitor (DMARD) for active disease after joint injections. Patients in the DMARD group showed higher levels of CXCL13, whereas levels of CD21-CXCR5-CD11c+IgG+ B cells, Tph cells and BAFF were comparable between the two groups. We stimulated cells of JIA patients ($n$=9) and controls ($n$=14) with CpG: after 7 days the fraction of plasmablasts was higher in patients than controls.
**Conclusion**
Our findings indicate that JIA is characterized by enhanced extrafollicular B cell activation, accompanied by an expansion of Tph cells. BAFF and TLR9 signaling likely contribute to this extrafollicular activation program. In parallel, we detected increased CXCL13 levels, consistent with augmented follicular B cell responses. CXCL13 elevation was most evident in patients who went on to require systemic therapy. Together, these data support a model in which extrafollicular B cell activation is a core feature of JIA, whereas heightened follicular responses (occurring in lymph nodes or synovial tertiary lymphoid structures) may mark a subset of patients with more severe disease who require systemic treatment.

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**Disclosure of interest**
None declared.

## O08

### A distinct t cell–mediated immunopathology underlies antibiotic-refractory lyme arthritis

J. M. Fischer[1,*], J. Dirks[1], F. Trost[1], J. Stumpf[2], C. Vollmuth[1], A. Holl-Wieden[1], J. Brunner[3], T. Hohmann[3], R. Borchers[5], H. Girschick[6], C. Kleemann[4], A. Schubert-Unkmeir[2], H. Morbach[1]
[1]Pediatric Inflammation Medicine, University Children's Hospital Würzburg; [2]Institute for Hygiene and Microbiology, University of Würzburg, Würzburg, Germany; [3]Department of Pediatrics, Medical University of Innsbruck, Innsbruck, Austria; [4]Pediatric Immunology, Rheumatology, and Infectiology, Leipzig University Hospital for Children and Adolescents, Leipzig; [5]Children's Hospital, University of Augsburg, Augsburg; [6]Pediatrics, Vivantes Klinikum im Friedrichshain, Berlin, Germany
**Correspondence:** J. M. Fischer
*Pediatric Rheumatology 2026*, **24(S1):**O08

**Introduction**
While most patients with Lyme arthritis (LA) respond to antibiotics, a subset develop antibiotic-refractory Lyme arthritis (ARLA), defined by persistent synovitis despite bacterial clearance. The mechanisms underlying ARLA and its distinction from other inflammatory arthritides remain unclear. Although T and B cell–mediated immune responses have been implicated, their molecular basis and population-level relevance are poorly understood.
**Objectives**
To dissect the T cell–mediated immunopathology of antibiotic-refractory Lyme arthritis in direct contrast to oligoarticular/polyarticular juvenile idiopathic arthritis (o/p-JIA).
**Methods**
We characterised a large German paediatric LA cohort ($n$ = 179) to compare clinical and immunological profiles of ARLA ($n$ = 29) with antibiotic-responsive LA ($n$ = 150) and o/p-JIA ($n$ = 52). Synovial fluid from ARLA ($n$ = 16) and JIA ($n$ = 35) was analysed using flow cytometry, single-cell RNA sequencing (scRNA-seq), and high-throughput T cell receptor (TCR) sequencing. ARLA was defined as positive *Borrelia* serology and persistent arthritis after two antibiotic treatment courses.
**Results**
ARLA patients were older at onset compared to responsive LA and typically presented with monoarticular disease. Antibiotic type and NSAID use did not affect LA outcomes. Compared to o/p-JIA, ARLA synovial fluid showed enrichment of PD-1$^+$ HLA-DR$^+$ CD4$^+$ effector T cells and reduced FOXP3$^+$ CD25$^+$ regulatory T cells. Effector T cells in ARLA produced higher amounts of IL-1β, CCL2, and TNF-α than their counterparts in o/p-JIA, but comparable amounts of IL-17A and IFN-γ. In contrast to o/p-JIA, the regulatory T cell compartment showed weaker signs of antigen-specific activation and clonal expansion compared to Tph cells.
**Conclusion**
ARLA affects older children and is characterised by a distinct, HLA-linked, T cell–driven immune response. scRNA-seq findings suggest an impaired regulatory T cell response as a potential contributor to disease pathogenesis, supporting a T cell–mediated immunopathology specific to ARLA and distinct from o/p-JIA. Ongoing work aims to identify ARLA-specific antigens and elucidate molecular mechanisms using synovial organoid models. A planned multicentre study will seek to identify predictive biomarkers to guide early immunomodulatory therapy.

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**Disclosure of interest**
None declared.

## O09

### Distinct trajectories of disease severity progression in juvenile localized scleroderma

G. Mastrangelo[1], A. Meneghel[1,*], F. Vittadello[2], F. Tirelli[1], F. Zulian[1]
[1]Division of Pediatric Rheumatology, Department of Women and Children Health, University of Padua; [2]Department of Research and Statistical Analysis, Explora, Padua, Italy
**Correspondence:** A. Meneghel
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**Introduction**
Juvenile localized scleroderma (JLS) is a rare fibrosing disorder with heterogeneous disease courses, yet longitudinal patterns of disease activity in affected children remain poorly characterised.
**Objectives**
To describe a large longitudinal cohort of children with JLS, and to identify latent subgroups of disease activity trajectories using growth mixture modelling.
**Methods**
Retrospective cohort study using prospectively collected data from 133 patients with JLS between January 2010 and December 2025. Children were included in the study if diagnosed according to the 2006 Padua Classification criteria, treated at the University Hospital of Padua, and with complete data over a minimum 4-year follow-up. According to disease classification, five subtypes were included: circumscribed morphea, linear scleroderma of the trunk and limbs, linear scleroderma of the head, generalized morphea, pansclerotic and mixed morphea grouped together. Demographic, clinical, and laboratory data were recorded alongside serial disease severity assessments. The primary outcome was change in the Localized Scleroderma Assessment Tool (LoSCAT) with a ≥25% shift from baseline defined as clinically significant. Growth Mixture Modelling was applied to uncover distinct longitudinal severity classes.
**Results**
Eighty-five patients with JLS (64% females), with ≥48 months of follow-up, were included. Twenty-five (29%) had linear scleroderma of the trunk/ limbs, 21% linear scleroderma of the head, 20% generalized morphea, 19% pansclerotic/mixed morphea, and 11% circumscribed morphea. ANA positivity was present in 24% of patients and RF in 1.5%, with most ANA-positive cases in the trunk/limb linear subtype (45%). Forty-four children received topical therapy, predominantly in the circumscribed morphea group, while 81% were treated with systemic therapy, most commonly methotrexate (55%), followed by mycophenolate mofetil (12%); 57% remained on methotrexate at 12-month follow-up. We identified two latent classes of disease severity. Class 1 comprised patients with low disease activity, favourable treatment response, and minimal long-term damage; it included all patients with linear scleroderma of the head, 89% with circumscribed morphea, nine with trunk/limb linear disease, three with generalized morphea, and two with pansclerotic/mixed subtype. Class 2 was characterized by high disease activity and substantial long-term damage, and included 16 patients with trunk/limb linear scleroderma, 14 with generalized morphea, and 14 with pansclerotic/mixed subtype.
**Conclusion**
We identified two distinct latent classes of disease severity in JLS with clear clinical relevance. Class 1 (low activity, minimal damage) included linear scleroderma of the head and circumscribed morphea, whereas Class 2 (high activity, severe damage) comprised trunk/limb linear, generalized, and pansclerotic/mixed forms. JLS subtype strongly predicted trajectory class membership.
**Disclosure of interest**
None declared.

## O10

### A novel variant in USP18 causing late onset recurrent HLH and severe CNS-HLH with acute necrotising encephalopathy

I. Z. Turtsevich[1,*], P. Cowie[2], M. Sa[3], D. Kelly[4], M. J. Carter[2], A. Kavirayani[1]
[1]Paediatric Rheumatology; [2]PICU; [3]Paediatric Neurology; [4]Infectious Diseases, Oxford University Hospitals NHS Foundation Trust, Oxford, UK
**Correspondence:** I. Z. Turtsevich
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**Introduction**
Ubiquitin-Specific Peptidase 18 (USP18) deficiency is an ultra-rare autosomal recessive type I interferonopathy caused by mutations in *USP18* gene, a key negative regulator of interferon signalling (1). Reported cases in literature typically present in the neonatal period with severe neuroinflammatory disease resembling pseudo-TORCH syndrome (2). Late-onset presentations have not been previously described.
**Objectives**
To describe a patient with a novel hypomorphic *USP18* variant presenting as recurrent haemophagocytic lymphohistiocytosis (HLH) with severe CNS-HLH in late childhood, expanding the known clinical spectrum of this condition.
**Methods**
An 11-year-old girl of consanguineous parents presented with recurrent self-limiting episodes of presumed HLH, characterised by fever, cytopenia, and marked hyperferritinaemia. Extensive infectious, autoimmune and immunological assays, including perforin expression and granule release assay were negative, with genetic testing remained pending. Whilst under outpatient follow-up, she presented with acute neurological deterioration and seizures, with MRI evidence of widespread haemorrhagic and inflammatory brain lesions, consistent with acute necrotising encephalopathy. She required intensive care with mechanical ventilation and inotropes with supportive therapy including antimicrobials and antiepileptics. Immunomodulatery treatment included intravenous (IV) corticosteroids, IV immunoglobulin, etoposide, and high-dose anakinra, with subsequent addition of a JAK inhibitor, Baricitinib, following genetic results. Whole genome sequencing identified a homozygous *USP18* splice-site variant (c.628–3C> G), classified as uncertain significance due to its extreme rarity and limited published data. RNA studies confirmed abnormal splicing with residual wild-type transcript, suggesting a hypomorphic effect. Interferon signature testing demonstrated mild type I interferon upregulation, likely attenuated by concurrent immunosuppressive therapy. She made a remarkable recovery following targeted immunosuppression and intensive multiprofessional care, with only mild residual neurological deficits on follow-up.
**Conclusion**
this case broadens the phenotypic spectrum of *USP18* deficiency, demonstrating that hypomorphic variants may present beyond infancy with recurrent HLH and life-threatening neuroinflammation. It highlights the importance of early genetic testing in unexplained or atypical HLH, to facilitate targeted JAKi therapy and improve outcomes.

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**Disclosure of interest**
None declared.

## O11

### FIRES/NORSE: a cerebral autoinflammatory syndrome

S. Barreiro Pérez[1], R. Alcobendas[1], C. Udaondo[1,*], A. Remesal[1], A. Galindo[2], M. Río-García[2], R. Velázquez[3], M. D. M. García Romero[3], A. Hercberg[1], L. Martínez[1]
[1]Pediatric Rheumatology; [2]Pediatric Intensive Care; [3]Pediatric Neurology, Hospital Universitario La Paz, Madrid, Spain
**Correspondence:** C. Udaondo
*Pediatric Rheumatology 2026*, **24(S1):**O11

**Introduction**
New-onset refractory status epilepticus (NORSE) and febrile infection-related epilepsy syndrome (FIRES) are rare clinical entities characterized by a biphasic course, with an acute phase followed by chronic refractory epilepsy and neurological impairment. Neuroimaging and autoimmune investigations are typically negative. The aetiology remains unknown; however, elevated levels of cytokines such as IL-1β,

IL-6, TNFα, IFNγ, CXCL10, and IL-8 have been identified in both serum and cerebrospinal fluid (CSF), supporting an inflammatory pathophysiology.

**Objectives**

To describe a paediatric patient with suspected FIRES/NORSE who showed a favourable response to intensive immunomodulatory treatment.

**Methods**

Case report: A 6-year-old girl was transferred to the Paediatric Intensive Care Unit due to a four-day history of generalised seizures associated with aphasia, agitation, and behavioural changes. At the referring hospital, she was treated with levetiracetam, lacosamide, and aciclovir following an initial positive HSV-1 PCR result in cerebrospinal fluid (CSF). On admission, she deteriorated neurologically, developing disorganised movements and psychomotor agitation. Continuous electroencephalographic monitoring revealed refractory non-convulsive status epilepticus, requiring escalation of antiepileptic therapywith brivaracetam, lacosamide, clobazam, valproate, and continuous infusions of midazolam and propofol. Given the initial suspicion of autoimmune encephalitis, intravenous methylprednisolone pulses, plasma exchange, intravenous immunoglobulins, and rituximab were initiated. However, brain magnetic resonance imaging was normal, and repeat CSF analysis at our centre was negative for HSV PCR and for autoimmune encephalitis-associated antibodies. On day 10 of admission, the patient developed a marked elevation of acute phase reactants with increased levels of IL-1, IL-6, and IL-8 in both serum and CSF, prompting initiation of anakinra at 100 mg every 6 h. Despite this, she subsequently developed episodes of intracranial hypertension associated with arterial hypertension, severe bradycardia, and desaturation, requiring cardiopulmonary resuscitation on one occasion. Suspecting sympathetic hyperactivity secondary to brain injury, propranolol was initiated and infliximab was added. On day 22 imaging revealed diffuse corticosubcortical atrophy, a finding previously described in this syndrome. However, the patient showed progressive clinical improvement, allowing extubation and transfer to the ward after 34 days of admission. At discharge, neurological examination was completely normal, permitting gradual tapering of corticosteroids and anakinra.

**Conclusions** The presence of elevated acute phase reactants in a patient with status epilepticus and otherwise unremarkable investigations should raise suspicion for this potentially life- threatening entity. Early initiation of correct immunomodulatory treatment is essential.

**Disclosure of interest**

None declared.

## O12

### Baricitinib in the treatment of new-onset juvenile dermatomyositis: a phase II trial"MYOCIT"

B. Bader Meunier[1,*], I. Melki[2], A. Welfringer-Morin[3], H. Reumaux[4], A. Carbasse[5], A.-L. Jurquet[6], I. Kone-Paut[7], P. Pillet[8], P. Quartier[1], M.-L. Frémond[1], U. Meinzer[9], A. Belot[10], B. Fournier[1], C. Gitiaux[11], F. Padavia[12], N. Bouazza[12]

[1]Pediatric Rheumatology, Necker Hospital; [2]Pediatrics, Trousseau Hospital; [3]Pediatric Dermatology, Necker Hospital, Paris; [4]Pediatrics, Jeanne de Flandre Hospital, Lille; [5]Pediatrics, Lapeyronie Hospital, Paris; [6]Pediatrics, North Hospital, Marseille; [7]Pediatric Rheumatology, Bicêtre Hospital, Le Kremlin Bicêtre; [8]Pediatric Rheumatology, Bordeaux Hospital, Bordeaux; [9]Pediatric Rheumatology, Robert Debré Hospital, Paris; [10]Pediatric Rheumatology, HFME Hospital, Lyon; [11]Pediatric Neurology; [12]Clinical Research Unit, Necker Hospital, Paris, France

**Correspondence:** B. Bader Meunier

*Pediatric Rheumatology 2026,* **24(S1):**O12

**Introduction**

The recommended first-line treatment with methotrexate (MTX) and corticosteroids is not efficient in a subset of patients with new-onset juvenile dermatomyositis (JDM). We aimed to assess the efficacy and safety of baricitinib in children with JDM.

**Objectives**

Our primary outcomes were the proportion of patients achieving a JDM PRINTO 20 level of improvement at month 6. Main secondary clinical outcomes were the proportion of patients achieving a juvenile JDM PRINTO 50, 70, and 90 levels of improvement, the assessment of the Total Improvement Score (TIS), safety, and the reduction in median corticosteroids dosage at month 6.

**Methods**

We did a single arm 6-months phase II study at 13 French pediatric rheumatology centres. The study used a Simon-two stage design based on the historical PRINTO 20 response rates in patients with new-onset JDM with a combination of corticosteroids and MTX, excluding severe JDM[1]. As our study was designed to include severe cases of JDM, the unacceptable response rate was conservatively set at 50%. Sixteen evaluable patients were required and our study would be considered positive if 11 or more responses achieved a PRINTO 20 level of improvement. We enrolled patients aged 3-16 years with new-onset JDM, with muscle (at least MMT < 74 and/or CMAS < 45) and cutaneous involvement. Baricitinib was administered orally once daily o at a dosage of 2 or 4 mg/day in children < 6 years or in children > 6 years respectively. Initial prednisone dosage was 1 or 2 mg/Kg/day according to the physician judgment.

**Results**

16 patients were enrolled. Patients had muscle (16/16), cutaneous (16/16), arthritis (6/16), lung (2/16) involvement. 5/16 (31%) of the patients had severe JDM. 13/16 (81%) of the patients had myositis specific autoantibodies: anti-NXP2 (5), anti-MDA5 (4), anti-Mi2 (2), anti-TIF-1 (1), anti-SAE (1). At month 6, JDM PRINTO 20 level of improvement were achieved in 12/16 (75%) of the patients, exceeding the predefined efficacy threshold. PRINTO 50, 70 and 90 scores were achieved in 13/15 (87%), 11/14 (79%) and 5/14 (36%) of the patients ; major clinical improvement on TIS was observed in 12/14 (86%) of the patients. Median dosage of prednisolone significantly decreased from 1.05 [0.88 ; 2.08] to 0.18 [0 ; 0.56] mg/Kg/d ($p$=0,003). Three severe adverse events (ankle and dorsal foot oedema, transient elevated transaminase levels> 8 N, phlegmon of the flexor tendon sheath of one finger) and 66 adverse events occurred. None of them resulted in a discontinuation of Baracitinib. No patient died.

**Conclusion**

Baricitinib met the predefined efficacy criteria of the Simon two-stage design, with encouraging short-term results compared with historical response rates from MTX-based cohorts, and the short-term safety of Baricitinb good. These results should be confirmed in a larger international randomised study and long-term safety should be assessed.

**References**

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**Trial registration identifying number**

NCT05524311.

**Disclosure of interest**

None declared.

## O13

### Interstitial lung disease in juvenile dermatomyositis: a single centre experience from North India

R. Singh[1,*], V. Pandiarajan[1], Y. Sharma[1], R. Semwal[1], R. K. Pilania[1], M. Dhaliwal[1], S. Sharma[1], D. Suri[1], A. Rawat[1], K. S. Sodhi[2], A. Gupta[1], S. Singh[1]

[1]Pediatric Allergy and Immunology; [2]Department of Radiodiagnosis and Imaging, PGIMER, Chandigarh, India

**Correspondence:** R. Singh

*Pediatric Rheumatology 2026,* **24(S1):**O13

**Introduction**

Interstitial lung disease (ILD) is a potentially life- threatening manifestation of juvenile dermatomyositis (JDM). Data on ILD in children with JDM from the Indian subcontinent are scarce. We sought to assess the clinical and immunological profile and outcome of ILD in JDM .

**Objectives**

To identify different ILD patterns in patients with JDMS and their association with myositis specific antiboides.To identify the mortality rate and patterns in various forms of ILD.

**Methods**

We performed a retrospective analysis of 209 children with JDM who were diagnosed and followed up at our centre, among whom 24 patients had ILD which was confirmed on high-resolution computed tomography (HRCT) of the chest. Their clinical features, laboratory investigations, treatment regimens, and outcomes were were analysed in detail.

**Results**
Of 209 patients with JDM, 24 (11.5%) were diagnosed with ILD. Median age at disease onset was 9.0 years (range: 2.5-15.0), median age at ILD diagnosis was 10.17 years (range 3.0-16.5), and median diagnostic delay was 4 months (range 0-60). The predominant clinical manifestations included proximal muscle weakness (18/24; 75%), Gottrón's papules (17/24; 71%), and fever (13/24; 54%). Nailfold capillaroscopy (NFC) abnormalities were detected in 16/24 (67%). Myositis-specific antibodies (MSA) testing was performed in 15/24 (63%), and 14 (93%) were MSA-positive. Anti-MDA5 was the most frequent antibody (10/15; 66.6%), followed by anti-Ro52 (9/15; 60%). On HRCT, the predominant pattern was non-specific interstitial pneumonia (NSIP) in 17/24 (71%), followed by usual interstitial pneumonia (UIP) in 4/24 (17%) and rapidly progressive ILD (RP-ILD) in 3/24 (12%). All patients were administered corticosteroids. Additional immunosuppression included methotrexate (18/24; 75%), cyclophosphamide (8/24; 33%), mycophenolate mofetil (6/24; 25%), tofacitinib (4/24; 17%), nifedipine (4/24; 17%), rituximab (2/24; 8%), azathioprine (2/24; 8%), pulse methylprednisolone (2/24; 8%), and intravenous immunoglobulin (2/24; 8%). Overall mortality was 5/24 (21%), 3 patients with RP-ILD died (100% mortality); all were anti-MDA5 positive with anti-Ro52 co-positivity.
**Conclusion**
This study being the largest single-centre cohort of JDM patients with ILD highlights early evaluation for ILD in all cases of JDM regardless of respiratory symptoms. ILD in JDM is associated with substantial mortality in our cohort. NSIP was the most common radiologic subtype, while anti-MDA5 antibodies were frequently identified with severe disease. Early and comprehensive MSA profiling is essential in JDM to risk-stratify patients and institute prompt aggressive immunosuppression.
**Disclosure of interest**
None declared.

## O14

### Targeting the type I interferon pathway: efficacy and safety of anifrolumab in refractory juvenile dermatomyositis—a multi-center experience

H. M. Natour[1,*], E. Iglesias[2], S. Carriqui[2], E. Baselga[3], A. Vicente[3], A. Alkandari[1], G. Filocamo[1], N. Nazer[1], K. Chiu[1], B. M Feldman[1], E. Jones[1], A. M Knight[1], D. M Levy[1], K. Whitney[1], R. S. M. Yeung[1], J. Antón[2]
[1]Division of Rheumatology, The Hospital for Sick Children, University of Toronto, Toronto, Canada; [2]Pediatric Rheumatology Department; [3]Pediatric Dermatology Department, Hospital Sant Joan de Déu, Barcelona, Spain
**Correspondence:** H. M. Natour
*Pediatric Rheumatology 2026*, **24(S1):**O14

**Introduction**
Juvenile dermatomyositis (JDM) is a rare autoimmune disease characterized by muscle inflammation and distinctive skin manifestations. Despite advances in treatment, a subset of patients remains refractory to conventional therapies, experiencing persistent disease activity and significant morbidity. Anifrolumab, a monoclonal antibody targeting the type I interferon alpha receptor subunit 1 (IFNAR1), has shown efficacy in adult systemic lupus erythematosus, but evidence in pediatric inflammatory myositis is limited.
**Objectives**
We aimed to detail the efficacy and safety of anifrolumab in pediatric patients with refractory inflammatory myositis from two tertiary centers.
**Methods**
We conducted a retrospective and prospective review of patients with refractory JDM treated with anifrolumab at two tertiary pediatric centers: Hospital Sant Joan de Déu, Barcelona, Spain, and The Hospital for Sick Children, Toronto, Canada. Data collected included demographics, disease manifestations, disease severity, laboratory findings (including Type I interferon-regulated gene [IRG] Z-scores), treatment details, and clinical responses.
**Results**
Six patients, four females and two males, aged 3.5 to 16 years at the time of diagnosis, were identified. The median time from diagnosis to starting anifrolumab was 12.5 months (IQR 7.3 to 23). All presented with muscle weakness and cutaneous features, patient 1 also had ulcerative skin disease and bowel perforation, while patient 2 had bulbar dysfunction. Autoantibody profiles included anti-NXP2 ($n$=3), anti-TIF1 ($n$=2) and anti MDA5 ($n$=1). Prior to commencing on anifrolumab infusions, all patients received prednisone, methylprednisolone pulses, and intravenous immunoglobulins (IVIG), along with two or more of methotrexate, tacrolimus, mycophenolate mofetil (MMF), tofacitinib, and cyclophosphamide. Patient 1 also needed three surgeries due to bowel perforation. Clinical responses were evaluated after ten anifrolumab infusions (5 mg/kg q4 weeks) for patient 1, two infusions for patient 2 and patient 6, eight infusions for patient 3 and 5, and six infusions for patient 4. All four patients demonstrated significant improvements in both skin involvement and overall disease activity. Patient 4 experienced a halt in the progression of the calcinosis lesion, while Patient 1 showed notable improvement in muscle strength and did not experience any recurrence of gastrointestinal issues. Additionally, the bulbar dysfunction in patient 2 resolved completely. The prednisone dosage was significantly reduced in patients 2 and 4, and it was completely discontinued for patients 1 and 3. Anifrolumab led to rapid, dramatic reduction in IFN signatures in all quantitative cases (Patients 3, 5, 6). No adverse effects related to anifrolumab were observed.
**Conclusion**
Anifrolumab demonstrated promising efficacy in this cohort of 6 patients with refractory JDM, resulting in clinically meaningful improvement in cutaneous and overall disease activity. The favorable safety profile, with no adverse effects reported in any patient, is encouraging. Type I interferon signature normalization in patients with available paired data provides biological evidence of successful IFNAR1 blockade and supports the mechanistic rationale for anifrolumab use in JDM.
**Disclosure of interest**
None declared.

## O15

### Developing 3D synovial membrane models derived from synovial fluid of patients with juvenile idiopathic arthritis

C. Kleimeyer[1,2,*], L. Finger[2], S.-J. Bremer[1,2], J. Pagel[1,2,3,4]
[1]Department of Pediatrics; [2]University Childrens' Research, University Medical Center Hamburg-Eppendorf; [3]Partner Site Hamburg-Lübeck-Borstel-Riems, German Center for Infection Research (DZIF); [4]Partner Site Hamburg, German Center for Child and Adolescent Health (DZKJ), Hamburg, Germany
**Correspondence:** C. Kleimeyer
*Pediatric Rheumatology 2026*, **24(S1):**O15

**Introduction**
The synovial membrane is the primary site of inflammation in juvenile idiopathic arthritis (JIA). Synovial fibroblasts (SF) contribute to disease pathogenesis through immune cell recruitment, angiogenesis, and extracellular matrix remodeling, making them an attractive therapeutic target [1, 2]. Compared with conventional two-dimensional cultures, three-dimensional (3D) cell culture models more closely recapitulate physiological tissue architecture and cellular interactions. To date, no 3D synovial membrane model has been described for JIA, representing a major limitation for studying disease mechanisms and evaluating therapeutic responses.
**Objectives**
To establish patient-derived 3D synovial membrane models with varying levels of complexity from synovial fluid of JIA patients, enabling applications ranging from high-throughput drug screening to mechanistic studies of disease pathogenesis.
**Methods**
SF were isolated from synovial fluid of eight JIA patients through serial passaging. Cell identity and purity were confirmed by qPCR and flow cytometry. Different 3D culture systems were established using ultra-low attachment plates, and extracellular matrix scaffolds. SF spheroids and organoids were stimulated with TNF-α (10 ng/ml), and IL-6 secretion was quantified by ELISA. In addition, spheroids were used in a proof-of-concept patient-specific drug screening assay. Organoids underwent histologic analysis.
**Results**
SF were successfully isolated and expanded from synovial fluid samples of JIA patients. Cells demonstrated a characteristic SF phenotype by qPCR (COL1A1+, PRG4+, PTPRC−) and flow cytometry (CD45−, CD31−,

CD90+, PDPN+). SF formed stable spheroids in ultra-low attachment culture conditions and exhibited increased IL-6 secretion following TNF-α stimulation. In proof-of-concept drug screening experiments, SF spheroids from four patients were stimulated with TNF-α and treated with ibuprofen, prednisolone, or baricitinib at varying concentrations, resulting in reduced IL-6 production. Furthermore, extracellular matrix-based cultures demonstrated structural features resembling the native synovial membrane, including lining and sublining-like layers.

**Conclusion**

We established the first patient-derived 3D synovial membrane models for JIA generated from synovial fluid rather than synovial biopsy tissue. These models provide versatile platforms with differing complexity and hold potential for personalized drug screening and investigation of JIA disease mechanisms, thereby offering a novel translational tool for pediatric rheumatology research.

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**Disclosure of interest**

None declared.

## O16

### PSTPIP1 and Pyrin, central regulators of macrophage differentiation

P. Berger[1,*], K. Tenbrock[1,2]
[1]Translational Rheumatology, RWTH Aachen, Aachen, Germany; [2]Pediatric Rheumatology, Inselspital Bern, Bern, Switzerland
**Correspondence:** P. Berger
*Pediatric Rheumatology 2026*, **24(S1):**O16

**Introduction**

Monocytes develop from hematopoietic stem cells and migrate into tissues, where they undergo stimulus–dependent and tissue–specific differentiation into macrophages, acquiring distinct inflammatory functions. The development of inflammatory properties during the differentiation of progenitor cells into macrophages remains incompletely understood.

**Objectives**

Our aim was to identify regulatory factors that govern monocyte/macrophage differentiation.

**Methods**

A genome–wide CRISPR/Cas9 knockout screen (GeCKO) was performed in ER–HoxB8 macrophages to identify key drivers of macrophage differentiation, which were subsequently validated in independent knockout and knock–in cell lines. Immunophenotyping was conducted by FACS; morphology and migration were assessed using fluorescence microscopy; and inflammatory responses were measured by ELISA. Transcriptomic data were generated by next–generation mRNA sequencing and validated by quantitative PCR and immunoblotting.

**Results**

The genome–wide CRISPR/Cas9 screen identified the cytosolic, cytoskeleton–associated adaptor protein PSTPIP1 (proline–serine–threonine phosphatase interacting protein 1) as a regulatory factor of macrophage differentiation. Notably, mutations in PSTPIP1 cause autoinflammatory disorders such as PAPA syndrome. Deletion of PSTPIP1 resulted in impaired differentiation, reduced inflammatory responses, altered morphology, and modified adhesion and migration properties. PSTPIP1 is a regulator of Pyrin inflammasome activity, which drives autoinflammation in familial Mediterranean fever (FMF). Deletion of Pyrin likewise led to pronounced alterations in macrophage cellular dynamics.

**Conclusion**

The genome–wide CRISPR/Cas9 screen identified the cytosolic, cytoskeleton–associated adaptor protein PSTPIP1 (proline–serine–threonine phosphatase interacting protein 1) as a regulatory factor of macrophage differentiation. Notably, mutations in PSTPIP1 cause autoinflammatory disorders such as PAPA syndrome. Deletion of PSTPIP1 resulted in impaired differentiation, reduced inflammatory responses, altered morphology, and modified adhesion and migration properties. PSTPIP1 is a regulator of Pyrin inflammasome activity, which drives autoinflammation in familial Mediterranean fever (FMF). Deletion of Pyrin likewise led to pronounced alterations in macrophage cellular dynamics.

**Disclosure of interest**

None declared.

## O17

### Interleukin 6 signaling restrains expression of interleukin 18 binding protein in Still's disease

L. Niemayer[1], M. Saers[1], S. Schleifenbaum[1], S. Fuehner[1], F. Dunst[2], C. Klemann[2], M.-A. Oestreich[3], K. Tenbrock[3,4], H. Wittkowski[1], T. Hinze[1,5], C. Hinze[1], D. Foell[1], E. L. Verweyen[1], C. Kessel[1,*]
[1]Pediatric Rheumatology & Immunology, University Hospital Muenster, Muenster; [2]Pediatric Immunology, Rheumatology and Infectiology, University Hospital Leipzig, Leipzig, Germany; [3]Pediatrics, Bern University Hospital, Inselspitall, Bern, Switzerland; [4]Pediatric Rheumatology, RWTH Aachen University Medical Faculty, Aachen; [5]Clinic of Paediatric and Adolescent Rheumatology, St.-Josef-Stift Sendenhorst, Sendenhorst, Germany
**Correspondence:** C. Kessel
*Pediatric Rheumatology 2026*, **24(S1):**O17

**Introduction**

Interleukin (IL) 18 binding protein (IL-18BP) is an endogenous soluble immune checkpoint of IL-18, a prominent inducer of T cellular IFNγ expression. IL-18BP is primarily produced by myeloid and epithelial cells and its expression is driven by IFNγ as result of a negative feedback loop in order to restrain excessive IL-18 signaling. In Still's Disease (SD) previous data suggest imbalanced expression of IL-18 compared to IL-18BP, resulting in unrestrained free IL-18 to contribute to systemic inflammation and cytokine storm conditions such as Macrophage Activation Syndrome (MAS).

**Objectives**

Here, we set out to better understand the immunological mechanisms behind a mutual IL-18/IL-18BP dysbalance in SD.

**Methods**

Whole blood (WB) and monocyte derived macrophage RNA sequencing (RNAseq) data sets form SD and poly/oligoarticular JIA patients or healthy controls (HC), in part including recombinant cytokine stimulation, were analyzed. Immortalized epithelial cells and ex vivo monocytes obtained from active and inactive SD patients and healthy controls were exposed to different in vitro stimuli and supernatants were analyzed for IL-18BP expression. SD patients' IL-18BP bioactivity in depleting IL-18 signalling in natural killer (NK) cells was tested.

**Results**

WB RNAseq data from SD patients compared to controls confirmed a disease-specific imbalanced expression of IL-18 *versus* IL-18BP. At the protein level, ex vivo analysis of SD patients' monocytes revealed significantly decreased IL-18BP expression by active *versus* inactive patients' cells. When cells were stimulated with different concentrations of IFNγ, active SD patients' monocytes produced consistently less IL-18BP compared to HC or inactive SD patients' cells. Regardless of expression level, when adjusted to equal concentration, IL-18BP expressed from SD patients' or healty control cells was equally competent in quenching IL-18 signaling on NK cells. When analyzing IL-18BP expression by myeloid and epithelial cells more broadly, we confirmed that IFNγ acts as its most prominent inducer, but IL18-BP expression could be further increased when cells were co-stimulated with specific inflammatory triggers. In striking contrast, on both protein and gene expression level IL-6 signaling readily reduced myeloid and epithelial cell baseline IL-18BP production and also restrained IL-18 PB expression upon cell stimulation with IFNγ. Following up on these findings we observed strong negative association of genes indicating IL-6 signaling with *IL18BP* transcription in SD patients' whole blood and Reactome analysis pinpointed IL-6 signaling as the one designated cytokine signaling pathway linking with reduced *IL18BP* expression in SD.

**Conclusion**

Our data identify IL-6 signaling as a prominent contributor in limiting IL-18BP expression in SD and thus potentially promoting IL-18/IL-18BP dysbalance and unrestricted IL-18 signaling. In conditions with high IL-18 in presence of elevated IL-6 this may argue for targeted (additive) therapeutic IL-6(R) inhibition in order to restore IL-18BP expression and thus re-establish physiological control over IL-18 signaling.

**Disclosure of interest**
None declared.

## O18

### Two-year outcomes in children with antineutrophil cytoplasmic antibody–associated vasculitis

I. Stojkic[1,*], D. Cabral[2], S. Shenoi[3], M. Twilt[4], V. Sivaraman[5], P. Riley[6], S. Tarvin[7], R. Luqmani[8], R. Beard [9], J. Bistrolarides [10], S. Charuvanij [11], A. Chun[12], K. Cook[13], S. Deepak[14], M. DeGuzman[15], B. A. Eberhard [16], M. E. Elder[17], S. Gagne[18], A. M. Huber[19], K. James[20], R. Khubchandani[21], S. Kim[22], T. Lee[23], L. N. Moorthy[24], A. Rosenberg[25], K. Brown[2], K. Morishita[2]
[1]Nationwide Children's Hospital, Columbus, USA; [2]University of British Columbia, Vancouver, Canada; [3]Seattle Children's, Seattle, USA; [4]University of Calgary, Calgary, Canada; [5]Nationwide Children's, Columbus, USA; [6]Manchester University, Manchester, UK; [7]Riley Children's, Indianapolis, USA; [8]Oxford University Hospitals, Oxford, UK; [9]London Health Sciences Centre, London, Canada; [10]Medical College of Wisconsin, Milwaukee, USA; [11]Mahidol University, Bangkok, Thailand; [12]Lurie Children's, Chicago; [13]Akron Children's, Akron, USA; [14]Nottingham Children's Hospital, Nottingham, UK; [15]Texas Children's Hospital, Houston; [16]Cohen Children's Medical Center, New Hyde Park; [17]University of Florida, Gainesville; [18]University of Pittsburgh, Pittsburgh, USA; [19]Dalhousie University, Halifax, Canada; [20]University of Utah, Salt Lake City, USA; [21]SRCC Children's Hospital, Mumbai, India; [22]University of Californa San Francisco, San Francisco; [23]Stanford, Palo Alto; [24]Rutgers University, New Brunswick, USA; [25]University of Saskatchewan, Saskatoon, Canada
**Correspondence:** I. Stojkic
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**Introduction**
While childhood-onset AAV (GPA, MPA) is characterized by greater disease severity and higher rates of organ dysfunction at disease onset compared to adult disease, knowledge and understanding of pediatric specific disease course and outcomes through the maintenance phase of therapy has been limited by disease rarity and the lack of large prospective datasets.
**Objectives**
Utilizing the largest available international registry-based dataset for pediatric AAV (ARChiVe), we aimed to characterize disease activity, treatment response, and damage accrual at the 24-months post diagnosis data entry point.
**Methods**
Data from ARChiVe patients with a diagnosis of GPA or MPA and complete data through 24 months were analyzed. Primary outcomes included inactive disease (PVAS=0), flare rates, and damage accrual (PVDI) at 24 months.
**Results**
The cohort comprised 157 children with completed 24-month data entry: 49% were white, 72% were female, 78% had GPA, median baseline PVAS 18 (IQR 14-22). Inactive disease (PVAS 0) was observed in 77% of patients, with 53% achieving corticosteroid-sparing remission (PVAS 0 and glucocorticoid dose < 0.2 mg/kg/day) by 24 months. Nearly all patients (98%) demonstrated a ≥50% reduction in PVAS from baseline to 12 months, and 67% showed a further ≥50% reduction in PVAS score between the 12- and 24-month assessments. 44% of patients experienced a flare during the 24 month period with 7.6% of these flares being major. We did a subset analysis looking at flare rates between subsets on rituximab ($n$ = 52) versus azathioprine ($n$ = 35) and these were not found to be significantly different. 24% of patients on rituximab maintenance were in flare at 24 months versus 20% of patients on azathioprine maintenance. The median PVDI at 24 months was 0 (range 0-7). 40% of patients had a PVDI ≥ 1 at 24 months and 28% had a PVDI ≥ 2.
**Conclusion**
This is the largest study reporting 24-month outcomes in pediatric AAV to date. While most patients responded to treatment and showed continued improvement between 12- and 24-months, nearly half of children flare despite maintenance therapy, and meaningful damage accrues in 40% by 24 months despite high remission rates highlighting the ongoing challenges of long-term disease control.
**Disclosure of interest**
None declared.

## O19

### Diagnosis characteristics from 205 children with childhood-onset Takayasu arteritis: an icTAK registry study

S. Gagne[1,2,*], J. Bistolarides[3], V. Sivaraman[4], K. Morishita[5,6], V. Podzolkova[7], M. Cüceoğlu[8], S. Al-Mayouf[9], M. Jelusic[10], M. Šestan[10], L. Wagner-Weiner[11], D. Cabral[5,6], S. Ozen[8] on behalf of The International Collaboration on Childhood-Onset Takayasu Arteritis (icTAK)
[1]Department of Pediatrics, University of Pittsburgh School of Medicine; [2]Division of Pediatric Rheumatology, University of Pittsburgh Medical Center, Pittburgh; [3]Medical College of Wisconsin, Milwaukee; [4]Division of Pediatric Rheumatology, Nationwide Children's Hospital, Columbus, USA; [5]Pediatric Rheumatology, British Columbia Children's Hospital; [6]University of British Columbia, Vancouver, Canada; [7]Department of Children's Diseases, N.F. Filatov Clinical Institute of Children's Health, Moscow, Russian Federation; [8]Division of Rheumatology, Hacettepe University, Ankara, Türkiye; [9]King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia; [10]University of Zagreb School of Medicine, Zagreb, Croatia; [11]Section of Pediatric Rheumatology, University of Chicago Comer Children's Hospital, Chicago, USA
**Correspondence:** S. Gagne
*Pediatric Rheumatology 2026*, **24(S1):**O19

**Introduction**
Takayasu arteritis is a rare chronic large vessel vasculitis predominantly affecting the aorta and its major branches. Approximately one-third of patients present in childhood, yet literature on childhood-onset Takayasu arteritis (cTAK) remains limited. Existing studies are further constrained by single center or single country, and small cohort sizes that have limited generalizability.
**Objectives**
To describe a large international cohort of childhood-onset Takayasu arteritis at diagnosis.
**Methods**
Data were obtained from the International Collaboration on Childhood-onset Takayasu Arteritis (icTAK) registry, established in 2023 in collaboration with the CARRA Chronic Childhood Vasculitis Work Group and PReS Vasculitis Working Party. Eligible patients were diagnosed in 2018 or later and fulfilled the 2008 Ankara criteria for cTAK. Data were collected at three timepoints: diagnosis, one-year, and final outcome. Continuous variables are reported as median (IQR) and categorical variables as frequency (%).
**Results**
205 children had complete diagnosis visit data, originating from 22 countries across 5 continents. Most were female (75.1%) and of Middle Eastern ethnicity (35.6%). Median age at diagnosis was 13.9 years (IQR 10.5–16.0). Multivessel involvement was common at diagnosis (median 4 vessels, IQR 3–6). The most affected vessels were the thoracic aorta (55.1%), ascending aorta/arch (53.2%), common carotid (49.8%), and celiac artery (45.9%). Cardiovascular (71.7%) and constitutional (66.3%) systems were most frequently involved, followed by renal (42.4%), neurological (35.6%), and abdominal (23.9%). Most patients received glucocorticoids (GC) at diagnosis (94.8%), with the majority (51.8%) receiving combined pulse intravenous methylprednisolone and oral GCs. Non-GC disease modifying agents (DMARDs) were initiated in 94% of patients: 57.9% received a conventional DMARD (cDMARD) alone, 10.4% a biologic DMARD (bDMARD) alone, and 25.7% received both. Methotrexate was the most common cDMARD (48.3%) and tocilizumab the most common individual bDMARD (13.2%), though TNF inhibitors were the most frequently prescribed class of bDMARDs (22%). Antihypertensives and anticoagulant/antiplatelet agents were prescribed to by 57% and 51% of patients respectively.
**Conclusion**
The icTAK registry represents the largest international cohort of cTAK ever assembled. There was a female preponderance, and most patients presented in early adolescence. While cardiovascular and constitutional manifestations predominated, multi-organ involvement was common. Use of glucocorticoids and DMARDs were near universal, though medication choice was highly varied reflecting the absence of established treatment protocols.
**Disclosure of interest**
None declared.

## O20

### Kawasaki disease and Anakinra: experience from a tertiary referral center

I. Mahé[1,*], F. Anselmi[1], L. Rossi-Semerano[1], C. Galeotti[1], M. Gattorno[2], I. Kone-Paut[1], C. Matucci-Cerinic[2], P. Dusser[1]
[1]Paediatric Rheumatology Department, Assistance Publique-Hôpitaux de Paris (AP-HP), Hôpitaux universitaires Paris-Sud - Hôpital Bicêtre, F-94270, Le Kremlin Bicêtre, France; [2]Rheumatology and Autoinflammatory diseases, IRCCS Istituto Giannina Gaslini, Genova, Italy
**Correspondence:** I. Mahé
*Pediatric Rheumatology 2026*, **24(S1):**O20

**Introduction**
Intravenous immunoglobulin (IVIG)-resistant Kawasaki Disease (KD) is associated with a high risk of coronary artery involvement.Anakinra, an interleukin-1 (IL1) receptor antagonist, is increasingly used as rescue therapy, but its impact on coronary outcomes remains poorly documented.
**Objectives**
To describe the characteristics of patients treated with Anakinra IVIG resistance KD and to explore the relationship between timing of Anakinra introduction and coronary evolution.
**Methods**
Retrospective single-center study at Bicêtre University Hospital, France, including children treated with subcutaneous Anakinra for IVIG-resistant Kawasaki disease (2016-2025).Data were collected at diagnosis (t0), Anakinra initiation (t1), last follow-up (t2).Data are presented as counts (%) and mean ± standard deviation/median [interquartile range].Among patients with coronary involvement at t1, exploratory linear regression analyses were performed to assess the association between treatment timing and coronary improvement.Two timing variables were examined: the delay from disease onset to Anakinra initiation and from IVIG administration to Anakinra initiation.Coronary improvement was defined as the decrease in maximal coronary Z-score between t1 and t2.Multivariable models were adjusted for age and baseline maximal Z-score at t1.Sensitivity analyses additionally adjusted for infliximab use, the concomitant therapy most likely to influence coronary outcomes.
**Results**
37 patients were included; 62% were male, with a median age of 10 months [1–89], and 22% (*n*=8) were younger than 6 months.The median time from diagnosis to Anakinra initiation was 14.6 days [6–33], and the median treatment duration was 15.8 days [1–60].At t1, 76% (*n*=28) had coronary artery involvement (Z-score >2), including 29% with giant aneurysms.At last follow-up, coronary abnormalities persisted in 56% (*n*=20) of patients.Among those with coronary involvement at t1, 33% (*n*=9) achieved normalization after a mean delay of 193 days. The mean change in maximal coronary Z-score was −0.9.Among the 22 patients with coronary involvement and complete follow-up data, a longer delay from disease onset to Anakinra initiation was independently associated with less coronary improvement after adjustment for age and baseline coronary severity (β = −0.84 per day; 95% CI −1.28 to −0.39; $p = 0.001$).Results were unchanged after additional adjustment for infliximab use and when using the delay from IVIG administration to Anakinra initiation as the exposure variable.
**Conclusion**
This cohort represents a severe population with a high proportion of giant coronary aneurysms, consistent with the use of Anakinra as rescue therapy in refractory KD.Earlier Anakinra initiation was associated with greater coronary improvement after adjustment for age and baseline coronary severity.These exploratory findings support the potential benefit of early IL-1 blockade and warrant confirmation in prospective studies.
**Disclosure of interest**
None declared.

## O21

### Clinical phenotypes and lupus low disease activity state in childhood-onset SLE: the multi-national CLASPER cohort

T. Arkachaisri[1], K. L. Teh[1,*], C. Bernal[2], S. P. Tang[3], S. Vilaiyuk[4], S. M. Al-Mayouf[5], S. Charuvanij[6], A. K. Guiang-Valerio[7], S. Chin Lim[8], B. Lerkvaleekul[4], N. Jidmahawong[9], N. Alamajed[5], A. Alsaleem[5], A. Alasmari[5], M. Sukharomana[6], S. Tangcheewinsirikul[6], H. M. R. Yeung[10], Y. X. Book[1], J. C. Huang[1], L. Das[1], S. Farman[11] on behalf of CLASPER (Childhood onset Lupus in ASia Pacific nEtwoRk)
[1]Rheumatology and Immunology, KK Women's and Children's Hospital, Singapore, Singapore; [2]Department of Pediatrics, Section of Pediatric Rheumatology, University of Santo Thomas Hospital, Manila, Philippines; [3]Department of Pediatrics, Selayang Hospital, Kuala Lumpur, Malaysia; [4]Division of Pediatric Rheumatology, Department of Pediatrics, Ramathibodi Hospital, Mahidol University, Bangkok, Thailand; [5]King Faisal Specialist Hospital & Research Center, Alfaisal University, Riyadh, Saudi Arabia; [6]Division of Pediatric Rheumatology, Department of Pediatrics, Siriraj Hospital, Mahidol University, Bangkok, Thailand; [7]Department of Pediatrics, Section of Pediatric Rheumatology, University of Santo Tomas Hospital, Manila, Philippines; [8]Department of Pediatrics, Parkcity Medical Centre, Kuala Lumpur, Malaysia; [9]Department of Pediatrics, Sawanpracharak Hospital, Nakorn sawan, Thailand; [10]Department of Paediatrics, Queen Elizabeth Hospital, Hong Kong SAR, China; [11]Department of Rheumatology, National Hospital Postgraduate Training and Medical Centre, Lahore, Pakistan
**Correspondence:** K. L. Teh
*Pediatric Rheumatology 2026,* **24(S1):**O21

**Introduction**
Childhood-onset systemic lupus erythematosus (cSLE) is a heterogeneous disease associated with substantial morbidity. However, multinational pediatric data from the Asia-Pacific region remain limited. The Childhood-Onset Lupus in Asia-Pacific Network (CLASPER) was established in 2025 as the first collaborative cSLE network in the region, with the overarching goal of advancing outcomes for affected children across Asia-Pacific.
**Objectives**
We aimed to describe the clinical and immunologic characteristics of the cohort and evaluate attainment and predictors of lupus low disease activity state (LLDAS).
**Methods**
Retrospective and prospective standardized clinical, laboratory, and outcome data were collected from six centers across five countries (Singapore, Thailand, Malaysia, Philippines, and Saudi Arabia). Patients fulfilling the 2012 SLICC classification criteria with available follow-up data were included. LLDAS was defined as clinical SLEDAI-2K ≤4 without major organ activity, Physician Global Assessment ≤1, no new disease activity, stable immunosuppressive therapy, and prednisolone ≤0.15 mg/kg/day (childhood LLDAS; cLLDAS) or ≤7.5 mg/day (adult LLDAS). Nonparametric statistics were applied for descriptive analyses. Kaplan–Meier analysis estimated time to LLDAS, while Cox regression identified predictors of attainment.
**Results**
A total of 1,408 patients were included in the baseline phenotype analysis (86.2% female; median age at diagnosis, 11.8 years [IQR, 9.6–14.0]). Disease burden at presentation was high, with a median baseline SLEDAI-2K of 13.0 (IQR 8.0–19.0). In this cohort, 13.9% of patients were diagnosed before age 8. Hematologic involvement was most common (63.9%), followed by fever (62.9%), acute cutaneous manifestations (52.7%), and renal disease (47.7%). Immunologic activity was prominent, with hypocomplementemia in 82.1%, anti-dsDNA positivity in 75.9%, and ANA positivity in 92.8%. Compared with the UK JSLE cohort, CLASPER patients had higher rates of renal, neuropsychiatric, and hematologic involvement but lower rates of arthritis and serositis. Among 495 patients with longitudinal follow-up, 94.5% achieved first LLDAS at a median of 14.2 months (IQR 12.8–15.5) from diagnosis. Cumulative probability of achieving LLDAS was 43% at 1 year, 73% at 2 years, and 83% at 3 years. In multivariable Cox regression, acute cutaneous manifestations (HR 0.78, 95% CI 0.65–0.94, $p$=0.008) and renal involvement (HR 0.83, 95% CI 0.69–0.99, $p$=0.045) independently predicted delayed attainment of LLDAS. Patients assessed using childhood-specific cLLDAS criteria had a longer time to target than those assessed using adult LLDAS definitions (log-rank $p$=0.014).
**Conclusion**
The CLASPER cohort demonstrates a severe and highly systemic cSLE phenotype across Asia-Pacific, with substantial renal and hematologic disease burden. Although LLDAS is achievable in most patients, attainment remains delayed in patients with renal and cutaneous involvement. Early

identification of high-risk phenotypes may enable stratified treatment approaches to accelerate disease control and improve long-term outcomes. Ongoing expansion of the CLASPER data will enable validation of cLLDAS as a treatment target across diverse populations in the region.

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**Disclosure of interest**
None declared.

## O22

### Rare variant burden and longitudinal outcomes in childhood-onset systemic lupus erythematosus

Y. Vyzhga[1,2,*], D. Dominguez[2], Z. Ding[2], A. M. Knight[1,2], D. M. Levy[1,2], L. T. Hiraki[1,2]
[1]University of Toronto; [2]The Hospital for Sick Children, Toronto, Canada
**Correspondence:** Y. Vyzhga
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**Introduction**
Childhood-onset systemic lupus erythematosus (cSLE) is clinically and genetically heterogeneous. Rare, highly penetrant variants can cause monogenic lupus and may contribute to earlier disease onset and phenotypic variation.
**Objectives**
We examined clinical manifestations, longitudinal disease activity, healthcare utilization, and cumulative damage across genetically stratified groups in cSLE.
**Methods**
Patients with cSLE from the SickKids Lupus Clinic with whole-exome or whole-genome sequencing data were included. Rare variants were filtered using QUAL >400 and gnomAD MAF <0.1%, annotated using ACMG-informed evidence, and prioritized by loss-of-function effect, ClinVar pathogenicity, or CADD ≥25 for missense variants. Patients were stratified into Group A, monogenic lupus/interferonopathy variants; Group B, high-impact rare variants without monogenic architecture; and Group C, no prioritized predicted pathogenic variants. Clinical features, longitudinal SLEDAI, admissions >2 days, and SLICC damage domains were compared across groups.
**Results**
The cohort included 148 patients; 84% were female. Median age at diagnosis was 9.0 years (IQR 7.5–10.6), with median follow-up of 10.4 years (IQR 7.1–13.9). The cohort was ancestrally diverse: European 26%, East Asian 20%, South Asian 16%, African 16%, mixed 14%, and American ancestry 7%, with no significant difference in ancestry distribution across groups ($p$=0.57). Eight patients (5%) were classified as Group A, 19 (13%) as Group B, and 121 (82%) as Group C. Group A had earlier disease onset (median 7.2 vs. 9.3 vs. 9.1 years; $p$=0.04) and a higher proportion of males (38% vs. 11% vs. 12%; $p$=0.03). Malar rash (38% vs. 58% vs. 79%; $p$=0.01) and arthritis (25% vs. 74% vs. 66%; $p$=0.04) were less frequent in Group A. Longitudinal SLEDAI trajectories, peak SLEDAI, and cumulative disease activity burden did not differ significantly. Inpatient admission >2 days occurred in 88% of Group A, 47% of Group B, and 53% of Group C ($p$=0.14). Recurrent hospitalization burden was highest in Group A (median 3.0 admissions/patient [IQR 3.0–3.5]) compared with Group B (1.0 [1.0–2.0]) and Group C (2.0 [1.0–4.0]) ($p$=0.07). Any documented damage occurred in 63%, 32%, and 32% of Groups A, B, and C, respectively ($p$=0.23), most commonly cataracts, avascular necrosis, osteoporosis/fracture, and end-stage renal disease.
**Conclusion**
In a cSLE cohort enriched for suspected rare SLE-risk variants, 5% had biallelic pathogenic recessive variants or heterozygous variants in genes with predicted dominant inheritance. This subgroup was younger at diagnosis and had lower frequencies of malar rash and arthritis, with numerically higher hospitalization and damage burden. These findings support further studies evaluating damage accrual and treatment exposure in relation to rare SLE genetic variant burden.
**Disclosure of interest**
None declared.

## O23

### Early cardiovascular vulnerability in childhood–onset systemic lupus erythematosus is not uniform but shaped by population–specific and regional factors: results from a large international cohort

C. Ciurtin[1,*], S. Arpaci[2], D. Demeneva[3], H. Stanescu[3], D. Schonenberg[4], N. Kifer[5], K.N. Au[6], A. Valikova[7], K. Ivankova[7], K. Bouchalová[7], E. Marrani[8], G. Simonini[8], R. Waller[9], S. Sener[10], H. I. Tüzen[11], B. Kasap Demir[12], M. Jelusic[13] on behalf of PReS Lupus Working Party
[1]Division of Medicine, Department of Ageing, Rheumatology and Regenerative Medicine; [2]Medical School; [3]Centre for Genetics and Genomics within the Department of Renal Medicine, University College London, London, UK; [4]Paediatric Rheumatology, Amsterdam UMC, Amsterdam, Netherlands; [5]Paediatric Rheumatology, and University Hospital Centre Zagreb, Zagreb, Croatia; [6]Department of Paediatrics and Adolescent Medicine, Tuen Mun Hospital, Hong Kong, China; [7]Paediatric Rheumatology, Palacký University Olomouc & University Hospital Olomouc, Olomouc, Czech Republic; [8]Paediatric Rheumatology, SODc Reumatologia, AOU Meyer, Florence, Italy; [9]Paediatric Rheumatology, Great Western Hospitals NHS Foundation Trust, Swindon, UK; [10]Paediatric Rheumatology, Adana City Training and Research Hospital, Adana; [11]Paediatric Rheumatology, İzmir City Hospital, Izmir, Türkiye; [12]Paediatric Rheumatology, İzmir City Hospital, Izmir, UK; [13]School of Medicine, University of Zagreb, Zagreb, Croatia
**Correspondence:** C. Ciurtin
*Pediatric Rheumatology 2026*, **24(S1):**O23

**Introduction**
Childhood-onset systemic lupus erythematosus (cSLE) carries an accelerated risk of cardiovascular disease (CVD) driven by chronic inflammation, immune dysregulation, and treatment-related metabolic effects.
**Objectives**
We aimed to identify most suitable CVD-risk assessment tools for use in cSLE and examine associations with patient and disease-related variables.
**Methods**
This multicentre retrospective–prospective cohort study recruited 309 cSLE patients from nine international centres across Europe and Asia. CVD risk at last follow–up was assessed using adult–derived tools (FRS, ASCVD, QRISK3, Globorisk) and the paediatric–derived PDAY score. Risk stratification, subgroup analyses, and correlation and regression analyses were performed to identify predictors of PDAY–estimated atherosclerotic burden.
**Results**
Among 309 patients, 81.2% were female; ethnicity was mainly White (63.8%), Asian (18.1%), and Black (11.7%). Median age at last follow–up was 20 years (IQR 17.6–23.3), age at onset 14.3 (12.0–16.3), disease duration 5.5 (2.6–10.1), and diagnostic delay 0.2 years (0.0–0.6). Mean BMI was 23.2 ± 5.7 kg/m². Median SLEDAI fell from 11.0 (6.0–16.0) at diagnosis to 1.0 (0.0–4.0) at last review; PedSDI was 0.0 (0.0–1.0). Adult–derived 10–year CVD risk was uniformly minimal: FRSlipids (100%), FRSBMI (99.3%), ASCVD (98.6%), and Globorisk (100%) classified almost all patients as very low risk, while QRISK3 showed broader distribution (75.5% very low, 12.6% low, 8.8% moderate, 3.1% high). In contrast, PDAY ($n$=175) showed higher burden: 28.0% very low, 33.1% low, 25.7% moderate, 13.1% high, with 38.9% scoring ≥6 points. PDAY scores were higher in European vs. Asian centres (median 5.0 [4.7–10.0] vs. 0.0 [0.0–3.5], $p$<0.001). High–PDAY patients were older (25.5 [21.0–30.0] vs. 18.8 [16.0–21.0] years, $p$<0.001), had longer disease duration (11.5 [7.0–17.0] vs. 5.1 [2.0–8.0] years, $p$<0.001), higher BMI (25.1 vs. 22.7 kg/m², $p$=0.019) and systolic BP (120 vs. 117 mmHg, $p$=0.034). Severe haematological (57.4% vs. 35.3%, $p$=0.006) and mucocutaneous involvement (47.1% vs. 27.1%, $p$=0.010) were more common. HCQ use (32.4% vs. 16.8%, $p$=0.017) and B–cell therapy (current 17.6% vs. 7.5%, $p$=0.042; ever 54.4% vs. 27.4%, $p$<0.001) were also higher. Serology, SLEDAI at last review, damage, and autoantibodies did not differ (all $p$>0.05). No patients received statins. Higher PDAY scores were independently associated with older age (β=0.773, 95% CI 0.64–0.91, $p$<0.001), male sex (β=3.56, 95% CI 1.74–5.37, $p$<0.001) and 'Other' ethnicity vs. White (β=7.03, 95% CI 3.09–10.96, $p$<0.001), while disease duration (β=0.004, $p$=0.89), SLEDAI at last assessment (β=0.22, $p$=0.26) and cumulative steroid dose (β=0.04, $p$=0.27) were not significant predictors in a multivariable regression analysis.

**Conclusion**
The PDAY score outperformed adult tools, revealing substantial early CVD risk burden in cSLE and clear demographic and geographical disparities. It identified a distinct high–risk subgroup driven by older age, male sex, and ethnic differences, with Black patients and those treated in European centres showing disproportionately elevated scores. PDAY therefore stands out as a measure with genuine discriminatory power for detecting early, clinically relevant CVD risk in cSLE.
**Disclosure of interest**
None declared.

## O24

### Treat-to-target in DMARD naive Juvenile idiopathic arthritis: 12-month clinical outcome of a randomised controlled trial comparing methotrexate monotherapy with polytherapy: the CHAMP study

B. van Dijk[1,*], L. van der Aa[2], M. Kruizinga[1], P. de Boer[1], M. van Hest[1], S. Kamphuis[3], J. M. van den Berg[4], E. Schatorjé[5,6], R. Verstegen[7,8], Y. Koopman-Keemink[9], P. van Pelt[10], S. A. Bergstra[11], R. ten Cate[1], P. Hissink Muller[1], D. Brinkman[1]
[1]Paediatrics – Division of paediatric rheumatology, Willem-Alexander Children's Hospital, Leiden University Medical Centre, Leiden; [2]Paediatrics, Zaans Medical Centre, Zaandam; [3]Paediatric Rheumatology, Sophia Children's Hospital, Erasmus University Medical Centre, Rotterdam; [4]Paediatric Immunology, Rheumatology and Infectious Diseases, Emma Children's Hospital, Amsterdam University Medical Centres, Amsterdam; [5]Paediatric Rheumatology, St Maartenskliniek; [6]Paediatric Rheumatology and Immunology, Amalia Children's Hospital, Radboud University Medical Centre, Nijmegen, Netherlands; [7]Paediatrics – Divisions of Rheumatology and Clinical Pharmacology/Toxicology, The Hospital for Sick Children; [8]Paediatrics, University of Toronto, Toronto, Canada; [9]Paediatrics, Juliana Children's Hospital, HagaZiekenhuis, The Hague; [10]Rheumatology, Erasmus University Medical Centre, Rotterdam; [11]Rheumatology, Leiden University Medical Centre, Leiden, Netherlands
**Correspondence:** B. van Dijk
*Pediatric Rheumatology 2026*, **24(S1):**O24

**Introduction**
First-line use of biologic DMARDs (bDMARDs) for non-systemic juvenile idiopathic arthritis (JIA) has been suggested instead of methotrexate (MTX) because of improved efficacy and shorter time to response, but would involve disadvantages such as higher costs, parenteral administration and limited access in low-income countries. We hypothesised that an alternative approach, polytherapy with three conventional DMARDs (csDMARDs) as initial treatment, could increase inactive disease attainment and decrease bDMARD use.
**Objectives**
To investigate whether MTX, sulfasalazine (SSZ) and hydroxychloroquine (HCQ) polytherapy increases attainment of inactive disease and decreases the need for bDMARD treatment at 6 months, compared to MTX monotherapy. Inactive disease and bDMARD treatment at 12 months were secondary outcomes.
**Methods**
This was an investigator-initiated, open-label randomised controlled trial (RCT) in DMARD naive patients aged 2–16 years with recent-onset (≤18 months) non-systemic JIA according to ILAR classification criteria. Participants were randomised into monotherapy (MTX) or polytherapy (MTX, SSZ and HCQ). Both arms included prednisolone bridging for 6 weeks. A treat-to-target protocol was used for 12 months. Every 3 months, blinded investigators assessed disease activity, which steered treatment adjustments. In both groups, active disease indicated switching MTX from oral to subcutaneous administration at 3 months; and initiation of a bDMARD (etanercept or adalimumab) at 6–12 months.
**Results**
63 JIA-patients were in the monotherapy and 68 in the polytherapy arm (56% and 59% polyarticular, respectively). At 6 months, inactive disease was attained in 54% (35/65) of patients in the polytherapy compared to 37% (23/62) in the monotherapy group ($p$=0.08). At 12 months, inactive disease was statistically significantly more frequent in the polytherapy (77%; 51/66) than in the monotherapy group (55%; 34/62, $p$=0.009). bDMARD use or initiation was numerically less frequent in the polytherapy than in the monotherapy arm: 42% (28/67) vs. 51% (32/63; $p$=0.38) at 6 months and 49% (33/67) vs. 65% (41/63; $p$=0.08) at 12 months. Disease activity (JADAS10) was not significantly different ($p$>0.05 on all timepoints). AEs were generally mild and comparable in both arms. Severe AEs (SAEs) concerned 3 hospital admissions due to intercurrent infections of patients in the monotherapy group, who recovered well and could continue the trial. No SAEs were recorded in the polytherapy arm.
**Conclusion**
Non-systemic JIA patients who started on polytherapy with three csDMARDs (MTX, SSZ, HCQ) attained inactive disease more often than patients who started on standard monotherapy (MTX), with numerically less bDMARD use. The difference regarding inactive disease was statistically significant at 12 months.
**Trial registration identifying number**
Dutch Trial Register NL-OMON53043.
**Disclosure of interest**
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## O25

### An investigation of the effectiveness of a biopsychosocial model-based exercise approach in children with scleroderma: preliminary results of a single-blind randomized controlled trial

O. Tüfekçi[1,*], M. O. Erkan[2], A. Barlak[1], S. Demirel[3], M. R. Onur[4], B. Oğuz[4], F. Korkusuz[3], A. Akdoğan[5], Y. Bilginer[2], S. Özen[2], E. Ünal[1]
[1]Department of Physical Tharapy and Rehabilitation, Faculty of Health Sciences, Alanya University, Antalya, [2]Department of Pediatrics, Division of Rheumatology, [3]Faculty of Physical Therapy and Rehabilitation, [4]Department of Sport Medicine, [5]Department of Radiology, [6]Department of Internal Medicine, Division of Rheumatology, Hacettepe University, Ankara, Türkiye
**Correspondence:** O. Tüfekçi
*Pediatric Rheumatology 2026*, **24(S1):**O25

**Introduction**
Scleroderma is a rare chronic connective tissue disease with two forms: localized and systemic. Juvenile scleroderma is challenging to evaluate and manage, with biopsychosocial impacts such as pain, fatigue, anxiety, depression, and lower quality of life. Thus, biopsychosocial interventions are needed. The Cognitive Exercise Therapy Approach (BETY - *Bilişsel Egzersiz Terapi Yaklaşımı*) is a biopsychosocial model-based exercise method for rheumatic diseases. While BETY's benefits have been demonstrated in adults with scleroderma, its efficacy in children remains unknown (1).
**Objectives**
The aim of this study was to examine the preliminary results of BETY's effects in children with scleroderma.
**Methods**
This single-blind RCT included children aged 7–18 with scleroderma, assigned to a study group (SG) or control group (CG). All participants received one session of function-oriented core stabilization exercise (a BETY component), and the SG also received chronic pain and mood management with a mentor physical therapist. The CG received a brochure with the same exercises for home use and kept an exercise diary. Both groups continued exercises twice weekly for three months. The participants were assessed using the Juvenile Arthritis Biopsychosocial Scale (JAB-Q Patient and Family Forms), the Childhood Health Assessment Questionnaire, the Maximum Mouth Opening (MMO), the Time Up and Go (TUG), the 30-Second Sit and Stand Test (30-SST), the 6-Min-

ute Walk Test (6MWT), inflammatory markers, and the Isokinetic knee strenght with dinamometer; their parents were assessed using the Juvenile Arthritis Biopsychosocial Questionnaire for Parents at baseline and at 3 months. Statistical analysis used the Wilcoxon signed-rank test for within-group comparisons and the Mann-Whitney U test for between-group comparisons.

**Results**

Baseline characteristics (age, BMI, and disease duration) were similar between groups (each with 9 participants: 3 systemic and 6 localized scleroderma). The study group showed significant within-group improvements in JAB-Q Patient and Family Forms, MMO, TUG, 30-SST, 6MWT, inflammatory markers, and isokinetic knee strength ($p < 0.05$).

**Conclusion**

This trial's preliminary results demonstrated that BETY significantly improved biopsychosocial, physical, and functional outcomes in children with scleroderma. BETY may offer a valuable biopsychosocial approach for pediatric scleroderma. Future studies should assess its long-term effects.

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**Trial registration identifying number**

ClinicalTrials.gov NCT07292961.

**Disclosure of interest**

None declared.

## O26

### Presence of nailfold capillary abnormalities is associated with more severe organ involvement in juvenile systemic sclerosis: results from the juvenile systemic scleroderma inception cohort

I. Foeldvari[1,*], J. Klotsche[2], K. Torok[3], O. Kasapcopur[3], A. Adrovic[3], B. Feldman[3], J. Anton[3], F. Sztajnbok[3], M. T. Terreri[3], A. P. Sakamoto[3], S. Johnson[3], S. Hajek[3], V. Stanevicha[3], G. Özomay Baykal[3], R. Khubchandani[3], C. Pain[3], D. Schonenberg-Meinema[3], E. Alexeeva[3], H. D. Dara Kar[3], M. Katsicas[3], S. Sawhney[3], V. Smith[3], S. Abu Al-Saoud[3], E. Al-Abadi[3], S. Appenzeller[3], T. Avcin[3], S. O. Hetlevik[3], M. Kostik[3], T. Lehman[3], S. Li[3], H. Malcova[3], E. Marrani[3], A. Patwardhan[3], W.-A. Sifuentes-Giraldo[3], N. Cabrera[3], P. Costa Reis[3], M. Janarthanan[3], M. Moll[3], D. Nemcova[3], M. J. Santos[3], N. Vasquez-Canizares[3], C. Battagliotti[3], L. Berntson[3], B. Bica[3], J. Brunner[3], D. Eleftheriou[3], L. Harel[3], A. Horne[3], G. Horneff[3], D. Kaiser[3], T. Kallinich[3], D. Lazarevic[3], J. M López Benítez[3], K. Minden[3], S. Nielsen[3], F. Nuruzzaman[3], B. Ogunjimi[3], M. Sparchez[3], É. Tuba[3], Y. Uziel[3], N. Helmus[1]

[1]Hamburg Centre for Pediatric and Adolescence Rheumatology, Hamburg; [2]German Rheumatism Research Center, Berlin; [3]jSSc collaborative group, Hamburg, Germany

**Correspondence:** I. Foeldvari

*Pediatric Rheumatology 2026*, **24(S1):**O26

**Introduction**

Juvenile systemic sclerosis (jSSc) is a rare pediatric rheumatic disease with significant morbidity. The Juvenile Systemic Scleroderma Inception Cohort (jSScC; www.juvenile-scleroderma.com) is the largest prospective cohort worldwide. In adult systemic sclerosis, nailfold capillary abnormalities are associated with more severe disease, but data in jSSc remain limited. This study evaluates whether the presence of nailfold capillary abnormalities correlates with distinct organ involvement and disease burden in jSSc.

**Objectives**

To assess the difference in patients with jSSc with normal (NF-) and abnormal NF(NF+) findings at the time of inclusion in the cohort.

**Methods**

Baseline data were analyzed from jSScC patients enrolled up to 15 March 2026 with documented nailfold capillary assessment at cohort entry. Nailfold capillary status was recorded on the standardized clinical research form as present (NF+) or absent (NF−), assessed using otoscope, ophthalmoscope, or capillaroscope. Demographic characteristics, clinical organ involvement, laboratory parameters, and patient- and physician-reported outcomes were compared between NF+ and NF− groups using chi-square and non-parametric tests.

**Results**

Among 280 patients, 218 (78%) had nailfold capillary abnormalities (NF+). Sex distribution, disease duration, and age at onset of Raynaud's phenomenon did not differ between groups. NF− patients were more often of Caucasian origin (82% vs. 69%, $p$=0.039). ANA positivity was significantly higher in NF+ patients (94% vs. 80%, $p$=0.002), without differences in extractable nuclear antigen (i.e. Scl-70). NF+ patients showed a more severe clinical phenotype. History of digital ulceration was more frequent (59% vs. 28%, $p$<0.001). Abnormal findings on high-resolution chest CT occurred more often in NF+ patients (49% vs. 28%, $p$=0.008). Gastrointestinal involvement, particularly oesophageal disease, was significantly more common (45% vs. 19%, $p$=0.002), with a trend toward lower BMI < −2 z-score (20% vs. 8%, $p$=0.056). Musculoskeletal manifestations were increased, including reduced joint range of motion (65% vs. 46%, $p$=0.005), muscle weakness (23% vs. 2%, $p$<0.001), and contractures (53% vs. 34%, $p$=0.008). Patient- and physician-reported global disease damage scores and patient-reported Raynaud activity were significantly higher in NF+ patients.

**Conclusion**

In this large jSSc cohort, clinically documented nailfold capillary abnormalities are associated with a distinct and more severe organ involvement pattern and higher disease burden. Nailfold capillary assessment provides meaningful clinical information in jSSc and supports its routine inclusion in patient evaluation.

**Disclosure of interest**

None declared.

## O27

### Distinct organ involvement and outcome profiles in diffuse versus limited juvenile systemic sclerosis: updated results from the juvenile systemic scleroderma inception cohort

I. Foeldvari[1,*], J. Klotsche[2], K. Torok[3], O. Kasapcopur[3], A. Adrovic[3], B. Feldman[3], R. Khubchandani[3], J. Anton[3], F. Sztajnbok[3], M. T. Terreri[3], A. P. Sakamoto[3], S. Johnson[3], S. Hajek[3], V. Stanevicha[3], G. Özomay Baykal[3], E. Alexeeva[3], D. Schonenberg-Meinema[3], H. D. Dara Kar[3], C. Pain[3], E. Al-Abadi[3], M. Katsicas[3], S. Sawhney[3], V. Smith[3], S. Abu Al-Saoud[3], S. Appenzeller[3], T. Avcin[3], N. Cabrera[3], S. O. Hetlevik[3], M. Kostik[3], T. Lehman[3], S. Li[3], H. Malcova[3], E. Marrani[3], A. Patwardhan[3], W.-A. Sifuentes-Giraldo[3], N. Vasquez-Canizares[3], P. Costa Reis[3], M. Janarthanan[3], M. Moll[3], D. Nemcova[3], M. J. Santos[3], C. Battagliotti[3], L. Berntson[3], B. Bica[3], J. Brunner[3], D. Eleftheriou[3], L. Harel[3], A. Horne[3], G. Horneff[3], D. Kaiser[3], T. Kallinich[3], D. Lazarevic[3], J. M López Benítez[3], K. Minden[3], S. Nielsen[3], F. Nuruzzaman[3], B. Ogunjimi[3], P. Seferna[3], M. Sparchez[3], É. Tuba[3], Y. Uziel[3], N. Helmus[1]

[1]Hamburg Centre for Pediatric and Adolescence Rheumatology, Hamburg; [2]German Rheumatism Research Center, Berlin; [3]jSSc collaborative group, Hamburg, Germany

**Correspondence:** I. Foeldvari

*Pediatric Rheumatology 2026*, **24(S1):**O27

**Introduction**

Juvenile systemic sclerosis (jSSc) is a rare, severe childhood-onset disease with an estimated prevalence of approximately 3 per 1,000,000 children. The Juvenile Systemic Scleroderma Inception Cohort (jSScC; www.juvenile-scleroderma.com) is the largest prospective cohort worldwide, enabling systematic evaluation of organ involvement and patient- and physician-reported outcomes over time. Previous analyses of the first 150 patients suggested subtype-specific differences between diffuse (djSSc) and limited (ljSSc) disease that diverge from adult systemic sclerosis. We now report updated baseline findings from 300 patients enrolled in the jSScC.

**Objectives**

To study and compare the clinical presentation of jSSc patients with djSSc and ljSSc subtypes at the time of inclusion in the cohort.

**Methods**

Baseline data from patients enrolled in the jSScC up to 15 March 2026 were analyzed. Demographic characteristics, clinical manifestations, laboratory parameters, organ involvement, and patient- and physician-reported outcomes at cohort entry were extracted. Comparisons between djSSc and ljSSc were performed using chi-square tests for categorical variables and non-parametric methods for continuous variables, as appropriate.

**Results**

A total of 300 patients were included; 68% ($n$=204) had djSSc and 32% ($n$=96) ljSSc. Overlap features were present in 14.7% of djSSc and 36% of ljSSc patients. The female-to-male ratio was lower in djSSc (3.5:1)

compared with IjSSc (5.4:1). Median age at onset of Raynaud's phenomenon (10.1 vs. 11.8 years) and of first non-Raynaud manifestation (10.5 vs. 12.0 years) was younger in djSSc. Antibody profiles differed between subtypes across ENA specificities. Elevated CRP was more frequent in djSSc (12% vs. 4%, $p$=0.092). Cutaneous and vascular involvement was significantly more pronounced in djSSc, including higher modified Rodnan skin scores, sclerodactyly (84% vs. 57%, $p$<0.001), telangiectasia (42% vs. 22%, $p$=0.001), active digital ulceration (21% vs. 10%, $p$=0.021), and history of ulceration (62% vs. 33%, $p$=0.001). Gastrointestinal involvement overall was common, with oesophageal involvement occurring more frequently in djSSc (44% vs. 32%, $p$=0.045), and severe underweight (BMI < −2 z-score) was markedly more prevalent in djSSc (21% vs. 6%, $p$=0.001). No statistically significant subtype differences were observed for cardiopulmonary, renal, musculoskeletal, or neurological involvement. Across all assessed physician- and patient-reported outcomes, djSSc patients demonstrated significantly higher disease activity, damage, and ulceration scores.

**Conclusion**

In this updated analysis of 300 patients, djSSc is characterized by earlier disease onset, more severe cutaneous, vascular, and gastrointestinal involvement, poorer nutritional status, and higher perceived disease burden compared with IjSSc. The organ involvement pattern differs from our earlier cohort analysis[2] and continues to contrast with adult systemic sclerosis, underscoring the importance of pediatric-specific disease characterization and longitudinal follow-up.

**Disclosure of interest**

None declared.

## O28

### Childhood–onset Sjogren's disease across the world: clinical and serological features, and early outcomes in 311 children

C. Ciurtin[1,*], J. Liu[2], M. Gorasia[2], S. Cooray[3], M. Shamsuddin[3], E. Legger[4], H. Bootsma[5], E. Kilic Konte[6], J. Antón[7], J. M. Mosquera Angarita[7], C. Zinterl[8], F. Oliveira Ramos[8], M. Spârchez[9], N. Gurbuz[10], R. Yatabe[11], Y. Bayindir[12], D. Ünal[12], D. Lazarevic[13], I. Madureira[14], M. Conde[14], S. Sampath[15], S. Abu Rumeileh[16], L. Paterson Brown[17], S. Parry[18], G. Varnier[19], B. Kasap Demir[20], K. Bouchalová[21], A. Abushhiwa[22], H. Emine Sönmez[23], M. Al-Obaidi[3], S. Sahin[6], S. Melo-Gomes[24] on behalf of PReS Sjogren Interest Group (Rosemary Waller; Bushra Aladaileh; Karla Vanessa Mendiola; Joana Baptista Lima; Soraia Azevedo; Marta Cabral; Sandra Sousa)

[1]Department of Ageing, Rheumatology and Regenerative Medicine; [2]Medical School, University College London; [3]Paediatric Rheumatology, Great Ormond Street Hospital for Children, London, UK; [4]Paediatric Rheumatology; [5]Department of Rheumatology and Clinical Immunology, University Medical Center Groningen, Groningen, Netherlands; [6]Paediatric Rheumatology, Cerrahpaşa Medical Faculty, Istanbul, Türkiye; [7]Servicio de Reumatología Pediátrica, Hospital Sant Joan de Déu, Barcelona, Spain; [8]Paediatric Rheumatology, Hospital de Santa Maria, Lisboa, Portugal; [9]2nd Department of Paediatrics, Iuliu Haţieganu University of Medicine and Pharmacy, Cluj-Napoca, Romania; [10]Paediatric Rheumatology, Ege University Faculty of Medicine, Izmir, Türkiye; [11]Paediatric Rheumatology, Tokyo Metropolitan Children's Medical Center & Tokyo University Hospital, Tokyo, Japan; [12]Paediatric Rheumatology, Hacettepe University Faculty of Medicine, Ankara, Türkiye; [13]Paediatric Rheumatology, University of Niš, Belgrade, Serbia; [14]Paediatric Rheumatology, Hospital Dona Estefânea, Lisboa, Portugal; [15]Paediatric Rheumatology, Great North Children's Hospital, Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle, UK; [16]Paediatric Rheumatology, AOU Pisana, Santa Chiara Hospital, Pisa, Italy; [17]Paediatric Rheumatology, Royal Hospital for Children, Glasgow; [18]Paediatric Rheumatology, Royal Manchester Children's Hospital; [19]Paediatric Rheumatology, [16] Royal Manchester Children's Hospital, Manchester, UK; [20]Paediatric Rheumatology, İzmir Katip Çelebi University, Izmir, Türkiye; [21]Paediatric Rheumatology, Palacký University Olomouc & University Hospital Olomouc, Olomouc, Czech Republic; [22]Paediatric Rheumatology, University of Tripoli, Tripoli, Libya; [23]Paediatric Rheumatology, Kocaeli University Faculty of Medicine, Kocaeli, Türkiye; [24]Paediatric Rheumatology, Evelina London Children's Hospital, London, UK

**Correspondence:** C. Ciurtin

*Pediatric Rheumatology 2026*, **24(S1):**O28

**Introduction**

Childhood–onset Sjögren's disease (cSjD) is rare, and its clinical presentation, investigations, and long–term burden remain poorly defined.

**Objectives**

We aimed to characterise clinical features at diagnosis, cumulative manifestations, treatment exposure, and disease burden in a large cSjD cohort, and to assess alignment with adult/paediatric SjD classification criteria.

**Methods**

This multicentre cross–sectional study (Oct 2025–Apr 2026) retrospectively collected consecutive clinician–diagnosed cSjD cases, recording baseline data and, at last review, cumulative features, ESSDAI/ESSPRI, SSDDI, investigations and treatments, using a harmonised proforma.

**Results** A total of 311 CYP were included. Median age was 13.0 years at diagnosis (IQR 10.29–15.19) and 16.08 years at last review (14.08–18.0). Median symptom duration before diagnosis was 0.92 years (0.33–2.50) and total duration 4.42 years (2.17–7.83). The cohort comprised 246 females and 65 males (F: M 3.78:1), with no sex differences in ethnicity ($\chi^2$ $p$=0.39) or age at onset ($p$=0.55). Serology was performed in 307/311 (99%), salivary gland (SG) ultrasound in 306/311 (98%), SG biopsy in 265/311 (86%), ocular dryness testing in 218/311 (70%), and oral dryness testing in 256/311 (82%). Overall, 53.6% met ACR/EULAR 2016 criteria, 45% Bartůňková criteria, and 89.1% the Tomiita algorithm. Ocular dryness (42.5%), oral dryness (39.4%), and parotitis (32.2%) were the most common glandular features. Systemic findings included hypergammaglobulinaemia (68.2%), articular (50.7%), haematological (48.4%), and cutaneous (42.5%) involvement. Only 130/265 (49.06%) SG biopsies were diagnostic, while 226/306 (73.86%) CYP had ultrasound features suggestive of cSjD. Anti–Ro was positive in 219/308 (71.57%), anti–La in 146/308 (47.71%), and rheumatoid factor in 141/307 (46.23%). MALT lymphoma was diagnosed in 8/308 (2.6%), while 50/307 (16.39%) CYP had pulmonary, 32/307 (10.49%) peripheral and 43/307 (14.10%) central nervous system manifestations attributable to cSjD by their clinician. Cumulatively, hydroxychloroquine (123/307; 40.06%) and mycophenolate mofetil (79/307; 25.90%) were most commonly used; 37/307 (12.05%) received rituximab and 3/307 (<1%) belimumab. At last review, median ESSDAI was 4 (2–11), ESSPRI 2.6 (1–5), and SSDDI 1 (1–2). Among 111 CYP with full ESSPRI domain assessment, fatigue was 3 (1–6), joint pain 3 (1–5), and dryness 3 (1–4). ESSDAI correlated with ESSPRI ($r$=0.47, $p$<0.001), but neither was associated with disease duration, diagnostic delay, age, sex, ethnicity or BMI.

**Conclusion**

CYP with cSjD showed frequent systemic involvement but low parotitis prevalence than previosuly reproted. Under half had biopsy–confirmed cSjD despite frequent supportive ultrasound findings. As the largest cSjD cohort, these data define core diagnostic and longitudinal features, and emphasise the need for child–specific criteria.

**Disclosure of interest**

None declared.

## O29

### Uncovering distinct disease severity trajectories in juvenile systemic sclerosis through machine learning

F. Zulian[1], G. Mastrangelo[1,*], B. Castaldi[2], E. Zanatta[3], F. Vittadello[4], F. Tirelli[1], A. Meneghel[1]

[1]Division of Pediatric Rheumatology, Department of Women and Children Health; [2]Division of Pediatric Cardiology, Department of Women and Children Health; [3]Division of Rheumatology, Department of Medicine-DIMED, University of Padua; [4]Research and Statistical Analysis, Explora, Padua, Italy

**Correspondence:** G. Mastrangelo

*Pediatric Rheumatology 2026*, **24(S1):**O29

**Introduction**

Juvenile systemic sclerosis (JSSc) often presents with overlapping features, making phenotypic classification and disease progression difficult to define.

**Objectives**

To describe longitudinal disease progression in a cohort of children with JSSc, identify latent subgroups with distinct disease severity trajectories over time, and evaluate baseline predictors associated with trajectory class membership.

**Methods**

Retrospective cohort study using prospectively collected data from 45 patients with JSSc between January 2005 and December 2024. Children were included if fulfilled both pediatric and/or adults' classifica-

tion criteria for JSSc/SSc and had a minimum follow-up of four years. We considered fourJSSc subtypes: limited cutaneous subtype (lcSSc), diffuse cutaneous subtype (dcSSc), systemic sclerosis sine-scleroderma subtype (ssJSSc), and the newly proposed fibrotic subtype (fJSSc). Demographic, clinical, laboratory, and disease severity data were collected. The primary outcome was progression-free survival (PFS), defined as percentage change in the Juvenile Systemic Sclerosis Severity score (J4S) over time. A change exceeding ±25% from baseline was considered clinically significant. To describe disease progression Kaplan–Meier survival analysis was performed to estimate PFS, and a Growth Mixture Model was used to identify latent disease severity classes characterized by distinct longitudinal trajectories and their baseline predictors. For clinical validation, Kaplan–Meier survival analyses were performed to compare time-to-event outcomes among the latent trajectory classes.

**Results**

Thirty-eight JSSc patients (66% females), followed for ≥48 months, entered the study. PFS remained high in the fibrotic and limited cutaneous JSSc subtypes (80–100%) over the observation period, whereas the diffuse and sine scleroderma subtypes showed marked deterioration, with PFS decreasing to 54% and 25%, respectively. We identified two latent classes of disease severity: Class 1 (74% of subjects) showed low severity and good treatment response, Class 2 (26% of subjects) showed an active, progressive course with poor long-term outcomes. Baseline predictors of poor outcome (i.e., Class 2 membership) were J4S >7.5, treatment initiation >9 months from disease onset, and >1.5 organs involved. Kaplan–Meier analysis revealed distinct PFS patterns across GMM-derived classes. The comparison between the Kaplan–Meier curves of the predefined clinical subtypes to those derived from the GMM-identified latent classes showed good concordance (κ = 0.66).

**Conclusion**

We identified two latent classes of disease severity in JSSc with clear prognostic significance. High baseline J4S, delayed treatment, and greater organ involvement predicted a more severe disease course, paving the way for precision health strategies and optimized.

**Disclosure of interest**

None declared.

## O30

### Treatment patterns and predictors of systemic therapy initiation in childhood-onset Sjögren's disease: a prospective nationwide cohort study

S. Ancona[1,2], S. Campus[3], A. Marino[4], G. Simonini[5], F. Baldo[4], E. Marrani[5], V. Ibba[3], A. Mauro[6], F. Zulian[7], D. Rigante[8,9], E. Del Giudice[10], S. Pastore[11], V. Manfrè[12], A. Civino[13], C. Covizzi[14], R. Naddei[15], M. F. Gicchino[16], A. Uva[17], R. Loconte[18], L. Breda[19], M. Cappella[20], S. D'Elios[21], E. Longoni[22], C. Proietti Ciolli[23], M. Gattorno[1], C. Malattia[1,24,*]

[1]Rheumatology and Autoinflammatory Diseases Unit, IRCCS Istituto Giannina Gaslini, Genoa; [2]Department of Pathophysiology and Transplantation, University of Milan, Milan; [3]Pediatric Clinic and Rare Diseases, Ospedale Microcitemico Antonio Cao, Cagliari; [4]Pediatric Rheumatology, ASST Gaetano Pini-CTO, Milan; [5]Rheumatology Unit, Meyer Children's Hospital IRCCS, Florence; [6]Department of Childhood and Developmental Medicine, Fatebenefratelli-Sacco Hospital, Milan; [7]Department of Woman and Child Health, University of Padova, Padova; [8]Department of Life Sciences and Public Health, Fondazione Policlinico Universitario A. Gemelli IRCCS; [9]Università Cattolica Sacro Cuore, Rome; [10]Department of Maternal Infantile and Urological Sciences, Sapienza University of Rome, Latina; [11]Department of Pediatrics, Institute for Maternal and Child Health IRCCS "Burlo Garofolo", Trieste; [12]Division of Rheumatology, Academic Hospital Santa Maria della Misericordia, Azienda Sanitaria Universitaria Friuli Centrale, Udine; [13]Unit of Pediatric Rheumatology and Immunology, Vito Fazzi Hospital, Lecce; [14]mmunorheumatology Unit, Regina Margherita Children's Hospital, AOU Città della Salute e della Scienza di Torino, Turin; [15]Department of Translational Medical Sciences, University of Naples Federico II; [16]Department of Woman, Child and General and Specialized Surgery, University of Campania "Luigi Vanvitelli"; [17]General Pediatrics and Dermato-Immuno-Rheumatology, AORN Santobono-Pausilipon, Naples; [18]Department of Pediatrics, Giovanni XXIII Pediatric Hospital, Bari; [19]Department of Paediatrics, University of Chieti-Pescara, Chieti; [20]Paediatric Rheumatology, AUSL-IRCCS di Reggio Emilia, Reggio Emilia; [21]Paediatric Unit, Santa Chiara Hospital, Pisa; [22]Paediatric Unit, ASST Lariana, Como; [23]University of Ferrara, Ferrara; [24]Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genoa, Genoa, Italy

**Correspondence:** C. Malattia

*Pediatric Rheumatology 2026*, **24(S1):**O30

**Introduction**

Childhood-onset Sjögren's disease (cSjD) is a rare and underrecognized condition. The lack of longitudinal studies has resulted in limited knowledge regarding disease course, therapeutic management, and prognostic factors [1].

**Objectives**

To characterize disease course and treatment patterns and to identify predictors associated with the initiation of systemic therapy in the largest nationwide multicentre cohort of patients with cSjD.

**Methods**

Patients fulfilling the 2016 ACR/EULAR classification criteria for primary SjD who were evaluated at Italian paediatric rheumatology centres between May 2023 and February 2026 were prospectively enrolled and followed every six months. Comprehensive data on therapeutic exposure were collected retrospectively from diagnosis to study entry and prospectively thereafter. Survival analyses were performed to identify factors associated with systemic treatment initiation. Kaplan-Meier estimates were used to assess the cumulative probability of treatment initiation over time, while multivariable Cox proportional hazards models were applied to identify independent predictors.

**Results**

Seventy-two patients from 22 centres were included; 64 (89%) were female. Median age at diagnosis was 13.6 years (IQR 10.5-15.8), and median follow-up was 26.4 months (IQR 14.3–64.6). Both disease activity and patient-reported symptoms significantly improved over time, with median ESSDAI decreasing from 6.0 at study entry to 3.5 at last follow-up ($p$=0.032), and median ESSPRI from 3.0 to 1.8 ($p$=0.037). During follow-up, 63/72 patients (88%) initiated systemic therapy. Kaplan-Meier estimates showed a cumulative probability of treatment initiation of 43% at 1 month, 53% at 3 months, 63% at 6 months, 70% at 12 months, 80% at 24 months, and 84% at 36 months after diagnosis. In univariable survival analyses, elevated ESR, hypergammaglobulinemia and anti-SSA/SSB positivity were significantly associated with earlier initiation of systemic treatment. In multivariable Cox regression analysis, elevated baseline ESR emerged as the only independent predictor of earlier systemic therapy initiation (HR 2.44, 95% CI 1.42–4.20; $p$=0.01).

**Conclusion**

This study provides important real-world insights into treatment strategies and determinants of systemic therapy initiation in cSjD. Specifically, in this large nationwide cohort, systemic therapy was frequently started early after diagnosis, particularly in patients with inflammatory features. The lack of association between ESSDAI or ESSPRI scores and treatment initiation suggests that adult-derived tools may not adequately capture paediatric disease burden, underscoring the need for cSjD-specific assessment instruments.

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**Disclosure of interest**

None declared.

## O31

### Towards a consensus-driven and data-informed definition of flare in non-systemic juvenile idiopathic arthritis: initial results from an international flare task force

V. Rypdal[1,*], H. Brunner[2], F. O. Ramos[3], J. Swart[4], B. Feldman[5], S. Benseler[6], F. Debenedetti[7], E. Balay-Dustrude[8], M. Glerup[9], G. Horneff[10], D. B. Horton[11], A. Marino[12], F. McErlane[13], A. Migowa[14], K. Minden[15], E. M. Morgan[8], P. Hissink Muller[16], S. M. Orsi[17], S. Ozen[18], M. Rypdal[1], R. Gutiérrez Suárez[19], K. Tenbrock[20], M. Twilt[21], P. Weiss[22], H. Wittkowski[23], S. Worral[24], A. Consolaro[25], J. Guzman[26]

[1]UiT The Arctic University of Norway, Tromsø, Norway; [2]Cincinnati Children's Hospital Medical Center, Cincinnati, USA; [3]ULS Santa Maria

Hospital, Lisbon University, Lisbon, Portugal; [4]Wilhelmina Children's Hospital, University Medical Center, Utrecht, Netherlands; [5]The Hospital for Sick Children, Toronto, Canada; [6]Children's Health Ireland University College, Dublin, Ireland; [7]IRCCS Ospedale Pediatrico Bambino Gesu, Rome, Italy; [8]Seattle Children's Hospital and Research Center, Seattle, USA; [9]Aarhus University Hospital, Aarhus, Denmark; [10]Pediatric Hospital Asklepios St. Augustin, Sankt Augustin, Germany; [11]Rutgers Robert Wood Johnson Medical School, New Brunswick, USA; [12]ASST G.Pini-CTO, Milan, Italy; [13]Pediatric rheumatology, Newcastle Hospitals, Newcastle, UK; [14]Aga Khan University Hospital, Nairobi, Kenya; [15]German Rheumatism Research Centre (DRFZ), Berlin, Germany; [16]Willem Alexander Children's Hospital Leiden University Medical Center, Leiden, Netherlands; [17]Istituto Giannina Gaslini, Università di Genova, Genoa, Italy; [18]Hacettepe University, Ankara, Türkiye; [19]Hospital para el Niño, Instituto Materno Infantil del Estado, Toluca, Mexico; [20]RWTH Aachen University, Aachen, Germany; [21]University of Calgary, Calgary, Canada; [22]Children's Hospital of Philadelphia, Philadelphia, USA; [23]The University Hospital Muenster (UKM), Muenster, Germany; [24]University of Manchester, Manchester, UK; [25]Istituto Giannina Gaslini, Università di Genova, Genova, Italy; [26]University of British Columbia, Vancouver, Canada

**Correspondence:** V. Rypdal

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**Introduction**

A recent systematic literature review showed heterogeneity in flare definitions used across JIA clinical trials, hindering consistent evaluation of treatment effects and management of flares. An international task force has been convened to establish a globally accepted definition of flare for non-systemic (nsJIA) applicable to clinical practice, treat-to-target strategies, observational studies, and clinical trials.

**Objectives**

To conduct a pre-consensus assessment of task force opinions about flare in nsJIA and how it should be measured.

**Methods**

An electronic survey was distributed to the flare task force, comprised of experts from major pediatric rheumatology global entities, cohorts, and registries: BIKER, CAPRI, CARRA, CHYRRP, ERN-RITA, JIRcohort, JuMBO, NordicJIA cohort, OMERACT JIA, PAFLAR, PANLAR, PRCSG, PReS, PReSTaR, PR-COIN, PROKIND, and UCAN. The survey assessed: the conceptual understanding of disease flare in nsJIA and the measurement tools used to evaluate flare.

**Results**

The task force ($n$=24) provided input on the conceptual definition and measurement of flare. There was substantial agreement at a conceptual level: most ($n$=22) preferred a definition including "Measurable worsening of disease activity" and "Requires consideration of treatment adjustment."75% agreed the definition should apply regardless of prior disease activity, and 92% considered flare a condition necessitating treatment intensification. Regarding the measurement of flare, over 80% identified the following data elements as essential: active joint count (AJC, 100%), physician global assessment (PhGA, 92%), active uveitis (92%), and active enthesitis (83%).Patient/parent global assessment was considered essential by 67%.There was less agreement on how to operationalize the definition. Two-thirds (67%) preferred using absolute change (e.g., ≥2 joints) while 21% favored a combination of absolute and relative changes (e.g., 30% worsening with ≥2 joints). There was variation in their opinions about available definitions and measures used in studies. The following was rated as very appropriate or appropriate: Therapeutic intensification (71%), a JADAS increase above moderate disease activity (63%), recurrence of disease activity defined by PhGA alone (46%), and PRINTO/PRCSG criteria (38%). For each of these definitions, several task force members expressed substantial reservations.

**Conclusion**

While there was good pre-consensus agreement conceptually, there were substantial differences in preferred ways to measure flare and reservations about available definitions. We are therefore testing different methods in the CAPRI cohort followed by a formal consensus process to ensure a final definition that is both consensus-driven and data-informed, advancing consistency and rigor in clinical trials, treat-to-target strategies, and patient care worldwide.

**Disclosure of interest**

None declared.

## O32

### Still's disease associated lung disease: update from the European registry

C. Bracaglia[1,*], F. Minoia[2], C. Kessel[3], S. Vastert[4], M. Pardeo[1], A. Arduini[1], S. Horackova Fingerhutova[5], I. Nikishina[6], O. Basaran[7], N. Kiper[7], K. Belozerov[8], M. Kostik[8], S. Sahin[9], M. Glerup[10], R. Caorsi[11], A. Horne[12], G. Filocamo[2], H. Wittkowski[3], M. Jelusic[13], J. Anton[14], S. Khaldi-Plassart[15], A. Belot[15], G. Horneff[16], S. Palmer Sarott[17], E. Cannizzaro Schneider[17], L. Fotis[18], S. Pastore[19], A. Calin[20], I. Kone-paut[21], O. Kasapcopur[9], P. Dolezalova[5], S. Ozen[7], A. Ravelli[22], F. De Benedetti[1] and on behalf of MAS/sJIA Working Party
[1]Division of Rheumatology, IRCCS Ospedale Pediatrico Bambino Gesù, Roma; [2]Fondazione IRCCS Ca' Grande Ospedale Maggiore Policlinico, Milano, Italy; [3]Department of Pediatric Rheumatology & Immunology, WWU Medical Center (UKM), Münster, Germany; [4]Pediatric Rheumatology & Immunology, University Medical Center Utrecht, Utrecht, Netherlands; [5]Centre for Paediatric Rheumatology and Autoinflammatory Diseases, Department of Paediatrics and Inherited Metabolic Disorders, 1st Faculty of Medicine, Charles University and General University Hospital in Prague, Prague, Czech Republic; [6]V.A. Nasonova Research Institute of Rheumatology, Moscow, Russian Federation; [7]Department of Pediatrics, Hacettepe University, Ankara, Türkiye; [8]Saint-Petersburg State Pediatric Medical University, Saint-Petersburg, Russian Federation; [9]Department of Pediatric Rheumatology, Istanbul University-Cerrahpasa, Istanbul, Türkiye; [10]Department of Pediatrics, Aarhus University Hospital, Aarhus, Denmark; [11]Department of Pediatrics and Rheumatology, IRRCS Istituto G. Gaslini, Genova, Italy; [12]Department of pediatric rheumathology, Karolinska University Hospital, Stockholm, Sweden; [13]Department of Pediatrics, University of Zagreb School of Medicine, University Hospital Centre Zagreb, Zagreb, Croatia; [14]Pediatric Rheumatology, Hospital Sant Joan de Déu, Universitat de Barcelona, Barcelona, Spain; [15]Pediatric Nephrology, Rheumatology, Dermatology Unit, Hôpital Femme Mère Enfant, Hospices Civils de Lyon, Lyon, France; [16]Pediatrics, Asklepios Clinic Sankt Augustin, Sankt Augustin, Germany; [17]Department of Pediatric Rheumatology, University Children's Hospital Zurich, Zurich, Switzerland; [18]Pediatric Rheumatology Division, 3rd Department of Pediatrics, National and Kapodistrian University of Athens, "ATTIKON" General University Hospital, Athens, Greece; [19]IRCCS Burlo Garofolo, Trieste, Italy; [20]Marie Curie Children's Emergency Hospital, Bucharest, Romania; [21]Pediatric Rheumatology Department, CEREMAIA Bicêtre hospital, APHP, Le Kremlin Bicêtre University of Paris Saclay, Paris, France; [22]IRRCS Istituto Giannina Gaslini and Università degli Studi di Genova, Genova, Italy

**Correspondence:** C. Bracaglia

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**Introduction**

Lung disease (LD) is an emerging severe life-threatening complication of Still's Disease (SD) characterized by peculiar features. Patients with SD-LD are increasing, and available data are essentially provided by North American series.

**Objectives**

To evaluate the burden of SD-LD in Europe.

**Methods**

Patients with diagnosis of SD-LD, followed at European paediatric rheumatology centres were identified through a survey sent to the members of the MAS/sJIA Working Party of PReS.

**Results**

Data from 64 patients with SD-LD, diagnosed at 21 European paediatric rheumatology centres between 2007 and 2025, were collected. 62 patients were White-Caucasian; 39 were female. The median age at SD onset was 3.5 years and LD occurred after a median time of 1.9 years. 3 patients (5%) had trisomy 21. 86% (55) of the patients had at least one episode of MAS, 39 (71%) patients had >1 MAS episode. Clinical and lung features were overall comparable to those reported in previous described cohort. IL-18 levels, measured in 52 (81%) patients, were markedly elevated at time of SD onset and of LD diagnosis (median of 56.686 and 137.500 pg/ml respectively). Eosinophils count was measured in 56 (87%) patients, and it was >1.000 cell/mmc in 10 (21%) at time of SD onset and in 15 (27%) at time of LD diagnosis. HLA-DRB1 genotyping was performed in half of the patients (32/64). The HLA-DRB1*15 allele was found positive in 21/32 (65%), while the HLA-DRB1*11 was positive in 15/32 (47%). Twenty-seven (42%) patients experienced adverse drug reaction to a cytokine inhibitor: 13 to tocilizumab, 9 to anakinra

and 5 to both. Twelve patients (19%) did not receive either IL-1 or IL-6 inhibitors (IL-1/IL-6i) before LD diagnosis. The age at SD onset was comparable to that of patients who received IL-1/IL-6i prior to LD onset. Interestingly, disease duration at LD onset was significantly shorter in patients who had not received IL-1/IL-6i ($p$=0.02), raising the hypothesis that treatment with IL-1/IL-6i may control better SD and therefore, potentially delay LD onset. History of MAS and outcome of SD (ICU admission and death) were comparable in the two groups. After LD diagnosis, 55 (86%) patients received intravenous or oral glucocorticoids and additional treatments that were very heterogeneous across the entire cohort. Eight patients underwent hematopoietic stem cell transplantation. At last follow-up, LD worsened in 14 patients and 21 developed complications: 12 hypoxia, 8 O2 requirement and 8 pulmonary hypertension. 29 patients (45%) required ICU admission and 11 (17%) died.

**Conclusion**

SD-LD is seen in European patients. Clinical and lung features are comparable to those described in North American cohorts. Around 1 out of 5 of our patients did not receive IL-1/IL-6i prior to LD onset and had shorter disease duration at LD onset. These observations argue against a direct role of IL-1/IL-6i in the development of LD.

**Disclosure of interest**

C. Bracaglia Consultant with: Sobi, Novartis, F. Minoia Consultant with: Sobi, Speaker Bureau with: Novartis, C. Kessel Grant / Research Support with: Novartis, Consultant with: Novartis, Speaker Bureau with: Sobi, S. Vastert Consultant with: Sobi, Novartis, M. Pardeo: None declared, A. Arduini: None declared, S. Horackova Fingerhutova: None declared, I. Nikishina Speaker Bureau with: Pfizer, Roche, Novartis, Sobi, MSD, R-Pharm, Ipsen, O. Basaran: None declared, N. Kiper: None declared, K. Belozerov: None declared, M. Kostik: None declared, S. Sahin: None declared, M. Glerup: None declared, R. Caorsi Consultant with: Sobi, Novartis, A. Horne: None declared, G. Filocamo Consultant with: Sobi, H. Wittkowski: None declared, M. Jelusic: None declared, J. Anton Grant / Research Support with: Sobi, Novimmune, Novartis, Abbvie, Pfizer, GSK, Roche, Amgen, Lilly, BMS, Sanofi, Consultant with: Sobi, Novimmune, Novartis, Pfizer, GSK, Speaker Bureau with: Sobi, Novimmune, Novartis, GSK, Pfizer, S. Khaldi-Plassart: None declared, A. Belot Consultant with: SOBI, Novartis, Roche, Pfizer, G. Horneff Grant / Research Support with: MSD, Novartis, Roche, Speaker Bureau with: Abbvie, Chugai, Lilly, Sanofi, Novartis, Pfizer, S. Palmer Sarott: None declared, E. Cannizzaro Schneider: None declared, L. Fotis: None declared, S. Pastore: None declared, A. Calin: None declared, I. Kone-paut: None declared, O. Kasapcopur: None declared, P. Dolezalova: None declared, S. Ozen Consultant with: Sobi, Novartis, Speaker Bureau with: Sobi, Novartis, A. Ravelli Consultant with: Abbvie, Alexion, BMS, Galapagos, J&J, Novartis, Pfizer, Roche, SOBI, F. De Benedetti Grant / Research Support with: Roche, Sobi, Novimmune, Sanofi, Novartis, Elixiron; Regeneron, BMS, Consultant with: Sobi, Novartis, Apollo, Kiniksa.

## O33

### Immune phenotype identification in systemic juvenile idiopathic arthritis patients using high parameter flow cytometry

H. Kessler[1,*], N. Gibson[1], A. Sproles[1], C. S. Lages[1,2], P. Dascani[3], S. Thornton[1,2], G. Schulert [1,2]

[1]Division of Rheumatology, Cincinnati Children's Hospital Medical Center; [2]Department of Pediatrics, University of Cincinnati Medical Center, Cincinnati; [3]Cytek Biosciences, Pittsburgh, USA

**Correspondence:** H. Kessler

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**Introduction**

Systemic juvenile idiopathic arthritis (sJIA) is a unique subtype of juvenile arthritis that can present with life threatening complications such as chronic lung disease including interstitial lung disease, pulmonary alveolar proteinosis, pulmonary fibrosis, and pulmonary alveolar hypertension[1]. Despite such complications, the immunopathogenesis of sJIA-associated lung disease (sJIA-LD) is largely unknown.

**Objectives**

Our goal was to compare myeloid and lymphoid subsets in peripheral blood mononuclear cells (PBMCs) from sJIA-LD, sJIA without LD, and healthy controls. Findings were correlated to disease activity markers such as the lung disease global assessment scale (PGALD) and Krebs von den Lungen 6 (KL-6) level, a glycoprotein expressed on type II alveolar cells and reportedly increased in interstitial lung disease[2].

**Table 1 (Abstract O33)** Comparisons of CD4+ and B-cell subsets

| Cell Type | Conclusion |
|---|---|
| CD4+ Central Memory<br>CD4+ TH1 | Decreased in sJIA-LD |
| CD4+ Effector<br>CD4+ TEMRA | Increased in sJIA-LD |
| B-cells<br>Naive B-cells | Increased in SJIA-LD |
| Memory B-cells<br>Plasmablasts | Decreased in sJIA-LD |

**Methods**

sJIA-LD PBMC samples were obtained from the CARRA Registry and Biobank. sJIA non-LD and control PBMCs were obtained from Cincinnati Children's biorepository. Full spectrum flow cytometry was performed using the Auorora platform. Cell populations were identified using manual gating. Previously reported from this laboratory, a high parameter flow cytometry panel (28 parameters) was created to visualize myeloid and lymphoid cell subsets. GraphPad PRISM was used to analyze and visualize data. Statistical analysis utilized Welch ANOVA and Spearman correlation. This study was approved by the IRB and all patients/parents provided informed consent.

**Results**

We analyzed data from 35 sJIA-LD patients, 10 sJIA non-LD patients, and 15 controls. Total live singlet cells and percentage of CD45+ cells were similar across groups. Both sJIA groups showed increased monocyte and decreased lymphocyte percentages versus controls. In sJIA-LD, monocyte percentage positively correlated to PGALD and KL-6, while lymphocyte percentage negatively correlated to both. CD4+ T-cell percentages were similar across groups. Comparisons of T-cell and B-cell subsets are shown in Table 1.

**Conclusion**

Our results demonstrate an increase in peripheral monocytes and decrease in circulating lymphocytes in sJIA broadly, and percentages correlated with measures of lung disease activity. sJIA-LD patients exhibited specific alternations in both T and B cell subsets. The implications of these changes in unknown but support distinct immunologic differences in sJIA-LD. Our future work involves unsupervised analysis and evaluation of activation markers.

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**Disclosure of interest**

H. Kessler: None declared, N. Gibson: None declared, A. Sproles: None declared, C. Lages: None declared, P. Dascani Employee with: Cytek Biosciences, S. Thornton: None declared, G. Schulert Consultant with: SOBI paid to CCHMC, $10,000, Speaker Bureau with: SOBI paid to CCHMC, $10,000.

## PT001

### Baseline characteristics of the prospective, multicentre, international cohort for the new classification criteria of juvenile idiopathic arthritides

S. La Bella[1,2,*], A. Alongi[1], S. Magni-Manzoni[3], M. Jelusic[4], C. Malattia[5,6], O. Boyarchuk[7], I. Calvo Penadés[8], M. Rygg[9], R. Haviv[10,11], C. Lazar[12], P. Pal[13], S. Rusoniene[14], S. Pastore[15], J. Sánchez-Manubens[16], Z. Mukusheva[17], M. Cattalini[18], M. Hufnagel[19], L. Breda[20], Y. Butbul Aviel[21], T. Dallos[22,23], O. Synoverska[24], G. Capitoli[1], S. Galimberti[1], D. J. Lovell[25], A. Martini[26], H. I. Brunner[25], N. Ruperto[1,2] on behalf of Paediatric Rheumatology INternational Trials Organisation (PRINTO) on behalf of Paediatric Rheumatology INternational Trials Organisation (PRINTO)/Pediatric Rheumatology Collaborative Study Group (PRCSG)

[1]University of Milano-Bicocca; [2]Fondazione IRCCS San Gerardo dei Tintori, SC Reumatologia Pediatrica, PRINTO, Monza; [3]IRCCS Ospedale Pediatrico Bambino Gesù, UOC Rheumatology, Roma, Italy; [4]University Hospital Centre Zagreb, Zagreb, Croatia; [5]IRCCS Istituto Giannina Gaslini, UOC Rheumatology and Autoinflammatory Diseases; [6]University of Genova, Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, Genova, Italy; [7]I. Horbachevsky Ternopil National Medical University, Ternopil, Ukraine; [8]University Hospital La Fe, Valencia, Spain; [9]St. Olavs University Hospital of Trondheim, Trondheim, Norway; [10]Meir Medical Centre, Kfar Saba; [11]Gray School of Medicine, Tel Aviv University, Tel Aviv, Israel; [12]Children Emergencies Hospital, Cluj-Napoca, Romania; [13]Institute of Child Health, Kolkata, India; [14]Vilnius University, Vilnius, Lithuania; [15]IRCCS Burlo Garofolo, Trieste, Italy; [16]Parc Taulí Hospital Universitari. Institut d'Investigació i Innovació Parc Taulí (I3PT-CERCA), Barcelona, Spain; [17]Corporate Fund "University Medical Center" National Research Center for Maternal and Child Health, Astana, Kazakhstan; [18]Clinica Pediatrica dell'Università di Brescia, Spedali Civili, Brescia, Italy; [19]University Medical Center Freiburg, Freiburg, Germany; [20]Ospedale Policlinico – Università degli Studi di Chieti, Chieti, Italy; [21]Rambam Medical Center, Haifa, Israel; [22]National Institute of Children's Diseases; [23]Department of Paediatrics, Faculty of Medicine, Comenius University, Bratislava, Slovakia; [24]Regional Children's Hospital, Ivano-Frankivsk, Ukraine; [25]Cincinnati Children's Hospital Medical Center, Cincinnati, USA; [26]University of Genova, Genova, Italy
**Correspondence:** S. La Bella
*Pediatric Rheumatology 2026*, **24(S1):**PT001

**Introduction**
Juvenile idiopathic arthritis (JIA) has historically been classified according to the International League of Associations for Rheumatology (ILAR) criteria. However, several ILAR categories lack biological and clinical homogeneity. The development of new, evidence-based classification criteria is a key objective of the Paediatric Rheumatology INternational Trials Organisation (PRINTO) and the Pediatric Rheumatology Collaborative Study Group (PRCSG).
**Objectives**
To describe the baseline demographic, clinical, and laboratory characteristics of the prospective PRINTO/PRCSG cohort employed to develop the new evidence-based classification criteria for JIA.
**Methods**
A prospective, longitudinal, international cohort was established between 2020 and 2025 to enrol at least 1000 patients with new-onset JIA. Inclusion criteria were: (i) a diagnosis of JIA by the treating physician, (ii) clinical evaluations at 3-month intervals and at least annually up to 5 years, and (iii) availability for biosample collection. Demographic, clinical, and laboratory data, including centralized assessment of key laboratory parameters, were collected at onset and after 3 months.
**Results**
Overall, 1579 patients were assessed for eligibility and 1509 (95.6%) were included from 32 countries. Most patients were females (970, 64.3%), with a median age at onset of 6.8 years (Q1-Q3: 2.8-11.8). The majority (997, 66.1%) had never received steroids or DMARDs prior to enrollment. Patients had a median active joint count of 2 (Q1-Q3: 1-4). Among all patients, 846 (56.1%) experienced morning stiffness lasting more than 15 min. Psoriasis and dactylitis were observed in 41 (2.7%) and 72 (4.8%) patients, respectively; enthesitis was documented in 143 (9.5%) patients, while sacroiliitis was documented by MRI in 47/1426 (3.3%). Notably, 104 patients (6.9%) had systemic fever, 89 (5.9%) an evanescent erythematous rash, 37/1481 (2.5%) hepatomegaly, 25/1481 (1.7%) splenomegaly; 37/879 patients with ophthalmologic data (4.2%) had concomitant chronic anterior uveitis. Central laboratory evaluation was available for at least one sample in 1372 patients (90.9%). At onset, ANA positivity was observed in 671/1269 patients (52.9%) at a titre ≥1:160 and in 501/1269 (39.5%) at a titre ≥1:320. RF was positive at disease onset in 50/1267 patients (4%); ACPAs were positive in 64/1326 patients (4.8%). HLA-B27 was positive in 199/1248 patients (15.9%). ANA positivity in two occasions ≥3 months apart was associated with a higher prevalence of oligoarthritis compared with ANA-negative patients (OR 1.61, 95% CI 1.26–2.05); HLA-B27 positivity was associated with a higher prevalence of axial involvement than negative patients (OR 1.75, 95% CI 1.13–2.71). RF positivity and ACPAs were highly positively correlated (Gwet's AC1 of 0.96). According to the ILAR criteria, most patients would have been classified as oligoarthritis (670, 44.4%; extended in 39, 2.6%), followed by undifferentiated JIA (475, 31.5%) and RF-negative polyarthritis (180, 11.9%).
**Conclusion**
This international cohort provides a detailed baseline characterization of over 1500 children with new-onset JIA. The new classification criteria will be available after the planned 2026 consensus conference, based on evidence-derived data from this large cohort.
**Disclosure of interest**
None declared.

## PT002

### Can altered synovial fluid protein profiles serve as prognostic markers in oligoarticular juvenile idiopathic arthritis?

D. Aydın[1,*], N. Şahin[1], E. Koçyiğit[2], H. Taştanoğlu[3], C. Arslanoğlu[4]
[1]Pediatric Rheumatology; [2]Medical Biology, Kocaeli University, Kocaeli; [3]Pediatric Rheumatology, Erciyes University, Kayseri; [4]Pediatric Rheumatology, Kocaeli City Hospital, Kocaeli, Türkiye
**Correspondence:** D. Aydın
*Pediatric Rheumatology 2026*, **24(S1):**PT002

**Introduction**
Intra-articular corticosteroid injection (IACI) is considered a first-line treatment for oligoarticular juvenile idiopathic arthritis (oJIA). However, treatment response varies among patients.
**Objectives**
We investigated whether baseline synovial fluid proteomic profiles in treatment-naive patients with oJIA were associated with early clinical outcomes and explored candidate proteins that may predict the need for treatment escalation.
**Methods**
Thirty-three treatment-naive patients with oJIA treated with IACI were prospectively included. Synovial fluid samples obtained during IACI were stored at −80 °C. Patients were classified according to remission status at 3 months. Following protein isolation and trypsin digestion, samples were analyzed using nHPLC LC-MS/MS (QExactive Orbitrap). Label-free quantification was performed, and proteins with ≥2-fold expression differences were considered differentially expressed. Protein–protein interaction analysis was performed using the STRING database.
**Results**
Among 33 patients, 15 (45.5%) achieved clinical remission and 18 (54.5%) did not. There was no significant difference between the groups regarding ANA positivity, ESR, and CRP levels at baseline and at month 3. At month 3, systemic steroid and DMARD use were significantly higher in the non-remission group, whereas biologic therapy use showed no significant difference between groups. Clinical characteristics are shown in Table 1. A total of 13 proteins were differentially expressed between the remission and non-remission groups, with 9 upregulated and 4 downregulated proteins. Upregulated proteins included acute-phase reactants (ORM1, ORM2), neutrophil-associated proteins (S100A9, PRTN3), and several carrier proteins. Downregulated proteins included coagulation and anticoagulation pathway components (F5, PROC). Functional analyses demonstrated that the differentially expressed proteins were mainly enriched in biological processes related to acute-phase response, neutrophil activation, and hemostasis. Upregulated proteins formed a broader and highly interconnected interaction network, whereas downregulated proteins showed a more limited but high-confidence interaction, mainly centered on the PROC–F5 axis.

| Variable | Remission at 3 Months (*n*=15) | No Remission at 3 Months (*n*=18) | *p* value |
|---|---|---|---|
| **At the Time of IACI** | | | |
| Age (years), mean ± SD | 12.24 ± 5.60 | 13.90 ± 3.60 | 0.31 |
| Female sex, n (%) | 7 (46.7) | 7 (38.9) | 0.73 |
| Active joint count, median (IQR) | 1 (1–1) | 1 (1–2) | 0.06 |
| **Clinical Status at Month 3** | | | |
| Active joint count, median (IQR) | 0 (0–0) | 1 (0.25–2) | 0.002 |

**Conclusion**
Baseline synovial fluid proteomic differences suggest that innate immune and hemostatic pathways may influence the early clinical course of oJIA. These exploratory findings require validation in larger cohorts and may contribute to future biomarker studies predicting response to IACI.
**Disclosure of interest**
None declared.

## PT003

### Long-term efficacy of upadacitinib in patients with pcJIA: insights from SELECT–YOUTH trial across multiple therapy lines

H. I. Brunner[1], A. V. Ramanan[2], M. Mori[3,4], H. Schmeling[5], L. D. Smith[6], T. Gao[6], A. Garrison[6], J. Kannan[6], M. Avadisian[7], G. Horneff[8,9,*]
[1] University of Cincinnati, Cincinnati, USA; [2]Bristol Medical School, Bristol, UK; [3]St Marianna University School Medicine, Kawasaki; [4]Institute of Science Tokyo, Tokyo, Japan; [5]University of Calgary, Calgary, Canada; [6]AbbVie, Chicago, USA; [7]AbbVie, Saint-Laurent, Canada; [8]Asklepios Clinic, Sankt Augustin; [9]University Hospital of Cologne, Cologne, Germany
**Correspondence:** G. Horneff
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**Introduction**
Upadacitinib (UPA), an oral selective JAK inhibitor, has demonstrated safety and efficacy in pcJIA patients.
**Objectives**
To evaluate long-term efficacy of UPA in pcJIA patients previously treated with csDMARDs, bDMARDs, or TNFi only (subset of bDMARD population), in the phase 1 SELECT-YOUTH trial.
**Methods**
A total of 122 patients aged 2 to <18 years with pcJIA received a weight-based UPA dose. All analyses were performed as observed and stratified by prior exposure to csDMARDs, bDMARDs, and TNFi. Efficacy was assessed from week 12 to 48 using JIA-ACR 50/70/90/100 response rates, JADAS-27 (CRP), C-HAQ, and patient-reported pain scores (VAS 1-100).
**Results**
Of 122 patients who initiated UPA, 89% (109/122) completed 48 weeks of treatment. Prior to UPA, 66 (54.1%) received csDMARDs, 27 (22.1%) received bDMARDs, and 14 (11.5%) received TNFi. JIA-ACR response rates improved across all groups from week 12 to 48. At week 48, response rates were comparable regardless of prior csDMARD exposure (csDMARD-experienced vs. naive: JIA-ACR 50, 93.1% vs. 94.1%; JIA-ACR 70, 86.2% vs. 92.2%; JIA-ACR 90, 72.4% vs. 72.5%; JIA-ACR 100, 55.2% vs. 60.8%). Responses were similar across bDMARD-experienced, TNFi-experienced and bDMARD-naïve patients (bDMARD-experienced, TNFi-experienced vs. naive: JIA-ACR 50, 87.5%, 81.8% vs. 95.3%; JIA-ACR 70, 83.3%, 81.8% vs. 90.6%, JIA-ACR 90, 66.7%, 63.6% vs. 74.1%; JIA-ACR 100, 54.2%, 36.4% vs. 58.8%). Rates of low disease activity (JADAS-27 ≤3.8) and inactive disease (JADAS-27 ≤1) at week 48 were similar irrespective of csDMARD status (csDMARD-experienced vs. naive: JADAS-27 ≤3.8, 83.1% vs. 75.0%; JADAS-27 ≤1, 49.2% vs. 50.0%). Similarly, bDMARD status groups showed comparable results (bDMARD-experienced, TNFi-experienced vs. naive: JADAS-27 ≤ 3.8, 73.1%, 76.9 vs. 81.5%; JADAS-27 ≤ 1, 46.2%, 53.8% vs. 50.6%). Mean reduction from baseline in C-HAQ was similar regardless of csDMARD exposure, whereas pain reduction was greater in csDMARD-naïve patients (csDMARD-experienced vs. naive: C-HAQ, -0.64 vs. -0.64; pain, -25.0 vs. -37.3). Mean reduction in C-HAQ and pain were greater in bDMARD-naïve (bDMARD-experienced, TNFi-experienced vs. naive: C-HAQ, -0.49, -0.24 vs. -0.68; pain, -21.3, -18.8 vs. -33.4).
**Conclusion**
Upadacitinib demonstrated marked and consistent efficacy in pcJIA patients, regardless of prior csDMARD or bDMARD exposure.
**Trial registration identifying number**
NCT03725007.
**Disclosure of interest**
H. Brunner Grant / Research Support with: AbbVie, A. Ramanan Grant / Research Support with: AbbVie, M. Mori Grant / Research Support with: Received unrestricted research grants for the former Department of Lifetime Clinical Immunology from AbbVie GK, Ayumi Pharmaceutical Corporation, Chugai Pharmaceutical Co., Ltd., CSL Behring K.K., Japan Blood Products Organisation, Nippon Kayaku Co., Ltd., UCB Japan Co., Ltd., and Asahi Kasei Pharmaceutical Corporation, Speaker Bureau with: AbbVie, H. Schmeling Grant / Research Support with: Pfizer, AbbVie, UCB, Sanofi, Consultant with: Sanofi, AbbVie, L. Smith Employee with: AbbVie, T. Gao Employee with: AbbVie, A. Garrison Employee with: AbbVie, J. Kannan Employee with: AbbVie, M. Avadisian Employee with: AbbVie, G. Horneff Grant / Research Support with: Novartis, Consultant with: Novartis.

## PT004

### Novel subgroups of early juvenile psoriatic arthritis identified using machine learning: results from a multinational collaboration of inception cohorts

S. J. W. Shoop-Worrall[1,*], K. L. Hyrich[2,3] on behalf of CAPS Investigators, J. Guzman[4] on behalf of ReACCh-Out Investigators, K. Minden[5,6], J. Klotsche[7], R. Berard[8] on behalf of CAPRI Investigators, R. Queiro[9], M. Yates[10,11], G. M. Alenius[12], F. McErlane[13], C. Ciurtin[14], G. Cleary[15], L. Coates[16], N. Geifman[17]
[1]The University of Manchester, Manchester, UK; [2]Centre for Musculoskeletal Research, The University of Manchester; [3]NIHR Manchester BRC, Manchester University NHS Foundation Trust, Manchester Academic Health Science Centre, Manchester, UK; [4]UCB, Vancouver, Canada; [5]Programme area Epidemiology and Health Services Research, Deutsches Rheuma-Forschungszentrum Berlin; [6]Department of Paediatric Respiratory Medicine, Immunology and Critical Care Medicine, Charité - Universitätsmedizin Berlin, corporate member of Freie Universität Berlin and Humboldt - Universität zu Berlin; [7]Programme area Epidemiology and Health Services Research, Deutsches Rheuma-Forschungszentrum Berlin, Berlin, Germany; [8]Children's Hospital, London Health Sciences Centre, Ontario, Canada; [9]Rheumatology Division & Oviedo University School of Medicine, Hospital Universitario Central de Asturias, Oviedo, Spain; [10]Norwich Medical School, University of East Anglia; [11]Department of Rheumatology, Norfolk and Norwich University Hospital, Norwich, UK; [12]Department of Public Health and Clinical Medicine/Rheumatology, Umeå University, Umea, Sweden; [13]Great North Children's Hospital, Newcastle Hospitals, Newcastle; [14]Centre for Adolescent Rheumatology, Department of Ageing, Rheumatology and Regenerative Medicine, Division of Medicine, University College London, London; [15]Alder Hey Children's Hospital, Liverpool; [16]Nuffield Department of Orthopaedics, Rheumatology and Musculoskeletal Sciences, University of Oxford, Oxford; [17]School of Health Sciences, Faculty of Health and Medical Sciences, University of Surrey, Surrey, UK
**Correspondence:** S. J. W. Shoop-Worrall
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**Introduction**
The heterogeneity of juvenile psoriatic arthritis (JPsA) implies biologically distinct disease subsets that could drive stratified treatment. A challenge in subgroup identification is multiple methods to identify JPsA.
**Objectives**
(i) To identify novel, phenotypically consistent and clinically distinct subgroups of children and young people with JPsA in early disease, (ii) To understand how subgroups differ across JPsA identification frameworks.
**Methods**
Children and young people were selected if enrolled to one of five multicentre inception/population cohorts of JIA: CAPS (UK), ICON (Germany), NPRD (Germany), ReACCh-Out (Canada) or CAPRI (Canada) between January 2001 and December 2023 with a physician's diagnosis of JPsA, enthesitis-related or undifferentiated JIA. Cohort follow-ups within 24 months of symptom onset were used to identify clusters of children with distinct cumulative disease burden within this time frame. Clustering was undertaken in those who (i) fulfilled ILAR criteria for JPsA, (ii) had a physician's diagnosis of JPsA (iii) fulfilled CASPAR criteria for PsA. Latent class analysis clustered children using an active joint count and the presence of psoriasis, dactylitis, and nail abnormalities. Univariable statistics compared outcomes within 24 months and both demographic and disease characteristics at first observation following symptom onset across clusters.
**Results**
In 3,282 children with a JPsA, ERA or uJIA diagnosis, symptom duration at first study documentation was median 11 months (IQR 3, 19). While 785 had a diagnosis of JPsA within two years of symptom onset, 332

fulfilled ILAR criteria for JPsA, and 449 fulfilled CASPAR criteria for PsA. In the 24 months following symptom onset, three JPsA clusters were consistently identified with significant differences across all outcomes: (i) *Low joint-Psoriasis*, (ii) *Psoriasis-Dactylitis* and (iii) *All Features*. These groups made up 26%, 40% and 24% of those fulfilling ILAR criteria for JPsA, and 59%, 11% and 30% of those with a JPsA diagnosis. For those fulfilling CASPAR criteria for PsA, 53% and 42% were assigned to *Psoriasis-Dactylitis* and *All Features* groups. However, a third group differed: *High joint-Dactylitis* (5%). A greater number of children in the Psoriasis-Dactylitis cluster had enthesitis (70-83%, versus 6-21%, $p<0.001$) and a positive family history of psoriasis (57-85% versus 24-46%, $p<0.001$). This group also experienced poorer patient/parent-reported outcomes (pain 4.6 vs. 2.6-3.0, disability 1.0 vs. 0.3-0.4; and wellbeing 4.6 vs. 2.6-3; all $p<0.01$) compared with other clusters. Those in the Low joint-Psoriasis cluster had lower inflammatory burden (physician global 2.0–3.0 cm versus 3.0–6.0 cm; ESR 10–12 mm/hr versus 14–20 mm/hr).

**Conclusion**

This large-scale international study reveals distinct JPsA clusters that could inform targeted treatment.

**Disclosure of interest**

None declared.

## PT005

### Treatment patterns in non-systemic juvenile idiopathic arthritis: a single-centre real-world cohort study

M. I. Gonzalez Fernandez[1,*], L. Lacruz Perez[2], M. Marti Masanet[2], I. Burgos Berjillos[2], B. Lopez Montesinos[3], I. Calvo Penades[1]

[1]Pediatric Rheumatology Research Group, Health Research Institute La Fe; [2]Pediatric Rheumatology Unit, Hospital Universitari i Politècnic La Fe, Health Research Institute La Fe; [3]Pediatrics, Campanar II Health Center, Valencia, Spain

**Correspondence:** M. I. Gonzalez Fernandez

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**Introduction**

Advances in pharmacological treatment have significantly improved outcomes in juvenile idiopathic arthritis (JIA); however, understanding real-world treatment patterns and sequences remains essential to optimise therapeutic strategies given the marked heterogeneity of JIA.

**Objectives**

To describe treatment patterns and sequences in non-systemic JIA, and to assess switching of targeted therapies in a real-world clinical setting.

**Methods**

Retrospective single-centre observational cohort study including consecutive patients with non-systemic JIA followed at a tertiary paediatric rheumatology unit, with ≥12 months of follow-up and exposure to disease-modifying anti-rheumatic drugs (DMARDs). Demographic and clinical data were collected from medical records. Treatment sequences, including conventional and targeted DMARDs, were analysed. Switching of targeted therapies was defined as the sequential use of ≥2 distinct targeted agents.

**Results**

A total of 228 patients (76.3% female) were included. Median age at diagnosis was 3.0 years (IQR 1.9–6.9), with median follow-up of 86.1 months (IQR 44.2–130.9). Most patients (92%) were diagnosed between 2011 and 2023. JIA categories at last follow-up were: oligoarthritis 54.8%, RF-negative polyarthritis 25.0%, RF-positive polyarthritis 0.9%, enthesitis-related arthritis 4.4%, juvenile psoriatic arthritis 7.9% and undifferentiated arthritis 7.0%. Conventional DMARDs were used in 227 (99.5%) patients, all receiving methotrexate. Targeted DMARDs were initiated in 141/228 (61.8%); the most frequently used first-line targeted agents were etanercept 85/141 (60.3%) and adalimumab 47/141 (33.3%). Among patients receiving targeted therapy, 100/141 (70.9%) had no switches, 29/141 (20.6%) had one switch and 12/141 (8.5%) underwent ≥2 switches. Treatment sequences were visualized using Sankey diagrams, revealing the diversity of treatment pathways in non-systemic JIA. Concomitant use of two targeted therapies was observed in 2/141 (1.4%) patients. In univariable analysis, younger age at diagnosis ($p=0.0358$), higher joint counts (both within the first 6 months [$p=0.0119$] and over the disease course [$p=0.0018$]), presence of dactylitis ($p=0.0096$), uveitis ($p=0.0099$), psoriasis ($p=0.0215$) and ILAR category ($p=0.0014$) were associated with increased number of targeted therapy switches (0, 1, ≥2).

**Conclusion**

In this single-centre real-world cohort of non-systemic JIA, treatment showed heterogeneous therapeutic trajectories over long-term follow-up. While most patients maintained the first targeted agent, a relevant subset required one or more switches. Switching was associated with clinical features suggestive of greater disease burden and distinct disease phenotypes. These findings highlight the importance of individualised therapeutic strategies in non-systemic JIA.

**Disclosure of interest**

M. I. Gonzalez Fernandez: None declared, L. Lacruz Perez Speaker Bureau with: Novartis, M. Marti Masanet: None declared, I. Burgos Berjillos: None declared, B. Lopez Montesinos: None declared, I. Calvo Penades Speaker Bureau with: Novartis, Sobi, Pfizer, GSK.

## PT006

### Calprotectin as a biomarker of disease activity in non-systemic juvenile idiopathic arthritis: JIA–COMPASS prospective international pilot study

M. Jelusic[1,*], M. Sestan[1], M. Sparchez[2], A. Loncar Vrancic[3], H. Musil[4], D. Turudic[1], M. Orhan Erkan[5], O. Basaran[5], B. Sener[6], A. Cochino[7], V. Bittermann[8], D. Misikova[9], T. Dallos[9], D. Rogic[3], J. Anton[10], S. Ozen[11]

[1]University Hospital Centre Zagreb, Department of Paediatrics, University of Zagreb, School of Medicine, Zagreb, Croatia; [2]Iuliu Hatieganu University of Medicine and Pharmacy, Cluj-Napoca, Romania; [3]University Hospital Centre Zagreb, Department of Laboratory Diagnostics; [4]University of Zagreb, School of Medicine, Zagreb, Croatia; [5]Department of Paediatrics, Division of Rheumatology; [6]Department of Infectious Diseases and Clinical Microbiology, Hacettepe University Faculty of Medicine, Ankara, Türkiye; [7]Department of Clinical Immunology, National Institute for Mother and Child Health Alessandrescu-Rusescu, University of Medicine and Pharmacy "Carol Davila", Bucharest, Romania; [8]Department of Pediatric Rheumatology, SJD Barcelona Children's Hospital Passeig Sant Joan de Déu, Barcelona, Spain; [9]2nd Department of Paediatrics, National Institute of Children's Diseases, Comenius University Medical School in Bratislava, Bratislava, Slovakia; [10]Department of Paediatrics, Division of Rheumatology, SJD Barcelona Children's Hospital Passeig Sant Joan de Déu, Barcelona, Spain; [11]Hacettepe University Faculty of Medicine, Department of Paediatrics, Division of Rheumatology, Ankara, Türkiye

**Correspondence:** M. Jelusic

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**Introduction**

Calprotectin is a promising biomarker of innate immune activation in systemic juvenile idiopathic arthritis (JIA), but prospective real-world data in non-systemic JIA are limited.

**Objectives**

To evaluate the association of calprotectin with disease activity in non-systemic JIA and explore its utility in differentiating treatment and remission cohorts.

**Methods**

This international multicenter prospective pilot study included patients with non-systemic JIA divided into a Treatment cohort (diagnosis/start of treatment) and a Remission cohort (treatment tapering/withdrawal). Patients were followed at tertiary pediatric rheumatology centers with assessments every 3 months. Disease activity was evaluated using JADAS-27 ESR and JADAS-27 CRP. Calprotectin was measured using the automated Gentian Calprotectin Immunoassay (GCAL®), an immunoturbidimetric assay intended for use in everyday clinical practice. Correlations with clinical and laboratory disease activity measures were analyzed using Spearman's coefficient, and exploratory ROC analyses assessed differentiation between treatment and remission cohorts.

**Results**

A total of 323 visits from 128 patients were analyzed (M: F ratio 1:2.76; mean age at diagnosis 7.5±5.37 years). Median calprotectin concentrations were higher in the Treatment cohort (1.21 [0.72–2.02] vs. 0.84 [0.49–1.31] µg/mL). Calprotectin significantly correlated with JADAS-27ESR ($\rho=0.357$,$p<0.001$), JADAS-27CRP ($\rho\approx0.36$,$p<0.001$), active joint count ($\rho=0.321$,$p<0.001$), PGA ($\rho=0.313$,$p<0.001$), PtGA/PaGA ($\rho=0.285$,$p<0.001$), ESR ($\rho=0.212$,$p<0.001$). Correlations were stronger

in the Treatment cohort compared with the Remission cohort: JADAS-27ESR (ρ=0.447 vs. 0.287), JADAS-27CRP (ρ≈0.45 vs. ≈0.29), active joint count (ρ=0.401 vs. 0.191), PGA (ρ=0.392 vs. 0.248), and PtGA/PaGA (ρ=0.341 vs. 0.214; all $p$<0.01). Calprotectin reflected inflammatory disease activity, demonstrating progressively higher concentrations across JADAS-27-defined disease activity states: inactive 0.0 (0.0–0.0), low 0.0 (0.0–0.84), moderate 0.46 (0.0–1.00), and high disease activity 1.46 (0.94–2.42) µg/mL. ROC analysis showed moderate discrimination between treatment and remission cohorts (AUC 0.624). A calprotectin cut-off of 1.89 µg/mL achieved 91.9% specificity.

**Conclusion**

Calprotectin concentrations were higher in the treatment cohort than in the remission cohort and increased progressively across JADAS-defined disease activity states, supporting its role as a biomarker of active inflammation in non-systemic JIA. Exploratory ROC analyses suggest potential utility of calprotectin for differentiation between clinically active and remission states. Extended prospective follow-up of this cohort is currently in progress. Support: NPOO 10106-25-2896(JIA-BARRIER-MARKERS), funded by EU-NextGenerationEU and Gentian AS.

**Disclosure of interest**

None declared.

## PT007

### Increased gut-derived LPS bioactivity in juvenile idiopathic arthritis

L. Lima-Antoine[1], B. Graziela Postal[1], J. Touneur-Marsille[1], B. Chassaing[2], C. Durand[3], L. A. Eveillard[3], M. Labouret[3], M. Roy[4], É. Viennois[5], U. Meinzer[5,*]
[1]Université Paris Cité, INSERM, Centre de Recherche sur l'inflammation, UMR 1149, F-75018, Paris; [2]Institut Pasteur; [3]Hôpital Robert-Debré, Paris; [4]Université Paris Cité, INSERM, Centre de Recherche sur l'inflammation, UMR 1149, Paris; [5]Université Paris Cité, INSERM, Centre de Recherche sur l'inflammation, UMR 1149, Paris, France
**Correspondence:** U. Meinzer
*Pediatric Rheumatology 2026*, **24(S1):**PT007

**Introduction**

Juvenile idiopathic arthritis (JIA) is a chronic inflammatory disease of unknown aetiology in which increasing evidence implicates the intestinal microbiota. Altered microbial composition and intestinal barrier dysfunction have been associated with immune activation in JIA, although the contribution of specific microbial products remains incompletely understood. Lipopolysaccharide (LPS), a potent pro-inflammatory component of Gram-negative bacteria, may promote chronic inflammation through activation of innate immune pathways and could contribute to joint inflammation.

**Objectives**

To investigate gut-derived LPS bioactivity in children with JIA and to explore its potential role in arthritis development using a spontaneous arthritis mouse model.

**Methods**

Faecal and serum LPS bioactivity, LPS-binding protein (LBP), and anti-LPS antibodies were quantified in children with JIA and healthy controls using TLR4 reporter assays and immunoassays. Mechanistic studies were performed in IL-1Ra$^{-/-}$ spontaneous arthritis mice, including antibiotic-induced dysbiosis and human-to-mouse faecal microbiota transplantation (FMT). Arthritis severity, intestinal inflammation, circulating LPS bioactivity, and microbiota composition were assessed longitudinally. Ex vivo stimulation assays were used to evaluate the inflammatory potential of faecal extracts.

**Results**

Children with JIA exhibited significantly increased faecal and serum LPS bioactivity, elevated circulating LBP levels, and reduced anti-LPS antibodies compared with healthy controls, suggesting an altered host response to gut-derived endotoxin. In IL-1Ra$^{-/-}$ mice, increased intestinal and systemic LPS bioactivity was observed during arthritis development. Antibiotic-induced dysbiosis increased intestinal LPS bioactivity and was associated with more severe arthritis, whereas FMT from low-LPS donors partially attenuated disease severity. Faecal extracts with high LPS bioactivity induced robust pro-inflammatory cytokine production ex vivo. Longitudinal microbiota analyses demonstrated that expansion of Gram-negative bacteria preceded clinical arthritis onset.

**Conclusion**

Increased gut-derived LPS bioactivity is associated with JIA and may contribute to systemic inflammation and arthritis progression. These findings support a role for host–microbiota interactions in paediatric inflammatory arthritis and suggest that modulation of intestinal microbial activity may represent a future therapeutic strategy.

**Trial registration identifying number**

NCT03092427; NCT04460144.

**Disclosure of interest**

None declared.

## PT008

### Dissecting the inflammatory signature of enthesitis-related arthritis: role of HLA-B27 and the IL-23/IL-17 axis

G. Olesea[1,2,*], R. Ninel[1,2], F. S. Gabriela[1,2], G. Olga[1,2]
[1]Pediatrics, SUMPh Nicolae Testemitanu; [2]Rheumatology, Institute of Mother and Child, Chisinau, Moldova, Republic of
**Correspondence:** G. Olesea
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**Introduction**

Enthesitis-Related Arthritis (ERA), a distinct ILAR subtype, is a juvenile spondyloarthritis characterized by a strong association with the HLA-B27 antigen and dysregulation of the IL-23/IL-17 axis. Clinically, ERA often leads to axial involvement, early disability, and resistance to conventional synthetic DMARDs (csDMARDs). Within the 'Treat-to-Target' paradigm, investigating these pathogenetic correlates is vital to optimize outcomes and prevent long-term functional impairment.

**Objectives**

To evaluate the genetic profile (HLA-B27) and specific pro-inflammatory cytokine pathways—specifically the IL-23/IL-17 axis and TNF-alpha— and correlate these biomarkers with clinical, functional, and imaging data in pediatric ERA.

**Methods**

This cross-sectional study included 40 pediatric patients (24 boys, 16 girls; mean age 14.47±2,3 years) diagnosed via ILAR criteria. Assessment utilized VAS for pain and JSpADA for disease activity. Laboratory analysis included HLA-B27 and ANA status, alongside a detailed evaluation of the IL-23/IL-17 and TNF alpha cytokine axis. Axial involvement was assessed using Magnetic Resonance Imaging (MRI).

**Results**

Patients were stratified into two cohorts: 12 (30%) HLA-B27 positive and 28 (70%) HLA-B27 negative. Clinical and immunological analyses revealed significant disparities. JSpADA score, was significantly higher in the HLA-B27 positive group compared to the negative group (4.81±1.48 vs.2.63±1.28; $p$ < 0.001). This increased clinical severity correlated with a robust activation of the IL-23/IL-17 axis; HLA-B27 positive patients exhibited markedly higher levels of IL-23 (94.3± 8.2 vs.28.3±4.1 pg/mL; $p$ < 0.0001) and TNF-alpha (35.6±4.5vs.11.3±2.8 pg/mL; $p$ < 0.001). In contrast, the HLA-B27 negative cohort showed a higher prevalence of ANA positivity (46.4% vs. 25%; $p$ = 0.045). Furthermore, MRI findings demonstrated more pronounced axial involvement and structural changes in the HLA-B27 positive group, underscoring the aggressive nature of the IL-23-driven phenotype in these patients. These patients also required, bDMARD, DMARDs and glucocorticoids more frequently (OR = 13.2; $p$ = 0.004), reflecting more severe disease and more difficult therapeutic control.

**Conclusion**

HLA-B27 positivity identifies a specific autoinflammatory phenotype in ERA driven by the IL-23/IL-17 axis. These findings suggest that HLA-B27 status is a critical prognostic indicator, highlighting the need for early targeted biological interventions in positive patients to mitigate high disease activity and irreversible structural damage.

**Disclosure of interest**

None declared.

## PT009

### Clinical and prognostic relevance of sacroiliac MRI findings in children with enthesitis-related arthritis: inter-reader reliability and longitudinal imaging patterns

M. T. Karadoğan[1,*], E. Ayaz[2], N. G. Akyel[2], S. Doğantan[1], T. Güzel[1], B. Çinpolat[1], Z. Hazıroğlu Ökmen[1], E. E. Fırat[1], F. G. Demirkan[3], F. Çakmak[1]
[1]Department of Pediatric Rheumatology; [2]Department of Pediatric Radiology, Başakşehir Çam and Sakura City Hospital; [3]Department of Pediatric Rheumatology, Kanuni Sultan Süleyman Training and Research Hospital, İstanbul, Türkiye

**Correspondence:** M. T. Karadoğan
*Pediatric Rheumatology 2026*, **24(S1):**PT009

**Introduction**
Sacroiliac involvement is central to enthesitis-related arthritis (ERA), yet clinical assessment and inflammatory markers may underestimate inflammatory and structural disease.
**Objectives**
To evaluate sacroiliac magnetic resonance imaging (SI-MRI) findings in children with ERA, focusing on inter-reader reliability, clinicoradiologic discordance and longitudinal imaging change.
**Methods**
This retrospective cohort included 45 children with ERA who underwent SI-MRI. Clinical, laboratory, treatment and outcome data were extracted from records. Each MRI was scored independently by two blinded paediatric radiologists for bone marrow oedema (BME)/osteitis, synovitis, capsulitis, enthesitis and structural lesions. Reliability was assessed with Cohen's kappa and intraclass correlation coefficient (ICC). Associations and longitudinal MRI changes were analysed using non-parametric tests.
**Results**
Twenty-six patients (57.8%) were male. Median age at diagnosis was 13.0 years and median disease duration at baseline MRI was 1.8 years. HLA-B27 was positive in 7/39 (17.9%). At baseline, any-reader positivity was 82.2% for BME/osteitis, 86.7% for any active sacroiliac inflammation and 75.6% for any structural lesion; erosions and sclerosis were detected in 46.7% and 66.7%, respectively. Agreement was substantial for synovitis (κ=0.733), BME/osteitis (κ=0.685) and erosions (κ=0.694), and moderate for sclerosis (κ=0.599). ICC for maximum BME depth was 0.749, whereas agreement for BME quadrant extent was lower (κ=0.329). Structural lesions were associated with older age at MRI (15.2 vs. 12.0 years, $p$=0.043). Deep BME (>10 mm) was associated with HLA-B27 positivity ($p$=0.008) and erosions ($p$=0.0035); BME involving ≥3 quadrants was also associated with erosions ($p$=0.017). CRP, ESR, JADAS-27 and visual analogue scores showed no consistent correlation with BME depth or extent. Fourteen patients had repeat MRI after a median of 20.1 months. BME depth decreased significantly (median change −2.5 mm, $p$=0.0069), as did BME quadrant burden (median change −0.8, $p$=0.0348), while structural lesions persisted.
**Conclusion**
SI-MRI showed a high burden of active and structural sacroiliac disease in children with ERA, often discordant with clinical and laboratory measures. Blinded paediatric radiology scoring was reliable for key lesions, but semi-quantitative BME extent required caution. Deep/extensive BME clustered with erosive damage, and longitudinal imaging suggested regression of inflammation with persistence of structural abnormalities, supporting SI-MRI for risk stratification and follow-up in paediatric ERA.

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**Disclosure of interest**
None declared.

## PT010

### Recurrence risk during systemic therapy tapering in juvenile idiopathic arthritis-associated uveitis

G. B. Beretta[1,*], A. Losi[1], M. Nassisi[2,3], S. Mazzitelli[1,2], A. Spagni[2], S. Lanni[1], G. Leone[3], C. Mapelli[3], F. S. Minoia[1]
[1]Pediatric Immuno-Rheumatology Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico; [2]University of Milan; [3]Ophthalmology Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy
**Correspondence:** G. B. Beretta
*Pediatric Rheumatology 2026*, **24(S1):**PT010

**Introduction**
Juvenile Idiopathic Arthritis-associated uveitis (JIA-U) relapse after systemic therapy tapering is common, while evidence regarding the optimal timing and strategy of treatment de-escalation remains limited[1].
**Objectives**
To evaluate relapse rates after tapering of systemic therapy in JIA-U and to identify predictors of flare during treatment de-escalation.
**Methods**
Clinical records of patients with JIA-U undergoing tapering of systemic therapy with Methotrexate (MTX) and/or Adalimumab (ADA) followed at a single tertiary Pediatric Rheumatology center were retrospectively reviewed. Demographic, clinical and treatment-related variables associated with tapering outcomes were analyzed.
**Results**
44 patients with JIA-uveitis undergoing a first systemic therapy tapering attempt were included, accounting for 55 tapering episodes. At tapering initiation, 30 patients were receiving combination therapy with Methotrexate and a biologic agent (28/30 ADA), 11 MTX monotherapy, and 5 ADA monotherapy. Patients were predominantly female (75%), ANA-positive (43/44), and affected by oligoarticular arthritis (65.9%). Median age at first tapering was 9.46 years, with a median follow-up of 3.85 years after treatment tapering. Overall, 38 MTX and 17 ADA tapering episodes were analyzed. In patients receiving combination therapy, MTX was tapered before ADA in all but one case. Median time from last uveitis flare to tapering initiation was 1.73 years, without significant differences between patients with or without relapse. Among MTX tapering episodes, 17 (44.7%) resulted in relapse (7 ocular and 7 articular flares). Flare frequency was lower in patients receiving concomitant biologic therapy compared with MTX monotherapy (37.9% vs. 67.7%). Patients with successful MTX discontinuation had started systemic therapy earlier after disease onset compared with those with relapse (median 0.47 vs. 1.1 years). In 5/17 relapses (29.4%), flare occurred after complete MTX withdrawal. Among ADA tapering episodes, 14/17 resulted in relapse (10 ocular and 9 articular flares), with 9/14 occurring after complete ADA discontinuation. Following relapse, 28/31 patients regained disease control after reintroduction of full-dose therapy, while only 3 required therapeutic step-up. After a first successful tapering attempt, 9 patients underwent a second treatment de-escalation, with relapse occurring in 6/9 cases. Ocular damage progression after tapering was observed in 5/44 patients (11.4%).
**Conclusion**
Relapse after systemic therapy tapering in JIA-U remains frequent. Earlier systemic treatment initiation may improve the likelihood of successful treatment withdrawal, suggesting a therapeutic window in JIA-U.

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**Disclosure of interest**
None declared.

## PT011

### A biomarker panel with IL-18 and IL8 discriminates between sJIA-SD from and pediatric malignancies

H. I. Abu-Tawil[1,*], A. Leek[2], M. mizuta [3], J. de Kanter[4], R. Scholman[1], V. de Haas[4], F. Meyer-Wentrup[4], L. Tytgat[4], S. Nierkens[1], J. Drylewicz[1], E. Delemarre[1], J. van Loosdregt[1], S. J. Vastert[1,2]
[1]Center for Translational Immunology; [2]Pediatric Rheumatology, University Medical Center Utrecht, Utrecht, Netherlands; [3]Pediatric Rheumatology, Prefectural Kobe Children's Hospital, Kobe, Japan; [4]Pediatric Oncology, Prinses Máxima Centrum, Utrecht, Netherlands
**Correspondence:** H. I. Abu-Tawil
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**Introduction**
Systemic juvenile idiopathic arthritis (sJIA/Still's disease) can closely mimic pediatric malignancies at presentation, particularly when arthritis is absent early after onset. Because delayed or incorrect diagnosis may lead to unnecessary invasive workup and postpone appropriate therapy, clinically actionable biomarkers are needed to support early discrimination.

**Objectives**
This study aimed to evaluate the performance of IL-18 discriminating between SD and childhood malignancy and to possibly create a panel of multiple biomarkers with an augmented performance, establishing cut-off values to be used in clinical practice. This could expedite SD diagnosis, avoid unnecessary (expensive) diagnostic procedures and diminish doctor's delay in initiating treatment for SD and childhood malignancy.
**Methods**
We retrospectively assembled a cohort of patients with sJIA ($n$=34) and pediatric malignancies with overlapping clinical characteristics at onset (total $n$=90: precursor B-ALL $n$=25, T-ALL $n$=14, AML $n$=12, Hodgkin lymphoma $n$=20, non-Hodgkin lymphoma $n$=4, neuroblastoma $n$=15). Samples obtained at first outpatient presentation (serum or sodium-heparin plasma) were analyzed using a customized 17-analyte Luminex multiplex panel. In addition, the same samples were profiled with the Olink Target 96 Immuno-Oncology panel as a discovery approach. Differential protein signals were explored using volcano/boxplot-based comparisons. Diagnostic performance was evaluated using ROC analyses with cut-off values derived via Youden's index. A multivariable model was constructed using stepwise selection.
**Results**
Discovery profiling identified IL-18 as strongly elevated in sJIA compared with malignancy, with an inverse signal for IL-8 as an additional discriminating feature. Luminex measurements confirmed these findings and supported high diagnostic performance for IL-18 alone and for a combined biomarker approach. A model combining IL-18 and inverse IL-8 achieved excellent discrimination between sJIA and malignancy (AUC 0.99), with clinically interpretable cut-offs (including IL-18 ≥1672 and IL-8 <20).
**Conclusion**
A biomarker panel centered on IL-18 and inverse IL-8 shows strong potential to distinguish sJIA from pediatric malignancies at disease onset. Prospective and multi-center validation and standardization of laboratory implementation are warranted before routine clinical adoption.
**Disclosure of interest**
None declared.

## PT012

### Efficacy of arumakimig, a bispecific IL-1β and IL-18 neutralizing antibody, in refractory Still's disease with recurrent macrophage activation syndrome and lung involvement

M. Trevisan[1,2,*], A. Ippoliti[1], M. Pardeo[1], A. De Matteis[1], L. Menchini[3], M. L. Mennini[3], A. M. Greco[4], I. Caiello[5], G. Prencipe[5], E. Marchettini[6], S. Ghirardo[6,7], A. Taddio[6,7], F. De Benedetti[1,5], C. Bracaglia[1]
[1]Division of Rheumatology, IRCCS Ospedale Pediatrico Bambino Gesù; [2]Department of Systems Medicine, University of Rome "Tor Vergata"; [3]Department of Radiology and Bioimaging; [4]Division of Hospital Pharmacy; [5]Laboratory of Immuno-Rheumatology, IRCCS Ospedale Pediatrico Bambino Gesù, Rome; [6]Department of Medicine, Surgery, and Health Sciences, University of Trieste; [7]Institute for Maternal and Child Health, IRCCS "Burlo Garofolo", Trieste, Italy
**Correspondence:** M. Trevisan
*Pediatric Rheumatology 2026*, **24(S1):**PT012

**Introduction**
Still's disease (SD) is a systemic inflammatory disorder in which some patients develop refractory disease, occasionally associated with lung involvement, requiring prolonged immunosuppressive treatments. Arumakimig, a bispecific monoclonal antibody targeting IL-1β and IL-18, may represent a promising treatment option for refractory SD, particularly in patients with recurrent macrophage activation syndrome (MAS) and lung disease (LD).
**Objectives**
To report the efficacy and safety of Arumakimig in patients with refractory SD-associated LD (SD-LD).
**Methods**
We retrospectively collected data on 5 patients with SD-LD from two tertiary pediatric hospitals who were treated with Arumakimig for at least 12 months. Clinical, laboratory, and radiological findings were analyzed before and after initiation of Arumakimig.
**Results**
Five patients with SD-LD were enrolled (3/5 female), with a median age at disease onset of 0.8 years (IQR 0.4–2.5). All patients presented an atypical urticarial rash at onset, and 3/5 had a history of MAS. LD was diagnosed at a median age of 1.9 years, and 2 patients developed LD without prior exposure to IL-1 or IL-6 inhibitors. Two out of four patients carrying the HLA-DRB1*15 allele experienced adverse reactions to tocilizumab, resulting in treatment discontinuation. One patient had severe LD requiring oxygen supplementation, with bronchoalveolar lavage suggestive of alveolar proteinosis. Except for two patients with pathological overnight pulse oximetry, pulmonary function tests were unremarkable in the remaining patients. High-resolution chest CT findings were consistent with SD-LD in all cases, mainly showing septal/peribronchial thickening and ground-glass opacities. Arumakimig was initiated after a median of 41.3 months (IQR 14.0–52.9) from SD onset. After 12 months of treatment, 3/5 patients showed clinical and laboratory improvement, with no further MAS episodes. IL-18 and CXCL9 levels markedly decreased during treatment (median IL-18 from 60,568 to 20,347 pg/mL; median CXCL9 from 1,856 to 200 pg/mL). LD improved in 3 patients and worsened in 2, although they reduced oxygen dependence and glucocorticoid need. Overall treatment burden decreased after treatment initiation, particularly glucocorticoid use. Two patients discontinued Arumakimig due to refractory arthritis and nonspecific colitis, respectively. No infusion-related reactions or major safety concerns were observed, and no deaths occurred after a median 5-year follow-up from LD onset.
**Conclusion**
Our cases suggest that simultaneous neutralization of IL-1β and IL-18 with Arumakimig may represent a promising therapeutic approach in refractory SD, improving lung involvement, MAS flares, and glucocorticoid need.

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**Disclosure of interest**
M. Trevisan: None declared, A. Ippoliti: None declared, M. Pardeo Consultant with: Sobi, Novartis, A. De Matteis: None declared, L. Menchini: None declared, M. Mennini: None declared, A. Greco: None declared, I. Caiello: None declared, G. Prencipe: None declared, E. Marchettini: None declared, S. Ghirardo: None declared, A. Taddio: None declared, F. De Benedetti Consultant with: Abbvie, SOBI, Novimmune, Novartis, Roche, Pfizer, C. Bracaglia Consultant with: Sobi, Novartis.

## PT013

### Age-dependent diagnostic value of IL-18 and phagocyte-specific S100 proteins in Still's disease

T. Thalheim[1,*], E. L. Verweyen[1], C. Girard-Guyonvarc'h[2,3], M. Saers[1], S. Schleifenbaum[1], S. Fühner[1], Y. M. Mueller[4], C. Park[1], H. Wittkowski[1], C. Hinze[1], P. D. Katsikis[4], C. Gabay[2,3], C. Kessel[1], D. Foell[1]
[1]Department of Pediatric Rheumatology and Immunology, University Children's Hospital, Muenster, Germany; [2]Department of Medicine, University Hospitals; [3]Department of Pathology and Immunology, University of Geneva, Geneva, Switzerland; [4]Department of Immunology, Erasmus MC, University Medical Center, Rotterdam, Netherlands
**Correspondence:** T. Thalheim
*Pediatric Rheumatology 2026*, **24(S1):**PT013

**Introduction**
Still's disease (SD) is a rare systemic autoinflammatory disease (SAID) with pediatric or adult onset. Recently, the EULAR/PReS guidelines for the diagnosis and management of SD recommended using phagocyte-specific S100 proteins (S100A8/A9, S100A12) and interleukin-18 (IL-18) as diagnostic biomarkers (1). However, few studies have directly compared their performance across a broader age range.
**Objectives**
Here, we evaluate the ability of S100 proteins and IL-18 to differentiate pediatric SD and adult SD from other SAIDs.
**Methods**
Using identical biomarker assay platforms, we analyzed serum levels of S100A8/A9, S100A12, and IL-18 in 307 patients with active SAIDs and

49 healthy controls. This included 78 pediatric and 44 adult SD patients from the European ImmunAID and a multicenter German cohort.

**Results**

S100 protein and IL-18 levels were elevated in SD when compared to other SAID patients and healthy controls. In SD patients, S100 protein serum levels were positively correlated with established inflammatory mediators, including ferritin and IL-6 blood levels, as well as peripheral blood leukocyte counts. IL-18 serum levels were associated with blood ferritin concentration but revealed only weak or no correlation with S100 protein levels in pediatric or adult SD patients, respectively. Longitudinal analysis demonstrated a significant decrease in S100 protein levels in SD patients under treatment ($P$<0.01). Interestingly, S100A8/A9 and S100A12 serum levels were significantly higher in pediatric (median=71,675ng/mL and 1,240ng/mL) compared to adult SD patients (median=24,237ng/mL and 310.5ng/mL; $P$<0.01). These findings were supported by negative correlations between S100 protein levels and patient age ($r$=-0.37 and $r$=-0.42). ROC analyses demonstrated that S100A8/A9 and S100A12 serum levels provided the best diagnostic discrimination in pediatric SD (AUC 0.88 and 0.89), whereas their performance in adult SD was limited (AUC 0.72 and 0.71). In contrast, IL-18 demonstrated strong diagnostic performance in adult (AUC 0.91) as well as pediatric SD (AUC 0.81).

**Conclusion**

This study confirms that S100 protein and IL-18 levels are elevated in SD compared to other SAIDs. However, the expression of S100 proteins differs between pediatric and adult SD patients. While these findings do not challenge the concept that pediatric and adult SD represent a single disease spectrum (1), our data strongly argue for age-adapted use of S100 proteins and respective cut-offs to optimize diagnostic accuracy and disease management.

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**Disclosure of interest**

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## PT014

### Transcriptomic profiling of CD38$^+$HLA-DR$^+$CD8$^+$ T cells in Still's disease and macrophage activation syndrome

G. Rogani[1,2,*], A. Bodelon[1], J. van Loosdregt[1], S. J. Vastert[1,2]
[1]Center for Translational Immunology; [2]Pediatric Rheumatology, University Medical Center Utrecht, Utrecht, Netherlands
**Correspondence:** G. Rogani
*Pediatric Rheumatology 2026*, **24(S1):**PT014

**Introduction**

CD38$^+$HLA-DR$^+$CD8$^+$ T cells (double-positive, DP-CD8$^+$) are markedly expanded in macrophage activation syndrome (MAS), where they may represent an important source of interferon-γ (IFN-γ)[1-3]. We have previously shown that these cells are already present in Still's disease (SD), including at disease onset. However, their biological profile remains incompletely defined.

**Objectives**

To define the transcriptomic landscape of DP-CD8$^+$ cells in SD and SD-associated MAS.

**Methods**

DP-CD8$^+$ and double-negative (DN; CD38$^-$HLA-DR$^-$) CD8$^+$ T cells were sorted from peripheral blood of patients with therapy-naïve new-onset SD and SD-MAS and subjected to bulk RNA sequencing. Differential expression analysis was performed using DESeq2. Functional enrichment analyses were conducted using g: Profiler and Gene Set Enrichment Analysis.

**Results**

Regardless of whether cells were derived from patients with new-onset SD or MAS, DP-CD8$^+$ cells displayed a distinct transcriptional program compared with DN-CD8$^+$ cells. This program was characterized by increased expression of genes involved in proliferation (MKI67), activation and checkpoint regulation (PDCD1, LAG3, TIGIT, CTLA4, FASLG), and cytotoxicity (GZMA, GZMB, GZMH, GZMK, PRF1). IFNG transcripts were upregulated, supporting the role of these cells as a potential source of IFN-γ. In contrast, IL18BP, encoding the natural antagonist of IL-18, was significantly downregulated. Pathway enrichment analysis confirmed overrepresentation of cell-cycle, DNA replication, lymphocyte activation, and cytotoxicity pathways. Unsupervised clustering based on proliferation- and cytotoxicity-related genes clearly separated DP- from DN-CD8$^+$ subsets. mTORC1 signaling genes were also more expressed in DP.

**Conclusion**

DP-CD8$^+$ cells in SD and MAS exhibit a hyperproliferative, highly activated, and cytotoxic transcriptional profile characterized by increased IFNG and reduced IL18BP expression. These findings support a role for this subset in amplifying IL-18- and IFN-γ-driven inflammation and identify DP-CD8$^+$ cells as a mechanistic link between SD and MAS and a candidate therapeutic target.

**References**

1. PMID: 37751296.
2. PMID: 37517575.
3. PMID: 35500103.

**Disclosure of interest**

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## PT015

### Macrophage activation syndrome in children: predictors of lethal outcome

S. Kurbanova[1,*], S. Valieva[1], O. Lomakina[1], V. Yudina[1]
[1]Morozov City Children's Clinical Hospita, Moscow, Russian Federation
**Correspondence:** S. Kurbanova
*Pediatric Rheumatology 2026*, **24(S1):**PT015

**Introduction**

Hemophagocytic lymphohistiocytosis (HLH) is a life-threatening hyperinflammatory syndrome resulting from pathological activation of T and NK cells, triggered by different stimuli. Early identification of patients at risk for fatal outcome remains challenging in HLH, especially in pediatric population.

**Objectives**

To analyze clinical and laboratory predictors of lethal outcome in children with HLH managed in a Morozovskaya children's city hospital (Moscow) between 2019 and 2025.

**Methods**

A retrospective analysis included 127 pediatric HLH patients (76 female, 51 male; median age – 8.9 years), including 24 cases with lethal outcome. Statistical analysis was performed using StatTech v. 4.12.7. Quantitative data were compared using Mann-Whitney U test or t-test, categorical data using Fisher's exact test, $p$<0.05 was considered significant.

**Results**

Overall, in-hospital mortality was 18.9% (24/127). Fatal cases differed significantly from survivors by higher frequency of underlying malignancies (30.4% vs. 4.5%, $p$=0.001), primary immunodeficiencies (28.6% vs. 4.7%, $p$=0.004), and infections (81.0% vs. 33.3%, $p$<0.001). Presence of rheumatologic disease was less common in fatal group (13.6% vs. 64.3%, $p$<0.001). Clinically, fatal outcome was associated with pleuritis (61.9% vs. 31.4%, $p$=0.013), hydroperitoneum (71.4% vs. 34.9%, $p$=0.003), CNS involvement (76.2% vs. 15.7%, $p$<0.001), and absence of arthritis (95.2% vs. 58.1%, $p$<0.001). Laboratory predictors at first day included thrombo-

cytopenia (88.2% vs. 48.1%, $p$=0.003), cytopenia of ≥2 lineages (93.3% vs. 55.0%, $p$=0.008), hypofibrinogenemia (62.5% vs. 32.4%, $p$=0.044), elevated procalcitonin (100% vs. 64.4%, $p$=0.012), and prolonged aPTT (median 41.1 vs. 30.1 s, $p$<0.001). At follow-up, persistent leukopenia (73.3% vs. 32.9%, $p$=0.007), neutropenia (69.2% vs. 32.0%, $p$=0.014), thrombocytopenia (100% vs. 39.0%, $p$<0.001), prolonged activated partial thromboplastin time (44.3 vs. 27.7 s, $p$=0.008), and prolonged prothrombin time (17.8 vs. 12.9 s, $p$=0.002) were significantly associated with mortality. In 66 patients with underlying rheumatic disease (macrophage activation syndrome (MAS) subgroup) 3 died (4.5%), and lethal outcome was associated with older age at symptom onset (15.3 vs. 9.2 years, $p$=0.028), higher triglyceride levels (4.46 vs. 1.95 mmol/L, $p$=0.016), and CNS involvement (100% vs. 10.0%, $p$=0.002).

**Conclusion**

Fatal outcome in pediatric HLH is predicted by underlying malignancies, primary immunodeficiencies, severe cytopenias, coagulopathy, and CNS involvement. Among MAS patients older age, hypertriglyceridemia and CNS involvement are key risk factors. These findings may help risk stratification and intensification of therapy in high-risk patients.

**Disclosure of interest**

None declared.

## PT016

### Limited prognostic value of plasma calprotectin (S100A8/A9) in patients with Still's disease treated with interleukin-1 inhibitors

M. Trevisan[1,2,*], G. Ferri[3], E. Loricchio[3], V. Matteo[3], I. Caiello[3], A. De Matteis[1], M. Pardeo[1], C. Bracaglia[1], F. De Benedetti[1,3], G. Prencipe[3]
[1]Division of Rheumatology, IRCCS Ospedale Pediatrico Bambino Gesù; [2]Department of Systems Medicine, University of Rome "Tor Vergata"; [3]Laboratory of Immuno-Rheumatology, IRCCS Ospedale Pediatrico Bambino Gesù, Rome, Italy
**Correspondence:** M. Trevisan
*Pediatric Rheumatology 2026*, **24(S1):**PT016

**Introduction**

Still's disease (SD) is a rare systemic inflammatory disease characterized by high-spiking fever, rash, arthritis, and/or hepatosplenomegaly. About 20% of patients develop macrophage activation syndrome (MAS), a life-threatening complication. We recently demonstrated the prognostic role of IL-18 levels in the early phases of SD for predicting disease activity, course, and MAS, whereas the prognostic value of serum calprotectin remains unclear.

**Objectives**

To evaluate the prognostic value of plasma S100A8/A9 levels at baseline and after 3 months of interleukin-1 inhibitor (IL-1i) therapy in predicting disease activity, disease course, and MAS in SD.

**Methods**

We retrospectively analyzed 66 biologic-naïve SD patients treated with IL-1i for ≥12 months. Demographic, clinical, and laboratory data were collected at baseline (before IL-1i initiation) and after 3 months (T3) of treatment. Plasma S100A8/A9 levels were measured by ELISA.

**Results**

At baseline, median S100A8/9 levels were 28,093 ng/ml (IQR 11,304-63,027 ng/ml), with no significant differences between patients with or without MAS ($p$=0.96). After three months of IL-1i, S100A8/A9 values significantly decreased (T0 median value 28,093 pg/ml to T3 median value 639 ng/ml, $p$ <0.0001). At T3, patients with AD showed similar calprotectin levels to patients with clinically inactive disease (CID) (AD 789 ng/ml vs. CID 557 ng/ml, $p$=0.29). S100A8/A9 levels showed a modest correlation with IL-18 both at baseline ($r$=0.25, $p$=0.04) and at T3 ($r$=0.33, $p$=0.008). Baseline S100A8/A9 levels did not discriminate patients by disease activity at 12 months, disease course (monocyclic/polycyclic vs. persistent), or subsequent MAS development. After three months of treatment, S100A8/A9 levels were significantly higher in patients who developed a persistent disease course compared to those with mono-/poly-cyclic course (1.671 vs. 555 ng/ml, $p$=0.01). A threshold >1,000 ng/mL at 3 months predicted persistent disease with moderate accuracy (area under the curve 0.73; CI 95% 0.58-0.89; $p$=0.01; Sensitivity 75%; Specificity 69%). However, in multivariate analysis, only IL-18 levels higher than 15,000 pg/ml (OR=53, $p$=0.01) and delayed IL-1i initiation (>3 months) independently predicted a persistent disease course (AUC 0.96, $p$<0.0001; Tjur's $R^2$ 0.70).

**Conclusion**

In SD, S100A8/A9 levels markedly decrease after IL1 inhibition, independently of disease activity, and do not predict disease course or MAS development. Although higher calprotectin levels at 3 months are associated with a persistent disease course, their independent prognostic value is limited. Conversely, IL-18 remains a robust biomarker for disease stratification and therapeutic decision-making.

**References**

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**Disclosure of interest**

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## PT017

### Spatial transcriptomics reveals divergent coagulation and immune programs in CpG– and perforin-induced HLH liver injury

E. Eloseily[1,*], C. Arana[2], A. Akpaninyang[3], R.. Chhaing[4], M. Jordan[4], A. Grom[4], G. Schulert[4]
[1]Pediatrics; [2]Immunology; [3]Bioinformatics, UT Southwestern Medical Center, Dallas; [4]Pediatrics, Cincinnati Children's Hospital Medical Center, Cincinnati, USA
**Correspondence:** E. Eloseily
*Pediatric Rheumatology 2026*, **24(S1):**PT017

**Introduction**

Hemophagocytic lymphohistiocytosis (HLH) and macrophage activation syndrome (MAS) are hyperinflammatory syndromes characterized by immune dysregulation and multiorgan injury. Despite the underlying pathological differences, hepatic inflammation and liver injury are cardinal clinical manifestations of both HLH and MAS.

**Objectives**

To define shared and divergent hepatic transcriptional programs in mouse models of MAS and primary HLH using spatial transcriptomics.

**Methods**

Murine liver tissue from the CpG-induced model of MAS ($n$=5), perforin-deficient mice infected with LCMV to induce HLH ($n$=4), and PBS controls ($n$=4) underwent Visium spatial transcriptomic profiling; gene expression was pseudobulked at the mouse level, and differentially expressed genes (DEGs) were identified using FDR < 0.05 and |log2 fold change| > 1. Pathway enrichment analyses were performed using Reactome and Gene Ontology approaches.

**Results**

Both CpG-induced MAS and perforin-deficient HLH mice demonstrated extensive inflammatory remodeling relative to PBS controls, with 1,025 and 796 significant DEGs identified, respectively. A total of 498 DEGs overlapped between CpG- and perforin-induced HLH relative to PBS controls, supporting a shared inflammatory program. Shared signatures included induction of inflammatory chemokines, interferon-response genes, and acute phase reactants, including Cxcl9 and Saa3. CpG-induced MAS demonstrated enrichment of inflammatory, stress-response, and metabolic pathways, whereas perforin-deficient HLH demonstrated stronger adaptive immune and T-cell receptor signaling. Direct MAS-versus-HLH comparison identified 251 significant DEGs and revealed divergence in coagulation and extracellular matrix pathways. CpG-induced MAS showed enrichment of hemostasis, platelet activation, fibrin clot formation, and extracellular matrix organization, supported by platelet-associated genes such as Gp1ba. Cell type–specific analyses showed coordinated multicellular differences, with hepatocyte stress responses, macrophage activation, and stromal remodeling in CpG-induced MAS, and greater adaptive immune activation in CD8+ T cell–enriched regions in perforin-deficient HLH.

**Conclusion**

CpG-induced MAS and perforin-deficient HLH mice share inflammatory liver programs but diverge in downstream remodeling and immune ac-

tivation. CpG-induced MAS preferentially induces coagulation, stromal remodeling, and hepatocyte stress responses, whereas perforin-deficient HLH demonstrates greater adaptive immune activation. These findings highlight distinct hepatic inflammatory states between MAS and HLH, and identify coagulation and extracellular matrix remodeling as key features of TLR9-driven liver injury. Spatial zonation and cell-type deconvolution analyses are underway.

**Disclosure of interest**

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## PT018

### Bead-based NK cell degranulation assay as a reliable alternative to K562 cell line

V. Boz[1,*], A. Agrusti[1], S. Pastore[1], A. Taddio[1,2], A. Tommasini[1,2], E. Valencic[1]
[1]Pediatrics, Institute for Maternal and Child Health IRCCS Burlo Garofolo, [2]Medicine, Surgery and Health Sciences, University of Trieste, Trieste, Italy
**Correspondence:** V. Boz
*Pediatric Rheumatology 2026*, **24(S1):**PT018

**Introduction**

NK cell degranulation, assessed by surface CD107a expression via flow cytometry, is a critical diagnostic test for familial hemophagocytic lymphohistiocytosis (fHLH) and macrophage activation syndrome (MAS) in rheumatological settings. The conventional assay relies on tumor cell lines, like K562, as NK stimulators, which presents significant logistical challenges related to cell culture maintenance and viability.

**Objectives**

The aim of this study was to develop and validate a novel NK cell degranulation assay based on antibody-coated beads, capable of overcoming the limitations associated with the use of the tumor cell lines.

**Methods**

Anti-biotin-coated beads were loaded with biotinylated anti-CD2 and anti-NKp46 antibodies (NK Cell Activation/Expansion Kit, human, Miltenyi Biotec) to generate a standardized, ready-to-use NK cell stimulus. Peripheral blood mononuclear cells (PBMCs) were isolated from 15 healthy donors and from one patient with familial HLH and defective NK cell degranulation. PBMCs were incubated for 4 h with the antibody-loaded beads and CD107a antibody (Miltenyi Biotec). Following incubation, cells were stained with CD3 (Miltenyi Biotec), CD45 (BD), CD56 (Miltenyi Biotec) antibodies to quantify NK cell degranulation by measuring the percentage of CD107a-positive NK cells by flow cytometry.

**Results**

In the 15 healthy donors, the percentage of CD107a-expressing NK cells following bead-based stimulation ranged from 16.2% to 74.7%, establishing a broad but consistent reference interval for normal degranulation capacity. When the assay was applied to the patient with a previously documented total NK cell degranulation defect, results confirmed a complete absence of CD107a upregulation, fully concordant with the findings obtained at the reference laboratory using the conventional K562-based assay.

**Conclusion**

The bead-based NK cell degranulation assay is a valid and practical alternative to the K562 cell line stimulation method. By eliminating the need for continuous tumor cell culture, this approach significantly reduces analytical variability, simplifies workflow, and ensures test availability regardless of cell line status. Preliminary validation data demonstrate both diagnostic accuracy and concordance with established methods. In rheumatological settings, MAS is a life-threatening complication of systemic inflammatory diseases. The identification of cases that are underpinned by defective NK cell cytotoxicity can be relevant for therapeutic choices. This assay shows promise for routine clinical implementation in the diagnostic workup of fHLH and in the assessment of NK cell dysfunction in MAS within rheumatological practice. Further validation on a larger patient cohort is warranted.

**Disclosure of interest**

None declared.

## PT019

### Updated population pharmacokinetic (PK)/pharmacodynamic (PD) analysis of emapalumab in patients with macrophage activation syndrome (MAS) associated with Still's disease and systemic lupus erythematosus (SLE)

P. Brossard[1,*], A. Facius[2], B. Jamieson[3]
[1]Sobi, Basel, [2]thinkQ2 AG, Baar, Switzerland, [3]Sobi, Inc., Morrisville, USA
**Correspondence:** P. Brossard
*Pediatric Rheumatology 2026*, **24(S1):**PT019

**Introduction**

MAS is a rare, life-threatening complication of rheumatic diseases that is characterized by interferon-γ (IFNγ)-driven hyperinflammation. Emapalumab, a fully human anti-IFNγ monoclonal antibody, binds and neutralizes free and receptor-bound IFNγ.

**Objectives**

To update a previously developed population pharmacokinetic (PK)/pharmacodynamic (PD) model that described the PD effect of emapalumab in patients with MAS in Still's disease.

**Methods**

A previously established population PK/PD model using data from patients with MAS in Still's disease treated with emapalumab (Study 06)[1] was built upon using an updated population PK model that was developed using additional data from a study of emapalumab in patients with MAS associated with Still's disease and SLE (Study 14).[2] Individual PK parameters from this model were used as fixed parameters in a PK/PD model. The PD effects of emapalumab on serum chemokine C-X-C motif ligand 9 (CXCL9; a specific marker of IFNγ activity), soluble CD25 (sCD25; a marker of T cell activity), and ferritin concentrations (a marker of MAS activity) were implemented as inhibition of the respective synthesis rates using maximum inhibition ($I_{max}$) models. These PD parameters and their treatment-induced changes were characterized by turnover models. Parameters were assumed to be in steady-state at baseline with exposure-induced inhibition after treatment start.

**Results**

The final PK/PD model comprised a two-compartment PK model linked with 3 turnover models for CXCL9, sCD25, and ferritin concentrations. The model had correlation parameters between baseline CXCL9 levels, $I_{max}$ of CXCL9 and $I_{max}$ of ferritin, as well as between baseline and $I_{max}$ of sCD25 concentrations implemented. Estimated baseline levels of CXCL9, sCD25, and ferritin were 4630 ng/L, 8310 ng/L, and 24,200 mg/L, respectively, in Study 06, and 109 ng/L, 8310 ng/L, and 5034 mg/L in Study 14. No continuous exposure–response relationship was identified; complete inhibition was observed immediately after treatment start. Emapalumab almost completely suppressed CXCL9, sCD25, and ferritin production (estimated reduction in synthesis rate: 98.7%, 85.9%, and 99.4%, respectively, in Study 06 and 96.0%, 75.1%, and 99.4% in Study 14). Standard errors of all parameters obtained by a bootstrap procedure were <50% of their respective bootstrap means, indicating good model precision. Goodness-of-fit plots and visual predictive checks indicated good alignment between model predictions and observed concentrations for all parameters.

**Conclusion**

Population PK/PD modelling indicated that emapalumab rapidly suppresses CXCL9, sCD25, and ferritin production in MAS associated with both Still's disease and SLE.

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**Trial registration identifying number**

ClinicalTrials.gov identifier: NCT03311854 and NCT05001737.

**Disclosure of interest**

P. Brossard Employee with: Sobi, A. Facius Consultant with: Sobi, B. Jamieson Employee with: Sobi, Inc.

## PT020

### CD38HIGH as a marker of inflammatory activity: a single-centre pilot study

S. Mazzitelli[1,*], F. Lucioni[1], G. B. Beretta[1], F. Di Stasio[1], M. Rossano[1], L. Baselli[1], D. Montin[1], L. Porretti[2], E. Trombetta[2], F. Minoia[1]
[1]Pediatric Immuno-Rheumatology Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico Milano, [2]Flow Cytometry Laboratory, Clinical Pathology, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico Milano, Milan, Italy
**Correspondence:** S. Mazzitelli
*Pediatric Rheumatology 2026*, **24(S1):**PT020

**Introduction**
Hemophagocytic lymphohistiocytosis (HLH) is a severe hyperinflammatory syndrome caused by uncontrolled immune activation. Secondary HLH may complicate infections, malignancies, and rheumatic diseases, where it is referred to as macrophage activation syndrome (MAS). Distinguishing MAS from systemic juvenile idiopathic arthritis (sJIA) flare remains challenging because of overlapping clinical and laboratory features. Expansion of activated CD38high/HLA-DR+ CD8+ T cells has been associated with increased IFNγ production and cytokine storm amplification in HLH.
**Objectives**
To describe CD38 expression in a single-centre cohort of patients evaluated for suspected secondary HLH and assess its potential role in distinguishing MAS from sJIA flare.
**Methods**
We conducted a single-centre retrospective observational study including patients, both adult and pediatric, evaluated for suspected secondary HLH (sHLH) in whom CD38 expression levels were assessed as part of the diagnostic work-up. According to discharge diagnosis, patients were classified into HLH and non-HLH groups. HLH cases were further subdivided into macrophage activation syndrome (MAS) and secondary HLH (sHLH), while non-HLH patients included systemic juvenile idiopathic arthritis (sJIA) flare, infections, and other rheumatologic conditions. Since CD38 values showed non-normal distribution (Shapiro–Wilk $p<0.001$), comparisons were performed using the Mann–Whitney U test with Welch correction. CD38 positivity rates were evaluated using a 10.15% cutoff (1) and compared using Fisher's exact test. Statistical significance was set at $p < 0.05$.
**Results**
A total of 52 patients were included. CD38 values were significantly higher in patients with HLH compared with non-HLH conditions ($p < 0.001$). Likewise, CD38 positivity rates above the 10.15% cutoff were significantly more frequent in the HLH group ($\chi^2$ $p < 0.001$). Subgroup analyses demonstrated that both MAS and sHLH patients had significantly higher CD38 expression compared with patients experiencing sJIA flare. In particular, MAS showed significantly higher CD38 levels than sJIA flare ($p = 0.01$). Similarly, positivity rates were significantly higher in MAS compared with sJIA flare ($p= 0.036$). Overall, CD38 expression appeared consistently elevated in hyperinflammatory conditions fulfilling HLH criteria, while lower values were observed in inflammatory conditions not associated with HLH, particularly in isolated sJIA flare.
**Conclusion**
CD38 expression was significantly increased in HLH, particularly in MAS and sHLH. Higher CD38 levels in MAS compared with isolated sJIA flare suggest a potential role for CD38 as an adjunctive biomarker of hyperinflammation and a possible tool to support the differential diagnosis between MAS and active sJIA.

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**Disclosure of interest**
None declared.

## PT021

### Innate immune cell dysregulation in periodic fever with aphthous stomatitis, pharyngitis, adenitis (PFAPA)

A. Dernstedt[1,*], P. Girard[1], E. Sallansonnet[2], E. Arsac[1], P. Fontannaz[1], W. Adouan[1], S. Lemeille[1], A. Didierlaurent[1], G. Blanchard-Rohner[2]
[1]Pathology and Immunology, University of Geneva; [2]Immunology, Vaccinology, and Rheumatology, Geneva University Hospitals, Geneva, Switzerland
**Correspondence:** A. Dernstedt
*Pediatric Rheumatology 2026*, **24(S1):**PT021

**Introduction**
PFAPA (Periodic Fever, Aphthous stomatitis, Pharyngitis, Adenitis) is the most common autoinflammatory periodic fever syndrome in children, yet its immunopathological mechanisms remain poorly understood.
**Objectives**
To characterise innate immune cell dysfunction, serum cytokine profiles, and transcriptional pathway dysregulation longitudinally across PFAPA disease states.
**Methods**
Whole blood was collected from children with PFAPA during flares, between flares, and at long-term follow-up ($n$=13), alongside age- and sex-matched healthy controls ($n$=10). Innate immune cells were phenotyped at baseline and functionally assessed after 6-hour ex vivo stimulation with LPS (TLR4) or Gardiquimod (TLR7/8). Serum cytokines were quantified by multiplex immunoassay. Whole-blood bulk RNA sequencing was performed with GSEA pathway enrichment analysis.
**Results**
Circulating plasmacytoid dendritic cells (pDCs) were reduced during flares but normalised between flares, whereas type 2 conventional DCs (cDC2s) were persistently decreased at all timepoints, suggesting constitutive innate immune perturbation. LPS stimulation elicited increased IL-12 from cDC2s at all timepoints, while IFN-α from pDCs was concomitantly reduced. TLR7/8 stimulation similarly enhanced cDC2 IL-12, particularly during flares. Serum cytokine profiling revealed heterogeneous proinflammatory signatures during flares (CXCL10, granzyme B, IFN-γ, PD-L1, IL-10, MCP-1, IL-1RA, CD40L), suggesting potential endotypes in PFAPA. Transcriptomic analysis showed that the PFAPA flare signature resembled acute viral infection while also more prominently engaging neutrophil-related pathways. Neutrophil pathway enrichment persisted between flares and resolved at long-term follow-up.
**Conclusion**
cDC2s and pDCs in PFAPA children exhibits constitutive TLR signalling dysregulation with enhanced IL-12 and impaired IFN-α responses. Persistent inter-flare neutrophil pathway enrichment indicates baseline immune dysregulation potentially driving recurrent flares and representing a therapeutic target. Heterogeneous cytokine profiles suggest clinically relevant endotypes, informing personalised treatment strategies.
**Disclosure of interest**
None declared.

## PT022

### Validation and comparison of classification criteria for chronic nonbacterial osteomyelitis in a monocentric cohort

G. Inguscio[1,2,*], E. Roberts[1,3], M. Mohanna[3], S. Romano[1,2], C. E. Pain[1,3], C. M. Hedrich[1,3]
[1]Department of Paediatric Rheumatology, Alder Hey Children's NIHR Foundation Trust, Liverpool, UK; [2]Paediatric Department, University of Florence, Florence, Italy; [3]Department of Women's and Children's Health, Institute of Life Course and Medical Sciences, University of Liverpool, Liverpool, UK
**Correspondence:** G. Inguscio
*Pediatric Rheumatology 2026*, **24(S1):**PT022

**Introduction**
Chronic nonbacterial osteomyelitis (CNO) is an autoinflammatory bone disease mainly affecting children and adolescents. Diagnosis can be challenging due to heterogeneous presentations and lack of disease-specific biomarkers. Classification criteria are essential to generate homoge-

**Table 1 (Abstract PT022)** Performance of the 3 CNO classification tools

| Criteria for CNO | Jansson et al. | Bristol | EULAR/ACR |
|---|---|---|---|
| Sensitivity | 100% | 96.1% | 96.1% |
| Specificity | 63.9% | 95.6% | 98.3% |
| PPV | 54.2% | 90.2% | 96.1% |
| NPV | 100% | 98.3% | 98.3% |

neous cohorts for clinical trials. Three sets of classification criteria have been proposed for CNO, including most recent EULAR/ACR criteria. Criteria have been validated recently in retrospective cohorts from Türkiye.

**Objectives**

To compare the performance of CNO classification criteria in a retrospective cohort including a broad spectrum of mimickers.

**Methods**

Patients with CNO or mimicking conditions (2012-25) were identified at a single UK centre. Electronic patient records were reviewed, and classification criteria were applied retrospectively.

**Results**

Seventy-seven CNO patients and 180 mimickers were included (66 juvenile idiopathic arthritis (JIA), 42 bone cysts/malignancies, 39 haematological conditions (malignancies, Langerhans cell histiocytosis (LCH), sickle cell disease, 33 infectious osteomyelitis). The CNO group had a median age of 11.0 years (interquartile range [IQR], 9.0–12.8; range, 4.0–16.5) and 61% were female, while the mimicker cohort had a median age of 8.1 years (IQR, 4.5–12.5; range, 0.25–17.0) with 42% female patients. Regarding ancestry, the CNO group consisted of 70 individuals (90.9%) identifying as White, 2 (2.6%) as Asian, and 5 (6.5%) from other ethnic backgrounds. In the mimicker cohort, ancestry distribution included 116 individuals (64.4%) identifying as White, 9 (5.0%) as Black, 5 (2.8%) as Asian, 2 (1.1%) as Mixed, and 10 (5.6%) from other ethnic backgrounds; ancestry data were unavailable for 38 patients. Comparing three classification systems, modified Jansson criteria showed the highest sensitivity but lowest specificity; EULAR/ACR and Bristol criteria demonstrated a more balanced performance (Table 1). Modified Jansson criteria correctly classified all CNO cases but misclassified 65/180 (36%) mimickers as CNO, compared with 8 (4.4%) for Bristol and 3 (1.6%) for EULAR/ACR. Jansson criteria misclassified malignant bone tumours, bone cysts, psoriatic JIA, and LCH cases as CNO. Bristol misclassifications included enthesitis-related arthritis (ERA, 3), Ewing sarcoma (2), haematological malignancies (1), and infectious osteomyelitis (2). EULAR/ACR misclassifications involved ERA.

**Conclusion**

All criteria for CNO have high sensitivity, but specificity varies substantially. Modified Jansson criteria risks mis-classification of malignant or structural bone diseases as CNO, while EULAR/ACR and Bristol criteria provide greater specificity. Findings emphasise the importance of validating classification criteria in multiethnic backgrounds against a range of relevant mimickers. They underscore that classification criteria should not be used to diagnose patients.

**Disclosure of interest**

None declared.

## PT023

### Blau syndrome: real-world data from a national multicenter cohort

E. Aslan[1,*], S. Sahin[1], V. Guliyeva[2], E. Tunce[3], H. Tastanoglu[4], H. Ozturk Aydin[3], F. Gundogmus[5], S. Sener[6], H. H. Ozguner Kucuk[7], S. N. Tigrak[8], O. Necipoglu[9], Z. A. Kurt[10], K. Deveci Seker[11], C. Yildiz[11], D. Gezgin Yildirim[11], U. Akca Kaya[10], S. Ucar[8], A. Tanatar[7], S. Ozdemir Cicek[4], O. Koker[7], M. Kalyoncu[5], A. Pac Kisaarslan[4], S. Ozen[9], B. Sozeri[3], N. Aktay Ayaz[2], O. Kasapcopur[1]

[1]Pediatric Rheumatology, Istanbul University- Cerrahpasa; [2]Pediatric Rheumatology, Istanbul University; [3]Pediatric Rheumatology, University of Health Sciences, Ümraniye Training and Research Hospital, Istanbul; [4]Pediatric Rheumatology, Erciyes University, Kayseri; [5]Pediatric Rheumatology, Karadeniz Technical University, Trabzon; [6]Pediatric Rheumatology, Adana City Hospital, Adana; [7]Pediatric Rheumatology, Marmara University, Istanbul; [8]Pediatric Rheumatology, Pamukkale University, Denizli; [9]Pediatric Rheumatology, Hacettepe University, Ankara; [10]Pediatric Rheumatology, Akdeniz University, Antalya; [11]Pediatric Rheumatology, Gazi University, Ankara, Türkiye

**Correspondence:** E. Aslan

*Pediatric Rheumatology 2026*, **24(S1):**PT023

**Introduction**

Blau syndrome (BS) is a rare autoinflammatory disorder caused by gain-of-function variants in NOD2 gene and characterized by the triad of granulomatous arthritis, uveitis, and dermatitis. Due to its rarity and clinical heterogeneity, evidence regarding the optimum treatment strategies and long-term outcomes remain limited.(1).

**Objectives**

To describe clinical characteristics and treatment responses in a national BS cohort.

**Methods**

In this retrospective multicenter study, clinical characteristics, genetic findings, and treatment response data of patients with BS followed in eleven pediatric rheumatology centers across Türkiye were evaluated.

**Results**

Among 30 patients with BS, 56.7% were female. Median age of symptom onset and diagnosis were 3.05 and 7.74 years, respectively, with a median diagnostic delay of 2.41 years. Arthritis was the most common manifestation (87%), followed by boggy tenosynovitis (73%), dermatitis (67%), and uveitis (56%). Arthritis was the earliest manifestation (88% at disease onset), whereas dermatitis and uveitis developed at a median of 11.6 and 27.9 months after onset, respectively. Wrist involvement was nearly universal among patients with arthritis (95%), followed by knee (77%), and ankle (72%). Axial involvement was uncommon (9%). Genetic analysis identified 35 NOD2 variants across 30 patients. 68% of variants localised to the NACHT domain. The most frequently affected residue was R334Q/W, detected in 7/35 (20%) variants. Glucocorticoids and methotrexate were used in 90% and 93%, respectively. At least partial response was achieved with methotrexate in arthritis (95%) and dermatitis (100%), but uveitis showed no response in 50% of cases. Among patients with uveitis, 88% (15/17) required at least one bDMARD. The most common bDMARD was anti-TNFs (87%, 26/30), 80% of them were adalimumab. Adalimumab was effective for all arthritis and dermatitis cases, but only 62% of the uveitis patients. At the last follow-up, 24 patients remained on bDMARD therapy, with successful discontinuation achieved in only 2 of 26 (7.6%).

**Conclusion**

BS presents in early childhood with a characteristic triad of arthritis, dermatitis, and uveitis. In contrast to the classical sequence reported in the literature, arthritis was the earliest manifestation in our cohort, preceding dermatitis and uveitis. (1) Wrist involvement was nearly universal among patients with arthritis. The majority of NOD2 variants localised to the NACHT domain, with R334Q/W being the most frequent. Uveitis develops later in the disease course and is refractory to DMARDs, requiring biologic therapy. The bDMARD requirement in our BS (84%) cohort significantly exceeded the 50-60% rate typically observed in JIA, underscoring the highly refractory nature of BS.

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**Disclosure of interest**

None declared.

## PT024

### Biallelic MVK deficiency with a novel hemizygous TLR8 variant drives IL-6 dominant autoinflammation responsive to tocilizumab

P. Pimpale Chavan[1,*], B. Matthiasardottir[2,3], I. Ramos[4], D. L. Kastner[2], R. Khubchandani[1], I. Aksentijevich[2]

[1]Department of Pediatric Rheumatology, NH SRCC Children's Hospital, Mumbai, India; [2]Inflammatory Disease Section, Medical Genetics Branch, National Human Genome Research Institute, National Institute of Health, Bethesda; [3]Dept. of Cell Biology and Molecular Genetics, University of Maryland, College Park; [4]Center for Human Immunology, Autoimmunity, and Inflammation (CHI), National Institute of Allergy and Infectious Diseases, National Institute of Health, Bethesda, USA

**Correspondence:** P. Pimpale Chavan

*Pediatric Rheumatology 2026*, **24(S1):**PT024

**Introduction**
Mevalonate kinase deficiency (MKD) is an autoinflammatory disease caused by biallelic pathogenic variants in *MVK*, typically associated with IL-1–mediated inflammation. However, marked phenotypic variability suggests that additional genetic modifiers may influence disease severity.
**Objectives**
To investigate the molecular basis of severe inflammatory disease in a patient carrying homozygous *MVK* variants and identify mechanisms underlying the atypical phenotype and therapeutic response.
**Methods**
Whole-exome sequencing and segregation analysis were performed in the proband and family members. Structural modeling was conducted using AlphaFold and crystal structures of inactive and ligand-bound TLR8 dimers. Functional effects were assessed using NF-κB reporter assays in HEK-DUAL cells following stimulation with the TLR8 agonist TL8-506. Immune profiling was performed using CyTOF, and serum cytokine measurements and longitudinal clinical data were integrated with molecular findings.
**Results**
The male proband, born to a consanguineous family, presented with infantile-onset recurrent fever, episodic vomiting, drowsiness, granulomatous uveitis, rash, hepatosplenomegaly, and arthritis. Genetic analysis identified a homozygous mild pathogenic MVK p.N205D variant together with a novel maternally inherited hemizygous p.N491S variant in the X-linked gene *TLR8*. Both germline and somatic gain-of-function variants in TLR8 have been reported in patients with a spectrum of immune dysregulation, including recurrent infections and autoinflammation/autoimmunity. Notably, the mother, also homozygous for MVK p.N205D, exhibited late-onset arthritis, suggesting that MVK alone could not explain the disease severity in the proband. Structural modeling localized p.N491S to the TLR8 dimerization interface adjacent to known GoF variants and predicted altered conformational dynamics favoring activation. NF-κB reporter assays demonstrated enhanced ligand-dependent signaling in p.N491S-expressing cells following TL8-506 stimulation, consistent with gain-of-function activity. CyTOF immunophenotyping revealed dendritic-cell and memory B-cell abnormalities together with reduced IκBα across immune subsets, indicating tonic NF-κB activation. Serum IL-6 was markedly elevated only in the proband. Treatment with the IL-6 receptor antagonist tocilizumab resulted in sustained clinical improvement, normalization of inflammatory markers, and reduction in disease flares. This enabled steroid tapering, cessation of topical therapy, and improved growth.
**Conclusion**
These findings support a dual-pathway model in which impaired prenylation caused by *MVK* deficiency cooperates with hyperresponsive TLR8 signaling to drive IL-6–dominant autoinflammation. This study highlights how interacting variants affecting convergent inflammatory pathways can reshape disease biology and influence therapeutic response.
**Disclosure of interest**
None declared.

## PT025

### Polyautoimmunity and lymphoproliferation as red flags for inborn errors of immunity: insights from a pediatric rheumatology cohort

G. Olivieri[1,*], L. De Nardi[1], F. De Benedetti[1], A. Insalaco[1]
[1]Division of Rheumatology, ERN_RITA Centre, IRCCS Bambino Gesù Children's Hospital, Rome, Italy
**Correspondence:** G. Olivieri
*Pediatric Rheumatology 2026*, **24(S1):**PT025

**Introduction**
Inborn errors of immunity (IEI) are increasingly recognized as an underdiagnosed cause of immune dysregulation, often masquerading as isolated autoimmune diseases. In pediatric rheumatology, polyautoimmunity and/or lymphoproliferation may represent powerful clues to an underlying monogenic disorder.
**Objectives**
To characterize the clinical and immunological features of patients with polyautoimmunity and/or lymphoproliferation, and to determine the proportion of patients harboring a monogenic IEI, thereby assessing the diagnostic yield of genetic testing in this population.
**Methods**
We retrospectively evaluated patients referred to a tertiary Pediatric Rheumatology Clinic for complex immune dysregulation, defined by polyautoimmunity (≥3 immune-mediated clinical manifestations) and/or lymphoproliferation, who underwent genetic testing because of refractory disease or atypical features. Demographic, clinical, immunological, and genetic data were collected.
**Results**
Twenty-four patients were included (72% female). Median age at symptom onset was 3 years (IQR 4.5), while median current age was 21.3 years (IQR 7.8). Patients presented a median of 4 immune-mediated manifestations (IQR 1); antinuclear antibodies were positive in 16 cases (64%). Two main clinical phenotypes were identified: a predominantly polyautoimmune phenotype in 19 patients (79.2%) and an ALPS-like phenotype in 5 (20.8%). All patients underwent NGS panels for autoinflammatory diseases and IEI. A definitive molecular diagnosis of IEI was established in 8/24 patients (33%): CTLA-4 deficiency (*n*=2), SOCS1 haploinsufficiency (*n*=2), ataxia-telangiectasia due to ATM mutation (*n*=1), chronic granulomatous disease due to homozygous CYBC1 or NCF4 variants (*n*=2), and homozygous LRBA deficiency (*n*=1). Ten additional patients carried VUS in genes involved in immune regulation. Among genetically confirmed IEI cases, lymphoproliferation was observed in 75%, arthritis in 50%, and inflammatory bowel disease in 37.5%. Notably, only two patients reported recurrent respiratory infections. The median diagnostic delay was 11 years (IQR 11), with a median of 3 treatment lines (IQR 1.5) prior to genetic diagnosis.
**Conclusion**
In selected pediatric patients referred to rheumatology centres with a history of polyautoimmunity and/or lymphoproliferation, IEI should be considered in the differential diagnosis. Early referral for genetic evaluation should be pursued to reduce diagnostic delay and enable targeted therapeutic strategies.
**Disclosure of interest**
None declared.

## PT026

### A clinical referral score to guide genetic testing for adenosine deaminase 2 deficiency

O. Necipoglu Banak[1,*], E. Sag[1], A. E. Banak[2], Y. Bayindir[1], V. Cam[1], D. Unal[1], H. Ercan Emreol[1], M. O. Erkan[1], H. D. Dara Kar[1], S. Ozkan[1], O. Basaran[1], Y. Bilginer[1], S. Ozen[1]
[1]Pediatric Rheumatology, Hacettepe University, Ankara; [2]invent.ai, Istanbul, Türkiye
**Correspondence:** O. Necipoglu Banak
*Pediatric Rheumatology 2026*, **24(S1):**PT026

**Introduction**
Adenosine deaminase 2 deficiency (DADA2) is a rare autosomal recessive immune disorder with heterogeneous manifestations, including vasculopathy, hematologic abnormalities, and immunologic dysfunction. Diagnostic delays are common because of clinical overlap with systemic vasculitides and autoinflammatory diseases.
**Objectives**
To develop a phenotype-weighted clinical scoring system to support early referral for adenosine deaminase 2 enzyme analysis and/or genetic testing in patients with suspected DADA2.
**Methods**
This single-center retrospective cohort study included 130 patients evaluated at a tertiary pediatric rheumatology center. Twenty-eight patients had genetically and/or enzymatically confirmed DADA2, while 102 disease controls had systemic vasculitides, autoinflammatory diseases, or neuroinflammatory mimickers. Demographic, clinical, and laboratory features at diagnosis were analyzed. Candidate variables were selected using L1-regularized logistic regression, followed by integer-weight optimization to generate a practical bedside score. Diagnostic performance was assessed using receiver operating characteristic curve analysis.
**Results**
Bicytopenia, hypogammaglobulinemia, and musculoskeletal involvement were the strongest discriminatory variables for DADA2. The final

DADA2 Genetic Referral Score (DGRS) incorporated weighted clinical and laboratory parameters, including neurologic involvement, elevated acute-phase reactants, family history, consanguinity, and thrombocytosis which was assigned a negative weight because it was more frequent among disease controls. At an optimal cutoff of ≥10 points, the DGRS showed strong diagnostic performance, with a sensitivity of 0.86, specificity of 0.96, Youden's J of 0.82, positive likelihood ratio of 21.8, and an area under the curve of 0.933. This cutoff correctly identified most confirmed DADA2 patients while maintaining a low false-positive rate among clinically relevant mimickers.

**Conclusion**

The DGRS is a practical phenotype-weighted referral tool that may help clinicians identify patients who warrant early ADA2 enzyme analysis and/or genetic testing. By combining hematologic, immunologic, inflammatory, and familial features, the score may be particularly useful in distinguishing DADA2 from vasculitic and autoinflammatory mimickers. This approach could support earlier diagnosis, improve testing prioritization, and guide referral decisions, especially in settings where access to enzymatic or genetic testing is limited.

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**Disclosure of interest**

None declared.

## PT027

### When the JAK/STAT regulation fails: comparing immune dysregulation in SOCS1 and PTPN2 haploinsufficiency


S. Palmeri[1,2,*], M. Jeanpierre[1], J. Cognard[3], F. Zeco[1], M. Tusseau[4], Q. Riller[1], C. Brunaud[1], M. C. Stolzenberg[1], B. Neven[1,5], R. Caorsi[6], R. Gallizzi[7], S. Volpi[8], J. Hadjadj[1], S. Ehl[9], A. Belot[3], A. L. Mathieu[10], M. Gattorno[6], M. Parlato[1], F. Rieux-Laucat[1]

[1]Laboratory of Immunogenetics of Pediatric Autoimmune Diseases, Imagine Institute, INSERM UMR 1163, Université Paris Cité, Paris, France; [2]Rheumatology and Autoinflammatory Diseases Unit, University of Genoa and IRCCS Istituto Giannina Gaslini, Genoa, Italy; [3]National Referee Centre for Pediatric-Onset Rheumatism and Autoimmune Diseases, Hospices Civils de Lyon; [4]Genetics Department, Lyon University Hospital, Lyon, France; CIRI, International Center of Research in Infectiology, Lyon University, INSERM, Lyon; [5]Pediatric Immunology-Hematology and Rheumatology Unit, Necker Hospital, Paris, France; [6]Rheumatology and Autoinflammatory Diseases Unit, IRCCS Istituto Giannina Gaslini, Genoa; [7]Paediatrics, Department of Health Sciences, University of Catanzaro "Magna Graecia", 88100, Catanzaro; [8]Clinical and Experimental Immunology, IRCCS Istituto Giannina Gaslini, Genoa, Italy; [9]Institute for Immunodeficiency, Center for Chronic Immunodeficiency, Faculty of Medicine, University Medical Center Freiburg, University of Freiburg, Freiburg, Germany; [10]CIRI, International Center of Research in Infectiology, Lyon University, INSERM U1111, CNRS UMR 5308, ENS, UCBL, Lyon, France

**Correspondence:** S. Palmeri



*Pediatric Rheumatology 2026*, **24(S1):**PT027


**Introduction**

SOCS1 and PTPN2 are key negative regulators of the JAK/STAT pathway, recently associated with haploinsufficiency with overlapping autoimmunity and autoinflammation. In both conditions, enhanced cytokine signaling downstream of JAK/STAT has been demonstrated (1, 2). However, it remains unclear whether: (i) the level and qualitative profile of cytokine activation differ between the two disorders, (ii) compensatory mechanisms are engaged in the presence of a single regulatory defect but fail to adequately control immune hyperactivation.

**Objectives**

The objectives of this study were: (i) to functionally validate novel variants in SOCS1 and PTPN2 in patients with immuno-rheumatological phenotypes; (ii) to compare plasma cytokine levels in the two patient cohorts; (iii) to haracterize and compare the lymphocyte subsets in the two patient groups.

**Methods**

We analyzed 15 patients with *PTPN2* variants of uncertain significance (VUS) and 25 with *SOCS1* VUS using JAK/STAT activation assays in activated T cells. In selected cases pSTAT6 in response to IL-4 was assessed. Plasma cytokines (24-plex) were measured in 23 SOCS1 and 11 PTPN2 patients and compared to age-matched controls. In 3 patients, cytokines were evaluated before and after treatment with JAK-inhibitors. CyTOF was performed in PBMCs from 14 SOCS1, 8 PTPN2 patients, and controls.

**Results**

We expanded the clinical spectrum to include recurrent Henoch-Schönlein purpura (SOCS1) and juvenile Still's disease, multiple sclerosis, interstitial lung disease (PTPN2). Distinct cytokine profiles were identified: SOCS1 patients showed increased IL-4, IL-8, IL-10, IP-10, IL-17, and IL-12p70, whereas PTPN2 patients exhibited elevated IL-1β and MCP-1. CyTOF revealed reduced CXCR5+ T follicular regulatory cells and Tregs in both groups. CD38+ naïve B cells were decreased in PTPN2 only, while non-classical monocytes were reduced specifically in SOCS1.

**Conclusion**

SOCS1 and PTPN2 haploinsufficiency display distinct cytokine signatures without effective compensatory mechanisms, alongside shared defects in immune regulation. These preliminary findings require validation in larger cohorts.

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**Disclosure of interest**

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## PT028

### Arthritis in inborn errors of immunity: a multicenter cohort study (ImmunoReum study)


A. Civino[1,*], C. Milito[2], F. Pulvirenti[2], A. Matucci[3], E. Vivarelli[3], V. Moschese[4], M. Sgrulletti[4], F. Diomeda[1], G. Loiacono[1], F. Maiolini[5], E. Bassanello[5], F. Conti[6,7], M. Moratti[6,8], A. Uva[9], L. Martemucci[9], P. Semeraro[10,11], P. Airò[10], S. Ricci[12,13], M. Peri[12,13], R. Castagnoli[14,15], E. Palmisani[16], F. Fioredda[16], E. Cirillo[17], S. Naviglio[18], S. Volpi[19], A. Marzollo[20], D. Cuzzubbo[21], R. Gallizzi[22], M. Maltese[23], G. Costagliola[24], B. Martire[25], M. Cortesi[26], M. Rossano[27], F. Castellana[28], R. Greco[1], M. Cattalini[26], D. Montin[27], S. Pastore[18], A. Pin[18], R. Consolini[29], A. Soresina[26], C. Cancrini[30,31], C. Pignata[17], A. Ravelli[32,33], M. Gattorno[19], R. Badolato[26], A. Tommasini[18,34] on behalf of ImmunoReum Study Group

[1]Pediatric Rheumatology and Immunology Unit, 'Vito Fazzi' Hospital, Lecce; [2]Department of Molecular Medicine, "Sapienza" University of Rome, Rome; [3]Immunoallergology Unit, Careggi University Hospital, Florence; [4]Pediatric Immunopathology and Allergology Unit, Policlinico TorVergata, University of Rome Tor Vergata, Rome; [5]Internal Medicine, Immunology Unit, University Integrated Hospital of Verona, Verona; [6]Pediatric Unit, IRCCS Azienda Ospedaliero-Universitaria di Bologna; [7]Department of Medical and SurgicalSciences, Alma Mater Studiorum, University of Bologna, Bologna; [8]Department of Biomedicine and Prevention, Molecular Medicine and Applied Biotechnology, University of Rome Tor Vergata, Rome; [9]AORN Santobono Pausilipon, Naples; [10]Rheumatology and Clinical Immunology Unit, ASST Spedali Civili of Brescia; [11]Department of Clinical and Experimental Sciences, University of Brescia, Brescia; [12]Paediatric Immunology Unit, Meyer Children's Hospital IRCCS; [13]Department of Health Sciences, University of Florence, Florence; [14]Pediatric Unit, Department of Clinical, Surgical, Diagnostic and Pediatric Sciences, University of

Pavia; [15]Pediatric Clinic, Fondazione IRCCS Policlinico San Matteo, Pavia; [16]Hematology Unit, IRCCS Giannina Gaslini Institute, Genoa; [17]Department of Translational Medical Sciences, Section of Pediatrics, Federico II University, Naples; [18]Institute for Maternal and Child Health, IRCCS Burlo Garofolo, Trieste; [19]Rheumatology and AutoinflammatoryDiseases Unit, IRCCS Giannina Gaslini Institute, Genoa; [20]Pediatric Hematology, Oncology and Stem Cell Transplant Division, Padua University Hospital, Padua; [21]Pediatric Oncohematology Unit, Rodolico-San Marco University Hospital, Catania; [22]Department of Health Sciences, Magna Graecia University of Catanzaro, Catanzaro; [23]Pediatric Oncohematology Unit, Parma, Parma University Hospital, Parma; [24]Section of Pediatric Hematology and Oncology, Azienda Ospedaliero Universitaria Pisana, Pisa; [25]Pediatrics and Neonatology Unit, Maternal-Infant Department, "Monsignor A.R. Di Miccoli" Hospital, Barletta; [26]Clinical and Experimental Sciences, Pediatric Clinic, AziendaSocio-Sanitaria Territoriale Spedali Civili di Brescia, University of Brescia, Brescia; [27]Pediatric Rheumatology, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan; [28]Department of Interdisciplinary Medicine (DIM), University of Bari Aldo Moro, Bari; [29]Section of Clinical and Laboratory Immunology, Pediatric Unit, Department of Clinical and Experimental Medicine, University of Pisa, Pisa; [30]Department of Systems Medicine, University of Rome"Tor Vergata"; [31]Research Unit of Primary Immunodeficiencies, Unit of Clinical Immunology and Vaccinology, Bambino Gesù Children Hospital, IRCCS, Rome; [32]Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics and Maternal-Infantile Sciences (DINOGMI), University of Genova, University of Genova; [33]Scientific Direction, IRCCS Istituto Giannina Gaslini, Genoa; [34]University of Trieste, Trieste, Italy

**Correspondence:** A. Civino

*Pediatric Rheumatology 2026*, **24(S1):**PT028

**Introduction**

Arthritis is the most frequent rheumatologic symptom in patients with inborn errors of immunity (IEI) and may precede the diagnosis. Better characterization of arthritis in IEI is needed to improve early recognition and support evolving targeted therapies.

**Objectives**

To analyze the clinical features, onset time, and prevalence of arthritis in a multicenter cohort of IEI patients.

**Methods**

Retrospective cross-sectional study across 24 centers of the Italian Network for Primary Immunodeficiencies, enrolling consecutive IEI patients with rheumatologic symptoms, excluding autoinflammatory diseases (Jan 2025–Jan 2026). For prevalence analysis, centers reported the total number of IEI patients evaluated during the study. A sub-analysis on arthritis, defined by standardized rheumatologic criteria, was performed.

**Table 1 (Abstract PT028)** Comparison of IEI patients with and without arthritis

| | All patients (*n*=201) | Patients with arthritis (*n*=108) | Patients without arthritis (*n*=93) | *p* |
|---|---|---|---|---|
| Females | 135 (67.2) | 80 (74.1) | 55 (59.1) | 0.036 |
| Age at IEI diagnosis, yrs | 19.58 [7.65, 47.37] | 30.45 [8.34, 52.59] | 14.78 [7.05, 37.07] | 0.02 |
| Time between first symptom and IEI diagnosis, yrs | 2.29 [0.42, 4.16] | 2.61 [0.65, 4.57] | 1.93 [0.23, 3.62] | 0.010 |
| Predominantly antibody deficiency | 146 (72.6) | 88 (81.5) | 58 (62.4) | <0.01 |
| ≥3 Therapeutic lines | 63 (31.3) | 39 (36.1) | 24 (25.8) | <0.01 |
| bDMARDs | 56 (28.1) | 41 (38.7) | 15 (16.1) | 0.001 |
| csDMARDs | 84 (42.0) | 64 (59.8) | 20 (21.5) | <0.001 |
| Systemic glucocorticoids | 90 (45.0) | 50 (46.7) | 40 (43.0) | 0.700 |
| NSAIDs | 59 (29.5) | 5 (42.1) | 14 (15.1) | <0.001 |

Data are expressed as number (%), and median [IQR]

**Results**

A total of 201 IEI patients with rheumatologic symptoms were enrolled. Arthritis was the most frequent symptom, occurring in 108 patients (53.7%), among whom predominantly antibody deficiency was the most frequent category (81.5%), followed by immune dysregulation disorders (9.3%). Females accounted for 74.1% (80/108) of patients; median age at IEI diagnosis 30.45 yrs (IQR 8.34-52.59). Arthritis preceded IEI diagnosis in 72 patients (66.7%) and represented the first symptom in 32 (29.6%), with a median diagnostic delay of 3.10 yrs (IQR 0.44–7.75). A defined rheumatic disease was diagnosed in 72.2% patients, most commonly undifferentiated inflammatory arthritis (28.0%), juvenile idiopathic arthritis (27.1%), rheumatoid arthritis (13.1%), and systemic lupus erythematosus (4.7%). Compared to patients with other rheumatologic symptoms, those with arthritis showed higher female prevalence, longer diagnostic delay, older age at IEI diagnosis, and greater therapeutic burden.

**Conclusion**

We found that arthritis is the most common rheumatologic manifestation in IEI, may precede diagnosis in more than half of cases, with a diagnostic delay of several years, and exposes patients to a significant therapeutic burden. Our findings highlight the importance of considering IEI in the differential diagnosis of arthritis and underscore the need for careful immunologic screening before immunosuppressive therapy.

**Disclosure of interest**

None declared.

## PT029

### TNFAIP3: a shared monogenic determinant of MIS-C and Kawasaki disease?

J. Cognard[1,*], M. Tusseau[1], F. Zekre[2], G. Arnold[3], E. Jeziorski[4], H. Reumaux[5], J. Tantot[6], F. Mestrallet[6], S. Benezech[6], P. S. Rohrlich[7], A. Laurent[1], A. L. Mathieu[8], D. Lee[9], V. Sancho Shimizu[10], J. L. Casanova[11], S. Y. Zhang[12], A. Belot[1]

[1]HFME, Lyon; [2]CHU Saint Etienne, Saint Etienne; [3]CH Valence, Valence; [4]CHU Montpellier, Montpellier; [5]CHU Lille, Lille; [6]HCL, Lyon; [7]CHU Nice, Nice; [8]INSERM, Lyon, France; [9]Harvard, Boston, USA; [10]Imperial College London, London, UK; [11]Institut Imagine, Paris, France; [12]Rockfeller University, New York, USA

**Correspondence:** J. Cognard

*Pediatric Rheumatology 2026*, **24(S1):**PT029

**Introduction**

MIS-C is a post–SARS-CoV-2 syndrome with frequent cardiac and vascular involvement. Its clinical overlap with Kawasaki disease (KD) suggests convergent pathways and supports the hypothesis that KD may represent a final common inflammatory outcome of diverse infectious triggers in genetically predisposed children. [1]

**Objectives**

Rare monogenic inborn errors of immunity (IEIs) have been reported in MIS-C. [2–4] TNFAIP3 encodes A20, a negative regulator of NF-κB signaling, and A20 haploinsufficiency is expected to lower the threshold for infection-triggered inflammation.

**Methods**

We identified a pathogenic TNFAIP3 variant in a child with recurrent inflammatory episodes presenting with severe MIS-C, leading to diagnosis of HA20. We then screened a large MIS-C exome dataset (*n*=558) for rare TNFAIP3 variants, and reviewed molecularly confirmed HA20 patients within the French inflammatory disease network for MIS-C or KD phenotypes.

**Results**

The index patient had recurrent fevers prior to MIS-C and developed shock-like hyperinflammation with TCR Vβ21.3+ T-cell expansion and a positive type I IFN signature. Exome sequencing identified rare non-synonymous TNFAIP3 variants in MIS-C (*n*=14) and KD (*n*=4) cases (MAF <0.01). Two molecularly confirmed HA20 patients with a KD phenotype and one MIS-C–like case carrying a TNFAIP3 VUS were additionally identified. Thirteen variants were functionally tested in an NF-κB luciferase reporter assay; 3/13 showed loss of function due to reduced protein expression. SARS-CoV-2 infection assays and PBMC single-cell RNA sequencing are ongoing.

**Conclusion**

These findings support a role for IEIs in a subset of MIS-C and identify TNFAIP3 as a shared monogenic contributor to MIS-C and KD, support-

ing a common genetic architecture. NF-κB dysregulation may underlie their phenotypic overlap and intersect with interferon responses. Recurrent or atypical KD and severe MIS-C should prompt consideration of monogenic IEIs, including TNFAIP3.

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**Disclosure of interest**
None declared.

## PT030

**Leukoglycemic index: a novel biomarker for detecting attack–related inflammation in familial Mediterranean fever?**


P. Esmeray Şenol[1,*], G. Gökulu[2]
[1]Department of Pediatric Rheumatology; [2]Department of Pediatric Emergency Medicine, Mersin City Training and Research Hospital, Mersin, Türkiye
**Correspondence:** P. Esmeray Şenol

*Pediatric Rheumatology 2026*, **24(S1):**PT030

**Introduction**
FMF is the most common hereditary autoinflammatory disease in children, caused by *MEFV* mutations. Distinguishing an active attack from subclinical inflammation remains challenging. The Leuko-Glycemic Index (LGI = Glucose [mg/dL] × Leucocyte [$10^3$/μL] / 1000) reflects systemic inflammation and metabolic stress, with proven value in cardiovascular disease, COVID-19, and DKA — yet unexplored in autoinflammatory diseases.
**Objectives**
This study aimed to evaluate the diagnostic capacity of LGI for detecting attack-related inflammation in pediatric FMF patients classified according to the 2019 Eurofever/PRINTO criteria.
**Methods**
69 patients followed at Mersin City Training and Research Hospital (Feb–Aug 2025) were retrospectively assigned to four groups: Group 1 — Active Attack ($n$=19; CRP >3.2 mg/L or SAA >0.5 mg/L + characteristic feature); Group 2 — Subclinical Inflammation ($n$=7; elevated markers without clinical attack); Group 3 — Attack-Free ($n$=38; ≥30 attack-free days, normal APR); Group 4 — MEFV Carrier ($n$=5; no FMF criteria, no colchicine). Exclusions: diabetes, infection, corticosteroids. LGI was calculated from fasting glucose and leucocyte count; Group 1 samples were drawn within 24 h of symptom onset.
**Results**
A total of 69 patients were included ($n$=19 active attack, $n$=7 subclinical inflammation, $n$=38 attack-free, $n$=5 MEFV gene carrier). Median age 10.0 years (IQR 7–14); 52.2% male; median disease duration 33 months. Attack-period LGI (median 0.90 [0.65–1.05]) was significantly higher than attack-free LGI in the same patients (0.60 [0.60–0.80]; $p$=0.049). ROC analysis (Group 1 vs. all others) yielded AUC=0.807 (95% CI: 0.674–0.918); optimal cut-off LGI ≥0.80; sensitivity 63.2%, specificity 84.0%. LGI did not correlate with CRP, ESR, NLR, or SAA (all $p$>0.05), suggesting independence from conventional markers. Attack-free LGI did not differ across groups ($p$=0.211).
**Conclusion**
LGI is a state-dependent biomarker that rises significantly during active FMF attacks and discriminates attack from other clinical states with AUC=0.807. Its calculation from routine tests makes it a practical adjunctive tool. Prospective multicentre validation is warranted.

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**Disclosure of interest**
None declared.

## PT031

**Kawasaki disease in Italy: TEN years of nationwide epidemiological data**


G. Inguscio[1,2,*], L. Gatti[2], S. Romano[1,2], I. Pagnini[2], I. Maccora[2], M. V. Mastrolia[2], E. Marrani[2], R. Massaro[3], C. Pescosolido[3], A. Marchesi[4], L. Gargiullo[4], M. Fabi[5], M. C. Maggio[6], M. Scortichini[7], G. Simonini[1,2], T. Giani[2]
[1]Paediatric Department, University of Florence; [2]Rheumatology Unit, AOU Meyer Children's Hospital IRCCS, Florence; [3]Rari ma Speciali – Kawasaki Disease Family and Patient Association; [4]Emergency, Acceptance and General Pediatrics Department, Bambino Gesù Children's Hospital, IRCCS, Rome; [5]Pediatric Emergency Unit, IRCCS Azienda Ospedaliero Universitaria di Bologna, Bologna; [6]Department of Health Promotion, Maternal and Infantile Care, Department of Internal Medicine and Medical Specialties "G. D'Alessandro", University of Palermo, Palermo; [7]Economic Evaluation and HTA (EEHTA)-CEIS, Faculty of Economics, University of Rome "Tor Vergata", Rome, Italy
**Correspondence:** G. Inguscio

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**Introduction**
Kawasaki disease (KD) is an acute pediatric vasculitis and the leading cause of acquired heart disease in children in high-income countries. Nationwide epidemiological and longitudinal data on KD in Italy remain limited.
**Objectives**
This study aimed to evaluate epidemiological trends, regional distribution, seasonality, hospitalization burden, and cardiac complications of KD in Italy.
**Methods**
We conducted a retrospective nationwide study using the Italian hospital discharge database (Scheda di Dimissione Ospedaliera, SDO), including pediatric patients hospitalized with KD between 2010 and 2019. Incidence estimates were calculated using pediatric population data from the Italian National Institute of Statistics (Istituto Nazionale di Statistica, ISTAT). A longitudinal cohort was created by including each subject at the first identifiable hospitalization and following them for 365 days. Since complete administrative data were available through 2020, patients hospitalized in 2019 were excluded from longitudinal analyses to ensure complete follow-up. Cardiac complications included coronary artery aneurysms, myocarditis, pericarditis, and myocardial infarction identified during index hospitalization or within one year.
**Results**
A total of 5,485 pediatric patients with KD were identified, accounting for 9,503 hospitalizations. Estimated incidence ranged from 4.5 to 8.2 cases per 100,000 children aged 0–15/16 years and from 15.8 to 28.7 per 100,000 in children <5 years. Hospitalization rates ranged from 3.1 to 8.8 per 10,000 pediatric inhabitants across Italian regions. Between 2010 and 2019, 5,447 patients accounted for 9,457 hospitalizations nationwide, corresponding to 1.7 hospitalizations per patient. The highest numbers of patients were reported in Lazio, Lombardia, Sicilia, and Campania, while the highest hospitalization burden per patient was observed in Lazio (3.2) and Campania (2.4). KD hospitalizations showed seasonal variation, with peaks in winter and spring and lower rates during summer. Index hospitalizations progressively decreased over time,

from 16% in 2010 to 9% in 2018. The longitudinal cohort included 4,965 patients. Cardiac complications occurred in 4.5% of patients during index hospitalization and in 6.4% within one year. Coronary artery aneurysms were identified in 2.9% of patients during index hospitalization and in 4.5% within one year. Myocarditis occurred in 0.8% of patients, pericarditis in 1.2%, and myocardial infarction in 0.1%. Male patients showed higher rates of cardiac complications than females (7.7% vs. 4.6%). Infants younger than one year and patients aged 9–16 years had the highest rates of cardiac complications (7.5% and 9.3%, respectively). Coronary artery aneurysms occurred in 5.5% of infants at one year, whereas myocarditis was more frequent among adolescents (3.1%).

**Conclusion**

KD remains a significant cause of pediatric hospitalization in Italy, with marked regional variability and a clear seasonal pattern. Although cardiac complications were overall uncommon, coronary artery aneurysms remained a relevant outcome, particularly in infants, while adolescents showed the highest rates of myocarditis and overall cardiac involvement. Male sex and extreme age groups were associated with a higher risk of cardiac complications.

**Disclosure of interest**

None declared.

## PT032

### Gastrointestinal involvement in pediatric Behçet's disease: a national multicenter study from Türkiye

E. Özçelik[1,*], H. Ercan Emreol[2], A. Alişan[3], H. Adıgüzel Dündar[4], N. Zeynep Özarslan[5], S. Şener[6], Z. Torunoğlu[7], Ş. Doğantan[8], T. Kasap[9], O. Bahçeci[10], H. H. Özgüner[11], H. Ö. Başaran[2], O. Köker Turan[11], Z. B. Özçakar[10], S. Yüksel[9], M. Yıldız[7], N. Şahin[5], R. M. Kışla Ekinci[3], S. Özen[2], B. Çelikel Acar[1]

[1]Pediatric Rheumatology, Bilkent City Hospital; [2]Pediatric Rheumatology, Hacettepe University, Ankara; [3]Pediatric Rheumatology, Cukurova University, Adana; [4]Pediatric Rheumatology, University of Health Sciences, Dr. Behçet Uz Children's Hospital, İzmir; [5]Pediatric Rheumatology, Kocaeli University, Kocaeli; [6]Pediatric Rheumatology, Adana City Hospital, Adana; [7]Pediatric Rheumatology, Istanbul University–Cerrahpaşa; [8]Pediatric Rheumatology, Basaksehir Cam and Sakura City Hospital, İstanbul; [9]Pediatric Rheumatology, Çanakkale Onsekiz Mart University, Çanakkale; [10]Pediatric Rheumatology, Ankara University, Ankara; [11]Pediatric Rheumatology, Marmara University, İstanbul, Türkiye

**Correspondence:** E. Özçelik

*Pediatric Rheumatology 2026*, **24(S1):**PT032

**Introduction**

Pediatric Behçet's disease (BD) is a chronic systemic vasculitis that can affect arteries and veins of all sizes and involve different organ systems with varying severity.

**Objectives**

This multicenter study conducted in Türkiye aimed to evaluate the clinical characteristics of pediatric BD patients with gastrointestinal (GI) involvement.

**Methods**

The study included pediatric BD under 18 years of age from various centers who met the ICBD or PEDBD criteria, had endoscopically/colonoscopically confirmed GI involvement, and were followed for at least 6 months.

**Results**

Twenty-eight pediatric BD patients with GI involvement (6.5%) were included among 428 patients followed across 11 centers. Fourteen of the patients (50%) were male. GI involvement was present at diagnosis in 12 patients (42.9%) and developed during follow-up in 16 patients (57.1%). At diagnosis, 26 patients (92.9%) had oral aphthae, 14 (50%) had genital ulcers, 17 (60.7%) had cutaneous involvement, 5 (17.9%) had ocular involvement, and 5 (17.9%) had neurological involvement. Pathergy test positivity was detected in 10 patients (35.7%). In 17 (60.7%) patients HLA-B51 was positive. The most common GI symptoms were abdominal pain in 26 patients (92.9%), diarrhea in 22 (78.6%), hematochezia in 5 (17.9%), reflux in 4 (14.3%), and constipation in 2 (7.1%). The course of GI involvement was monophasic in 10 patients (35.7%), recurrent in 14 (50%), and chronically persistent in 4 (14.3%). Acute abdomen was observed in 5 patients (17.9%); however, no perforation occurred, and no surgical intervention. Endoscopic evaluation revealed antral gastritis in 6 patients (21.4%) and pangastritis in 5 (17.9%). Ulcers were detected in 5 patients (17.9%), and 3 of them (60%) had multiple ulcers localized to the antrum. Colonoscopy demonstrated ileal inflammation in 8 patients (40%) and colonic inflammation in 7 (35%). Colonic ulcers were detected in 15 patients (53.6%), amongthese patients, 12 (80%) had multiple ulcers. The ileum was the most frequently involved site, affecting 16 patients (57.1%), followed by the stomach in 15 (53.6%), colon in 13 (46.4%), ileocecal region and rectum in 8 each (28.6%), and esophagus in 5 (17.9%). Following the diagnosis of GI involvement, corticosteroid treatment was initiated in 20 patients (71.4%). Azathioprine was added in 20 patients (71.4%), methotrexate in 2 (7.1%), mesalazine in 6 (21.4%), adalimumab in 8 (28.6%), and infliximab in 3 (10.7%). During follow-up, remission was achieved in 22 patients (75%).

**Conclusion**

GI involvement was identified in 6.6% of pediatric BD patients. GI involvement was observed during followup in 57.1% of patients, highlighting the importance of monitoring GI symptoms throughout the disease course. Remission was achieved in 75% of patients, although a substantial proportion required biologic therapy, emphasizing the need for early treatment escalation.

**Disclosure of interest**

None declared.

## PT033

### Gut microbial dysbiosis and immune remodelling after intravenous immunoglobulin in Indian children with Kawasaki disease

R. K. Pilania[1,*], V. Thakur[1], M. Dhaliwal[1], S. Sarkar[1], A. Kumar[1], S. Sharma[1], A. Rawat[1], A. Gupta[1], S. Singh[1]

[1]Pediatric Allergy Immunology Unit, Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research, Chandigarh, India

**Correspondence:** R. K. Pilania

*Pediatric Rheumatology 2026*, **24(S1):**PT033

**Introduction**

Kawasaki disease (KD) is an acute childhood vasculitis in which gut microbial dysbiosis may contribute to systemic inflammation and immune dysregulation.

**Objectives**

To evaluate gut microbial composition, cytokines, Th17 and T regulatory cells, and related transcriptional markers in Indian children with KD before and after intravenous immunoglobulin (IVIg) treatment.

**Methods**

Forty-one children with KD were studied during the acute pre-IVIg phase and after IVIg. Median age was 3.0 years (IQR 1.0–6.0), and 31 (75.6%) were male. Gut microbiota was assessed by 16S rRNA sequencing, cytokines by ELISA, Th17/Treg subsets by flow cytometry, and RORγT/FOXP3 expression by qRT-PCR.

**Results**

Pre-IVIg KD showed gut microbial dysbiosis, with high *Enterococcus* abundance (30.53 ± 36.40%) and lower abundance of commensal genera including *Blautia* (4.02 ± 7.57%), *Faecalibacterium* (6.65 ± 14.77%), *Bifidobacterium* (34.20 ± 26.41%) and *Lactobacillus* (1.83 ± 2.52%). After IVIg, *Enterococcus* decreased to 4.14 ± 10.25% ($p$=0.0002), while *Blautia* increased to 9.91 ± 9.71% ($p$=0.0008), *Faecalibacterium* to 13.64 ± 17.07% ($p$=0.059) and *Bifidobacterium* to 47.31 ± 20.93% ($p$=0.062). Shannon diversity increased from 7.7207 ± 0.7869 to 8.2076 ± 0.3040 ($p$=0.0034), and beta diversity differed after IVIg (PERMANOVA F=3.54, $p$=0.0023). Cytokine profiling revealed elevated baseline levels of inflammatory and regulatory mediators. After IVIg, IL-6 decreased from 389.70 ± 1056.63 to 72.30 ± 260.39 pg/mL ($p$=0.0020), IL-17A from 3.65 ± 3.41 to 1.23 ± 1.18 pg/mL ($p$=0.0166) and IL-10 from 7.97 ± 11.32 to 1.31 ± 1.35 pg/mL ($p$<0.0001), while TGF-β1 remained elevated (674.87 ± 302.39 to 634.13 ± 274.68 pg/mL; $p$=0.5118). Compared with controls, baseline levels of TGF-β1, IL-6, and IL-10 were significantly higher, whereas IL-17A was not. At baseline, Th17 frequency was comparable between KD and controls (0.41% [0.125–1.53] vs. 0.67% [0.345–1.445]; $p$=0.151). Treg frequency was higher in KD (8.785% [5.0–14.3] vs. 5.68% [4.185–7.645]; $p$=0.003), while FOXP3+ Tregs were comparable (4.94% [2.002–9.845] vs. 3.06% [0.925–5.52]; $p$=0.111). RORγT expression was higher in baseline KD than controls (ΔCt −3.16 ± 6.01 vs. 3.89 ± 3.31; $p$=0.019), whereas FOXP3 expression was unchanged.

**Conclusion**

Acute KD was characterised by gut microbial dysbiosis, cytokine imbalance and altered regulatory immune responses. IVIg was associated

with microbiome restructuring and reduction of inflammatory cytokines, while persistent TGF-β1 and increased RORγT expression support ongoing immune activation/remodelling.

**Trial registration identifying number**

Not applicable.

**Disclosure of interest**

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## PT034

### Performance of the Kawanet score in Kawasaki disease: external validation and contribution of biomarkers in a french pediatric cohort

E. Barbieri[1,2,*], I. Koné-Paut[2], P. Dusser-Benesty[2], F. Anselmi[2]
[1]Department of Medicine and Health Sciences "Vincenzo Tiberio", University of Molise, Campobasso, Italy; [2]Department of Paediatric Rheumatology and CEREMAIA, Bicêtre University Hospital, Paris, France
**Correspondence:** E. Barbieri
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**Introduction**

Prediction of intravenous immunoglobulin (IVIG) resistance in Kawasaki disease (KD) remains a major clinical challenge, particularly in non-Asian populations where most available risk scores show limited generalisability and inconsistent performance across heterogeneous cohorts.

**Objectives**

To assess the external validation of the Kawanet score in a French cohort of children with KD. Secondary exploratory analyses assessed whether selected inflammatory and biochemical biomarkers could improve its predictive performance. To identify factors associated with IVIG resistance and to explore whether their integration into the Kawanet score improves its predictive performance.

**Methods**

We conducted a retrospective monocentric cohort study including children diagnosed with KD and managed at a tertiary referral centre in France between 2015 and 2025. Both complete and incomplete forms of KD were included, reflecting real-world clinical practice. The Kawanet score was reconstructed using four admission variables (alanine aminotransferase >30 U/L, hepatomegaly, lymphocyte count >2400/mm$^3$, and IVIG administration ≤5 days from fever onset). IVIG resistance was defined pragmatically as the need for a second IVIG infusion and/or second-line treatment. Diagnostic performance was assessed using sensitivity, specificity, predictive values and receiver operating characteristic (ROC) curve analysis. Associations were evaluated using logistic regression, Fisher's exact test and Spearman correlation. Exploratory analyses assessed C-reactive protein (CRP) and serum sodium at admission, based on their potential role as markers of disease severity.

**Results**

Fifty-six patients were included (64.3% male; median age 1.86 years), with a predominance of complete KD forms. IVIG resistance occurred in 25/56 patients (44.6%), consistent with the tertiary-care setting and referral bias towards more severe phenotypes. Coronary artery aneurysms were observed in 20/56 patients (35.7%) during follow-up, highlighting the clinical relevance of early risk stratification. Using the standard cut-off (≥2), the Kawanet score showed limited predictive performance, with sensitivity 76% and specificity 26%, indicating poor ability to correctly classify non-resistant patients. ROC analysis confirmed limited discrimination (AUC 0.61). No significant association was observed between the Kawanet score and IVIG resistance or coronary artery aneurysms, suggesting limited utility of the score in this population. CRP ($p$ = 0.017) and hyponatremia ($p$ = 0.029) were significantly associated with IVIG resistance and each showed moderate discriminative performance (AUC = 0.71). In exploratory analyses, a modified score integrating these biomarkers showed improved performance (AUC 0.69), with increased sensitivity (84.6%) and moderate specificity (56.5%).

**Conclusion**

In this French tertiary-care cohort, the Kawanet score alone showed limited external validity for predicting IVIG resistance in KD and did not reliably identify patients at risk of coronary complications. Integration of inflammatory and biochemical biomarkers may improve risk stratification and support a more individualised approach to early treatment intensification. These findings support the need for multicentre European studies to develop and validate more robust and generalisable prediction models.

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**Disclosure of interest**

None declared.

## PT035

### The invisible burden in pediatric Behcet's disease: fatigue, sleep quality and treatment adherence

B. Danışman Avcı[1,*], T. Güzel[2], A. Doğru Kılınç[1], B. Başer Taşkın[1], A. Tanatar[3], F. G. Demirkan[4], F. Çakmak[2], Ö. Akgün[1], N. Aktay Ayaz[1]
[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine; [2]Pediatric Rheumatology, Basaksehir Cam and Sakura City Hospital; [3]Pediatric Rheumatology, Marmara University Pendik Training and Research Hospital; [4]Pediatric Rheumatology, Kanuni Sultan Suleyman Training and Research Hospital, İstanbul, Türkiye
**Correspondence:** B. Danışman Avcı
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**Introduction**

Behçet's disease (BD) is a chronic, relapsing, multisystemic inflammatory vasculitis that may begin during childhood. Clinical assessment in pediatric BD (pBD) primarily focuses on disease activity and organ involvement, whereas patient-reported outcomes such as fatigue, sleep quality, and treatment adherence are frequently overlooked.

**Objectives**

To evaluate fatigue burden, sleep quality, and medication adherence in pediatric patients with BD and compare these parameters with healthy controls.

**Methods**

This controlled cross-sectional study included 40 pediatric patients fulfilling PEDBD criteria and 55 age- and sex-matched healthy controls. Disease activity was assessed using the International Behçet's Disease Dynamic Activity Measure (IBDDAM) and Behçet's Disease Current Activity Form (BDCAF). Fatigue, sleep quality, and medication adherence were evaluated using the Pediatric Quality of Life Inventory Multidimensional Fatigue Scale (PedsQL-MFS), Pittsburgh Sleep Quality Index (PSQI), and 8-item Morisky Medication Adherence Scale (MMAS-8), respectively. Visual Analog Scale (VAS) scores were recorded for pain, fatigue, patient global assessment, and physician global assessment.

**Results**

The mean age at diagnosis of patients with pBD was 146.30±36.28 months, with a diagnostic delay of 24.0±27.54 months and a mean follow-up duration of 31.18±24.21 months. The most frequent clinical manifestations were oral aphthae (95%), arthralgia (47.5%), and pseudofolliculitis (35%). Mean baseline IBDDAM and BDCAF scores decreased from 4.18±2.54 and 3.03±1.20 to 1.55±1.43 and 1.30±1.18 at the final visit, respectively. Total and subscale PedsQL-MFS scores, including general fatigue and sleep/rest fatigue, were lower in patients with BD compared with healthy controls ($p$<0.001, $p$<0.001, and $p$=0.003, respectively). In contrast, total and subscale PSQI scores, including subjective sleep quality, sleep disturbance, and daytime dysfunction, were higher in the pBD group ($p$=0.028, $p$<0.001, $p$=0.003, and $p$<0.001, respectively). Low treatment adherence was identified in 70% of patients. Mean patient and physician global assessment scores were 4.4±2.2 and 2.95±1.2.

**Conclusion**

Despite low disease activity, fatigue and sleep disturbances persisted in patients with pBD, while low treatment adherence was frequently ob-

served. To the best of our knowledge, this is the first study evaluating fatigue, sleep quality, and treatment adherence together in pBD. These findings suggest that factors contributing to disease burden in pBD should be evaluated using a multidisciplinary and patient-centered approach.

**Disclosure of interest**
None declared.

## PT036

### Independent effects of age and IVIg resistance and the impact of first-line glucocorticoids on coronary outcomes in Kawasaki disease

M. V. Mastrolia[1,*], V. Pandiarajan[2], M. Cattalini[3], A. Taddio[4], S. Vergnano[5], J. Anton[6], W. Sontichai[7], B. Sozeri[8], S. Ozen[9], S. Rosina[10], J. Sánchez-Manubens[11], R. Haviv[12], P. Pal[13], A. Rodrigues Fonseca[14], A. Gagro[15], N. Kumar Bagri[16], L. Verdoni[17], D. Montin[18], M. C. Maggio[19], A. Thangaraj[2], R. Pilania[2], M. Mosquera[6], J. Calzada[6], R. Sittiwangkul[7], K. Ulu[8], Z. Balik[9], Y. Bilginer[9], M. Garrone[10], E. Patrone[10], G. Carreras[1], N. Ruperto[20], G. Simonini[1] on behalf of for the Paediatric Rheumatology European Society (PReS) Vasculitis Working Party and for the Pediatric Rheumatology INternational Trials Organisation (PRINTO)

[1]Meyer Children's Hospital IRCCS, Florence, Italy; [2]Postgraduate Institute of Medical Education and Research, Chandigarh, India; [3]University of Brescia and Spedali Civili di Brescia, Brescia; [4]IRCCS "Burlo Garofolo" and University of Trieste, Trieste, Italy; [5]Bristol Royal Hospital for Children, Bristol, UK; [6]Hospital Sant Joan de Déu, Barcelona, Spain; [7]Chiang Mai University, Chiang Mai, Thailand; [8]Umraniye Training and Research Hospital, Instanbul; [9]Hacettepe University, Ankara, Türkiye; [10]IRCCS Istituto Giannina Gaslini, Genova, Italy; [11]Institut d'Investigaciò I Innovaciò Parc Taul' (I3PT-CERCA), Barcelona, Spain; [12]Meir Medical Center, Tel Aviv, Israel; [13]Institute of Child Health, Kolkata, India; [14]Instituto de Puericultura e Pediatria Martagao Gesteira (IPPMG), Rio de Janeiro, Brazil; [15]Children's Hospital Zagreb, Zagreb, Italy; [16]All India Institute of Medical Sciences, New Delhi, India; [17]Hospital Papa Giovanni XXIII, Bergamo; [18]Regina Margherita Children's Hospital, Torino; [19]University of Palermo, Palermo; [20]Fondazione IRCCS San Gerardo dei Tintori di Monza, Monza, Italy

**Correspondence:** M. V. Mastrolia

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**Introduction**
Coronary artery aneurysms (CAAs) represent the most serious complication of Kawasaki disease (KD). Although young age and IVIg resistance are recognized risk factors, their independent impact remains uncertain, and the benefit of early adjunctive glucocorticoid therapy on coronary outcomes is still controversial.

**Objectives**
To assess the effects of age and IVIg resistance on CAAs, and evaluate the impact of first-line glucocorticoids on coronary sequelae.

**Methods**
An analysis of the KIWI international multicenter cohort was conducted to identify predictors of cardiac involvement using multivariable logistic regression adjusted for age, sex, ethnicity, fever duration, IVIg resistance, and CRP levels. Patients were stratified by age group. A secondary subanalysis focused on patients presenting with CAAs at diagnosis to assess the impact of first-line glucocorticoid therapy on coronary sequelae at 6–12 months. Relative risks were estimated using Poisson regression models adjusted for age, sex, ethnicity, fever duration, CRP at onset, baseline coronary Z-score, and IVIg resistance.

**Results**
Overall, 19.8% of patients (142/722) were IVIg-resistant, with no difference across age groups. Infants <6 months exhibited the highest risk of both overall cardiac involvement and CAAs. Compared with infants, older children had significantly lower odds of cardiac involvement (6–11 months: OR 0.31, 95% CI 0.15–0.62; 12–47 months: OR 0.32, 95% CI 0.17–0.59; >47 months: OR 0.32, 95% CI 0.17–0.60). A similar pattern was observed for CAAs, with significantly reduced risk in children aged 12–47 months (OR 0.24, 95% CI 0.08–0.66) and >47 months (OR 0.17, 95% CI 0.05–0.48). IVIg resistance remained an independent predictor of cardiac involvement (OR 2.79, 95% CI 1.87–4.18). In patients with CAAs at diagnosis (181/722), first-line glucocorticoids were not associated with a reduction in coronary sequelae, even in high-risk subgroups including infants ≤6 months and patients with baseline Z-score ≥5 (Table 1).

**Table 1 ( Abstract PT036)**

| | RR | 95% CI | *p* value | RR | 95% CI | *p* value | RR | 95% CI | *p* value |
|---|---|---|---|---|---|---|---|---|---|
| **First-line glucocorticoids** | 1.01 | 0.77–1.32 | 0.95 | 0.98 | 0.53–1.84 | 0.96 | 1.06 | 0.64–1.77 | 0.82 |
| *Age at diagnosis (months)* | 0.99 | 0.95–1.04 | 0.78 | 1.61 | 0.07–40.01 | 0.77 | 0.98 | 0.90–1.07 | 0.63 |
| *Gender* | 0.97 | 0.75–1.24 | 0.78 | 0.98 | 0.49–1.97 | 0.96 | 0.97 | 0.55–1.72 | 0.91 |
| *Ethnicity* | 1.03 | 0.90–1.17 | 0.72 | 0.97 | 0.65–1.44 | 0.87 | 0.96 | 0.71–1.30 | 0.79 |
| *Fever duration (days)* | 0.99 | 0.98–1.01 | 0.56 | 0.99 | 0.95–1.03 | 0.50 | 1.00 | 0.98–1.03 | 0.98 |
| *CRP (mg/dL)* | 0.99 | 0.98–1.01 | 0.50 | 0.99 | 0.95–1.03 | 0.53 | 0.99 | 0.96–1.02 | 0.51 |
| *IVIg resistance* | 0.97 | 0.75–1.27 | 0.83 | 1.08 | 0.57–2.04 | 0.81 | 0.91 | 0.53–1.57 | 0.73 |
| *Baseline Z score* | 0.97 | 0.95–1.00 | 0.06 | 0.99 | 0.94–1.05 | 0.72 | 0.98 | 0.94–1.02 | 0.27 |

**Conclusion**
Young age and IVIg resistance independently increase the risk of coronary involvement in KD, with infants <6 months at highest risk for CAAs. First-line glucocorticoids were not associated with reduced coronary sequelae suggesting limited efficacy on vascular remodelling.

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**Trial registration identifying number**
NCT06305611.

**Disclosure of interest**
None declared.

## PT037

### Multi-modal and longitudinal immune profiling uncovers a distinct monocytes phenotype distribution and function in Kawasaki disease

G. Olivieri[1,*], A. Neri[2], A. Rotili[2], M. Piccari[2], A. Marchesi[3], N. Cotugno[4], P. Palma[4], F. De Benedetti[1]

[1]Division of Rheumatology, ERN-RITA Centre, IRCCS Bambino Gesù Children's Hospital; [2]University of Rome Tor Vergata; [3]Division of Emergency, Acceptance and General Pediatrics; [4]Division of Clinical Immunology and Vaccinology, ERN-RITA Centre, IRCCS Bambino Gesù Children's Hospital, Rome, Italy

**Correspondence:** G. Olivieri

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**Introduction**
Kawasaki disease (KD) is an acute childhood vasculitis characterized by monocyte (MO)-driven innate immune activation.

**Objectives**
To comprehensively profile systemic inflammation and MO biology in KD by integrating plasma proteomics with deep immunophenotyping and functional assays, and to assess MO remodeling following IVIG therapy.

**Methods**
We enrolled 10 KD patients alongside age- and sex-matched healthy controls (HC, *n*=10) and febrile controls (FC, *n*=7). Blood samples from KD patients were collected at three timepoints: acute disease (T0), 48 h

post-IVIG (T1), and 4 weeks post-IVIG (T2). Plasma proteins ($n$=180) were quantified using Olink assay. MO function was assessed using a two-day stimulation assay, in which adherent MO were cultured and stimulated with LPS or left unstimulated, followed by application of a 16-marker functional panel to quantify subsets, activation states, chemokine-receptor expression, and intracellular cytokine production. Data analysis included unsupervised clustering using the Leiden algorithm.

**Results**

A total of 120,204 cells were profiled using the phenotype panel, identifying 11 clusters by Leiden clustering. We defined 9 MO subsets (cluster 1-9), alongside two clusters consistent with low-density neutrophils (LDNs, clusters 0 and 10). Compared with HC, KD patients exhibited* marked expansion of cluster 6 (activated classical MO) and cluster 9 (proliferating MO expressing Ki67/CD38). Both clusters correlated positively with inflammatory proteins linked to vascular injury. Notably, cluster 6 correlated with APBB1IP, previously identified by our group as associated with coronary artery involvement (1), suggesting a potential mechanistic role in vascular inflammation. Conversely, clusters 1, 7, and 8, (representing homeostatic MO) were reduced in KD. Cluster 10 (LDNs) was markedly enriched in KD children and displayed the strongest proteomic signature, with positive correlations to IL-17A, CXCL9, IL-10, CSF1, and mediators of Th17/IFN-driven inflammation, monocyte– dendritic cell crosstalk, and tissue repair pathways. KEGG and GO analyses revealed significant enrichment in cytokine–cytokine receptor interactions, leukocyte migration, and chemotaxis. Functional profiling showed that KD classical MO exhibited elevated baseline TNF-α production, whereas non-classical and intermediate MO subsets from FC patients displayed higher baseline IL-6 levels. When cytokine responses were analyzed as Δ values, significant differences in TNF-α and IL-6 production were observed across all monocyte subsets in KD patients compared with FC. Following IVIG therapy, MO composition partially normalized, evidenced by increases in clusters 1 and 8 and reductions in activated subsets (clusters 3 and 10) at T2. Post-IVIG, we observed an increase in delta TNF-α levels compared with baseline, whereas IL-6 responses remained unchanged.

**Conclusion**

This study provides a characterization of MO and LDNs in KD, revealing disease-specific alterations and a central role for activation and extravasation pathways. MO displayed disease-specific priming with elevated baseline TNF-α yet retained strong responsiveness to secondary stimulation. The increase in TNF-α delta after IVIG may reflect trained immunity. IVIG dampens pathological inflammation while preserving MO responsiveness, contributing to clinical improvement and vascular protection.

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**Disclosure of interest**

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## PT038

### A comparative clinical analysis of Kawasaki disease and MIS-C (multisystem inflammatory syndrome in children): results from the KIWI and Hyper-Ped COVID registries

R. Caorsi[1,*], M. Pasquinucci[2], M. V. Mastrolia[3], B. Sozeri[4], V. Pandiarajan[5], F. Haerynck[6], M. Cattalini[7], S. Ozen[8], J. Anton[9], A. Taddio[10], W. Sontichai[11], E. Faugier[12], R. Athimalaipet[13], A. Ravelli[1], M. Gattorno[1], G. Simonini[3] on behalf of for the Hyper-Ped COVID and the KIWI registries

[1]IRCCS Istituto Gaslini; [2]Università degli studi di Genova, Genova; [3]Meyer Hospital, Firenze, Italy; [4]University of Health Sciences, Instambul, Türkiye; [5]Postgraduate Institute of Medical Education and Research, Chandigarh, India; [6]Ghent University Hospital, Ghent, Belgium; [7]Spedali civili di Brescia, Brescia, Italy; [8]Hacettepe University, Ankara, Türkiye; [9]Sant Joan de Déu Hospital, Barcelona, Spain; [10]IRCCS Burlo Garofolo, Trieste, Italy; [11]Chiang Mai University, Bangkok, Thailand; [12]Hospital Infantil de México Federico Gómez, Mexico City, Mexico; [13]Bristol Medical School (THS), Bristol, UK

**Correspondence:** R. Caorsi

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**Introduction**

Kawasaki Disease (KD) and Multisystem Inflammatory Syndrome in Children (MIS-C) represent two major hyperinflammatory disorders of childhood that share significant clinical and pathogenic similarities. Despite extensive research, it remains unclear whether they are distinct entities or part of a single continuum of pediatric hyperinflammation triggered by infectious agents in genetically predisposed individuals.

**Objectives**

To compare two large international cohorts of patients diagnosed with either KD or MIS-C.

**Methods**

Patients with KD were extracted from Kiwi registry, collecting patient between 2015 and 2023, and patients with MIS-C from The HyperPed-COVID registry, which retrospectively collected MIS-C patients from 2020; demographic, clinical, laboratory, and instrumental parameters were compared using Student's t test or Mann-Whitney U test for continuous data, and $\chi^2$ or Fisher's exact tests for categorical data. A multivariable logistic regression model was developed to identify the most discriminative clinical and laboratory predictors; age-stratified analyses (<1 year, 1–4 years, 4–8 years) assessed phenotypic overlap across age groups.

**Results**

1701 patients (722 KD, 979 MIS-C) were included in the study. KD patients were significantly younger (3.2 ± 2.6 years vs. 7.9 ± 4.6 years, $p$<0.0001). Mucocutaneous manifestations were nearly universal in KD (99.7%), but also highly prevalent in MIS-C (82.1%). Gastrointestinal involvement — particularly abdominal pain — was the strongest distinguishing marker for MIS-C (62.6% vs. 15.1%; Cramér's V 0.48). Coronary abnormalities, especially aneurysms (33.9% KD vs. 7.7% MIS-C), were strongly associated with KD, whereas myocarditis (39.5% vs. 9.8%) and pericarditis (24.3% vs. 10.2%) were more frequent in MIS-C. The final multivariable model demonstrated excellent discriminative performance (AUC = 0.912; Pseudo-$R^2$ = 0.46). The strongest independent predictors of KD were coronary aneurysm (OR 7.36), cheilitis (OR 3.93), and cervical lymphadenopathy (OR 2.22); the strongest predictors of MIS-C were abdominal pain (OR 0.20), myocarditis (OR 0.33), and older age (OR 0.33 per SD). Age-stratified analysis revealed a significant drop in diagnostic performance in children aged 1–4 years (AUC = 0.805), a range in which phenotypic overlap is greatest and laboratory markers are least discriminating.

**Conclusion**

Despite notable clinical differences that allowed the development of a multivariable model with high accuracy, KD and MIS-C present a clear overlap in younger children.

**Disclosure of interest**

None declared.

## PT039

### Glycocalyx-related biomarkers in pediatric IgA vasculitis: associations with gastrointestinal and renal involvement

H. Musil[1], N. Kifer[2], D. Grguric[2], A. Kozmar[3], M. Frkovic[2], M. Sestan[2], M. Jelusic[2,*]

[1]University of Zagreb School od Medicine; [2]Department of Paediatrics; [3]Department of Laboratory Diagnostics, University of Zagreb School of Medicine, University Hospital Centre Zagreb, Zagreb, Croatia

**Correspondence:** M. Jelusic

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**Introduction**

IgA vasculitis (IgAV) is the most common childhood vasculitis and may be associated with gastrointestinal (GI) and renal involvement. Endothelial glycocalyx damage contributes to vascular permeability and endothelial dysfunction during inflammation. Glycocalyx-related biomarkers may therefore reflect disease severity in IgAV.

**Objectives**

To evaluate serum and urine glycocalyx-related biomarkers in children with IgAV and analyze their association with disease manifestations and activity.

**Methods**

Serum and urine Syndecan, Glypican and Spectrin concentrations were measured in children with IgAV at disease onset and after 6 months, and in healthy controls. Associations between biomarker levels and GI

manifestations, renal involvement, relapse number and laboratory parameters were analyzed using nonparametric and correlation analyses.
**Results**
Forty-nine children with IgAV (25 males, 24 females) and 17 healthy controls were included. Nephritis was present in 20 patients (40.8%), severe GI manifestations in 16 (32.7%), and severe skin involvement in 7 (14.3%). Median (Q1, Q3) age was 6.6 (4.85–8.15) years and median Paediatric Vasculitis Activity Score (PVAS) was 9 (3–13). Compared with controls, Serum Spectrin concentrations were significantly higher in IgAV patients ($p$=0.0001), while serum Glypican concentrations also differed between groups ($p$=0.035). Lower Syndecan concentrations were associated with severe GI manifestations, including lower baseline serum Syndecan (2.77 vs. 4.28 ng/mL, $p$=0.007), baseline urine Syndecan (0.71 vs. 1.61 ng/mL, $p$=0.010), and 6-month urine Syndecan levels (0.48 vs. 1.56 ng/mL, $p$=0.012). Lower baseline urinary Syndecan concentrations were also associated with combined proteinuria and hematuria (0.64 vs. 1.43 ng/mL, $p$=0.042), while higher baseline serum Spectrin concentrations were associated with isolated hematuria (7.08 vs. 5.54 ng/mL, $p$=0.050). Higher 6-month serum Spectrin concentrations correlated with relapse number (rho=0.305, $p$=0.039) and creatinine levels (rho=0.360, $p$=0.026).Spectrin was associated with persistent disease activity and renal involvement, Syndecan with severe GI and renal manifestations, while Glypican mainly differed between IgAV patients and controls, suggesting endothelial activation and remodeling.

| Biomarker | Association | *p*-value |
|---|---|---|
| Serum Spectrin | Higher in IgAV patients vs. controls | 0.0001 |
| Serum Glypican | Altered in IgAV patients vs. controls | 0.035 |
| Urine Syndecan | Lower in proteinuria+hematuria | 0.042 |
| Serum Spectrin | Higher in isolated hematuria | 0.050 |
| 6-month serum Spectrin | Positive correlation with relapse number | 0.039 |
| 6-month serum Spectrin | Positive correlation with creatinine | 0.026 |

**Conclusion**
Glycocalyx-related biomarkers are altered in pediatric IgAV and demonstrate distinct associations with GI and renal involvement. Lower Syndecan concentrations may reflect glycocalyx depletion during severe organ involvement, while higher Spectrin concentrations may indicate persistent endothelial and cellular injury. These findings support a potential role of endothelial glycocalyx injury in IgAV pathophysiology. Support: Croatian Science Foundation project HRZZ-IP-2024-05-6848 and NPOO 10106-25-2897 (GUT-VAS), funded by the EU – NextGenerationEU.
**Disclosure of interest**
None declared.

## PT040

### Comparison of pediatric vasculitis activity score, Indian Takayasu clinical activity score, and physician global assessment at diagnosis in 205 children with childhood-onset Takayasu arteritis: an icTAK registry study

S. Gagne[1,2,*], J. Bistolarides[3], V. Sivaraman[4], K. Morishita[5,6], V. Podzolkova[7], M. Cüceoğlu[8], S. Al-Mayouf[9], M. Jelusic[10], M. Šestan[10], L. Wagner-Weiner[11], D. Cabral[5,6], S. Ozen[8] on behalf of The International Collaboration on Childhood-Onset Takayasu Arteritis (icTAK)
[1]Department of Pediatrics, University of Pittsburgh School of Medicine; [2]Division of Pediatric Rheumatology, University of Pittsburgh Medical Center, Pittburgh; [3]Medical College of Wisconsin, Milwaukee; [4]Division of Pediatric Rheumatology, Nationwide Children's Hospital, Columbus, USA; [5]Pediatric Rheumatology, British Columbia Children's Hospital; [6]University of British Columbia, Vancouver, Canada; [7]Department of Children's Diseases, N.F. Filatov Clinical Institute of Children's Health, Moscow, Russian Federation; [8]Division of Rheumatology, Hacettepe University, Ankara, Türkiye; [9]King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia; [10]University of Zagreb School of Medicine, Zagreb, Croatia; [11]Section of Pediatric Rheumatology, University of Chicago Comer Children's Hospital, Chicago, USA
**Correspondence:** S. Gagne
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**Introduction**
Takayasu arteritis is a rare chronic large vessel vasculitis predominantly affecting the aorta and its major branches. Disease assessment is challenging due to symptom variability, and repeated imaging is often required. Currently no validated disease activity scores exist specifically for childhood-onset Takayasu arteritis (cTAK), although the Paediatric Vasculitis Activity Score (PVAS) is validated for all chronic childhood vasculitides and the Indian Takayasu Clinical Activity Score (ITAS-2010) is validated in adult Takayasu.
**Objectives**
To evaluate correlation and agreement between PVAS, ITAS-2010, and physician global assessment (PGA) in cTAK.
**Methods**
Data were obtained from the International Collaboration on Childhood-onset Takayasu Arteritis (icTAK) registry. PVAS and ITAS-2010 were scored from available clinical data. PGA was recorded by the contributing physician on a 0–100 visual analog scale. Continuous variables are reported as median (IQR). Pearson's r was used to assess correlation, intraclass correlation coefficient (ICC) to assess agreement, Spearman's rho for correlation with non-continuous variables, and Bland-Altman analysis to characterize the nature of disagreement between measures.
**Results**
Of 205 children with diagnosis visit data, all had available PVAS and ITAS-2010 scores; PGA was missing in 15 patients. Median scores for PVAS, ITAS-2010, and PGA were 10 (IQR 6–13), 5 (IQR 3–11), and 69 (IQR 50–80) respectively. Significant mild-to-moderate correlations were observed between PVAS and ITAS-2010 ($r$=0.514, 95% CI 0.406–0.609), PVAS and PGA ($r$=0.332, 95% CI 0.199–0.452), and ITAS-2010 and PGA ($r$=0.277, 95% CI 0.140–0.403), all $p$<0.001. ICC between PVAS and ITAS-2010 was moderate (0.433, 95% CI 0.191–0.602, $p$<0.001). PGA demonstrated poor, non-significant agreement with both PVAS (ICC 0.026, 95% CI -0.029–0.098) and ITAS-2010 (ICC 0.017, 95% CI -0.022–0.069). Bland-Altman analysis revealed a systematic bias with PGA consistently scoring approximately 50 points higher than both PVAS and ITAS-2010 on a 0–100 scale, with disagreement widening at higher disease activity levels. Both ITAS-2010 (ρ=0.316, $p$<0.001) and PGA (ρ=0.190, $p$=0.008) correlated with the number of vessels involved at diagnosis; only PGA correlated with ESR (ρ=0.171, $p$=0.024).
**Conclusion**
PVAS and ITAS-2010 demonstrated moderate correlation and agreement with each other but poor agreement with PGA. PGA showed weak but significant correlations with vessel burden and ESR, neither of which were captured by PVAS or ITAS-2010. These findings suggest that current scoring systems fail to capture important elements of disease activity in cTAK, likely reflecting the predominance of vascular over systemic manifestations. Adaptation and further validation of existing tools for cTAK is warranted.
**Disclosure of interest**
None declared.

## PT041

### Divergent temporal patterns of anti-dsDNA in childhood-onset SLE: a predictive window for lupus nephritis but limited utility in non-renal flares

N. Akay[1,*], S. Sahin[1], A. Agbas[2], Esra Karabag Yilmaz[2], Zeynep Torunoglu[1], Ece Aslan[1], Umit Gul[1], Elif Kilic Konte[1], Seha Saygili[2], Mehmet Yildiz[1], Kenan Barut[1], Yasemin Ozluk[3], Ozgur Kasapcopur[1], Nur Canpolat[2]
[1]Department of Pediatric Rheumatology; [2]Department of Pediatric Nephrology, Istanbul University-Cerrahpasa, Cerrahpasa Faculty of Medicine; [3]Department of Pathology, Istanbul University, Istanbul Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** N. Akay
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**Introduction**
Anti-dsDNA antibodies are widely used to monitor disease activity in childhood-onset SLE (cSLE), yet their temporal dynamics preceding organ-specific flares remain poorly defined.
**Objectives**
This study aimed to characterize pre-flare anti-dsDNA trajectories in cSLE and assess whether distinct temporal patterns are associated with renal versus non-renal flares.

**Methods**
This single-center retrospective study included 102 children with cSLE (2009–2025) classified by renal involvement status during follow-up. Significant organ involvement requiring treatment escalation with high-dose corticosteroids and/or immunosuppressive therapy was evaluated at the episode level and categorized as renal and non-renal flares. Anti-dsDNA positivity was defined as >24 IU/mL (>2× ULN). Measurements obtained at −24, −18, −12, −6, and −3 months before flare were analyzed to compare temporal trends across flare types using linear mixed-effects models.
**Results**
Among 102 children with cSLE, 43 had renal involvement, and 59 had no renal involvement. In total, 58 renal flares, including nephritis presenting at disease onset and subsequent renal flare episodes, and 88 non-renal flares were identified. Overall, renal flares showed significantly higher anti-dsDNA levels than non-renal flares ($p$=0.011), with divergent temporal patterns confirmed by a significant group-by-time interaction ($p$=0.023). Longitudinal analysis of 26 renal and 27 non-renal flares with available serial pre-flare anti-dsDNA data revealed distinct anti-dsDNA trajectories. In renal flares, median anti-dsDNA levels remained persistently elevated throughout the 24-month pre-flare period (range 228-300 IU/mL), with consistently high proportions exceeding 2× ULN (87–94%) and 5× ULN (78–86%) at all time points. A relative nadir was observed at −18 months (median 228 IU/mL; the only time point significantly lower than flare, $p$=0.008), followed by higher levels toward the renal flare, peaking at −3 months and at flare (median 300 IU/mL). In non-renal flares, median anti-dsDNA levels showed a more variable trajectory (range 189.5–300 IU/mL), peaking at −3 months (300 IU/mL), with lower proportions exceeding 2× ULN (68–85%) and 5× ULN (55–74%), and no sustained pre-flare elevation.
**Conclusion**
Temporal anti-dsDNA patterns differed markedly by organ involvement: sustained elevation characterized renal flares, while non-renal flares showed no consistent pre-flare rise. These findings support the role of serial anti-dsDNA monitoring in the follow-up of lupus nephritis, while suggesting a more limited discriminatory utility for non-renal disease activity.

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**Disclosure of interest**
None declared.

## PT042

### Monogenic lupus: genetic results, clinical spectrum, and outcomes from a nationwide multicenter cohort

S. Sahin[1], N. Akay[1,*], S. Atamyildiz Ucar[2], A. Alisan[3], E. Tunce[2], E. Kayhan[4], Y. Ugur Es[5], A. Alkaya[6], B. Baser Taskin[7], D. Unal[8], L. Koru[9], H. K. Zora[10], A. Uzun Bektas[11], H. M. Kacmaz[12], H. Kazanasmaz[13], S. Caglayan[14], S. Sener[15], B. Acun[16], G. Ozomay Baykal[17], Y. Demir Yigit[18], S. E. Albayrak[19], B. Bozkaya[20], O. Nurlu[21], F. Haslak[9], M. Yildiz[1], A. Adrovic[1], K. Barut[1], H. I. Yilmaz[22], M. Dogan[22], A. Gezdirici[22], M. Eser[23], S. Ozdemir Cicek[4], S. Balci[24], M. Kaya Gurgoze[19], A. Balat[21], S. Bakkaloglu[16], B. Bora[11], S. S. Kilic[10], M. Kalyoncu[13], S. Elmas[20], O. Koker[25], N. Aktay Ayaz[7], S. Ozen[8], B. Celikel Acar[5], A. Pac Kisaarslan[4], R. M. Kisla Ekinci[3], B. Sozeri[2], O. Kasapcopur[1]
[1]Department of Pediatric Rheumatology, Istanbul University-Cerrahpasa, Cerrahpasa Medical Faculty; [2]Department of Pediatric Rheumatology, Umraniye Training and Research Hospital, University of Health Sciences, Istanbul; [3]Department of Pediatric Rheumatology, Cukurova University Faculty of Medicine, Adana; [4]Department of Pediatric Rheumatology, Erciyes University Faculty of Medicine, Kayseri; [5]Department of Pediatric Rheumatology, University of Health Sciences, Ankara Bilkent City Hospital; [6]Department of Pediatric Rheumatology, University of Health Sciences, Etlik City Hospital, Ankara; [7]Department of Pediatric Rheumatology, Istanbul University Faculty of Medicine, Istanbul; [8]Department of Pediatric Rheumatology, Hacettepe University Faculty of Medicine, Ankara; [9]Department of Pediatric Rheumatology, Istanbul Medeniyet University Faculty of Medicine, Istanbul; [10]Department of Pediatric Rheumatology, Uludag University Faculty of Medicine, Bursa; [11]Department of Pediatric Rheumatology, Dokuz Eylul University Faculty of Medicine, İzmir; [12]Department of Pediatric Rheumatology, Diyarbakır Children's Hospital, Diyarbakır; [13]Department of Pediatric Rheumatology, Karadeniz Technical University Faculty of Medicine, Trabzon; [14]Department of Pediatric Rheumatology, Antalya Training and Research Hospital, Antalya; [15]Department of Pediatric Rheumatology, Adana City Training and Research Hospital, Adana; [16]Department of Pediatric Rheumatology, Gazi University Faculty of Medicine, Ankara; [17]Department of Pediatric Rheumatology, Mardin Training and Research Hospital, Mardin; [18]Department of Pediatric Rheumatology, Gazi Yasargil Training and Research Hospital, Diyarbakır; [19]Department of Pediatric Rheumatology, Fırat University Faculty of Medicine, Elazığ; [20]Department of Pediatric Rheumatology, Samsun Training and Research Hospital, Samsun; [21]Department of Pediatric Rheumatology, Gaziantep University Faculty of Medicine, Gaziantep; [22]Department of Medical Genetics, Basaksehir Cam and Sakura City Hospital; [23]Department of Medical Genetics, Umraniye Training and Research Hospital, University of Health Sciences, Istanbul; [24]Department of Pediatric Rheumatology, Dicle University, Faculty of Medicine, Diyarbakır; [25]Department of Pediatric Rheumatology, Marmara University Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** N. Akay
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**Introduction**
Monogenic lupus remains incompletely characterized, with limited data on its genetic landscape, clinical phenotype, and outcomes.
**Objectives**
To characterize real-world data describing the genetic landscape, clinical phenotype, and outcomes of a monogenic lupus cohort.
**Methods**
We evaluated all genetically confirmed monogenic lupus patients across Türkiye and compared clinical, laboratory, treatment, and outcome data between groups defined by pathogenic variants in distinct pathways.
**Results**
A total of 88 patients were included. Complement pathway deficiencies were identified in 28 patients (31.8%). Defects in nucleic acid metabolism and clearance constituted the largest subgroup in 36 patients (40.9%), predominantly involving DNASE1L3 and TREX1. Type I interferon signaling defects were detected in 6 patients (6.8%), including DDX58, IFIH1, ADAR, IRF5, and IRAK1. Immune regulation and lymphocyte signaling defects were identified in 17 patients (19.3%), involving genes associated with immune tolerance and JAK–STAT dysregulation: ACP5, PRKCD/PRKCQ, STAT3 gain-of-function, SOCS1, SHOC2, ZAP70, RAG1, HDAC7, FOXP3, and RELA. Consanguinity was present in 56 (63.6%), and positive family history in 34 (38.6%). ANA was positive in 66 (75%), anti-dsDNA in 33 (37.5%), and hypocomplementemia in 56 (63.6%). Mucocutaneous manifestations were the most prevalent clinical feature, with acute cutaneous involvement predominating (61.3%), followed by hematologic (44.3%), articular (37.5%), and renal involvement (35.2%). Lupus nephritis occurred in 31 patients (35.2%), with class IV nephritis being the most frequent histopathological subtype (32%). Neurological manifestations not captured by the ACR nomenclature for neuropsychiatric SLE were observed in 18 patients (20.4%), including spasticity, Babinski sign, hyperreflexia, developmental delay, and, especially, intracranial calcifications, which were detected in 13.6%. Vasculopathy/vasculitis-related features were common, including Raynaud phenomenon ($n$=22, 25%), chilblain lesions ($n$=18, 20.4%), livedo reticularis ($n$=18, 20.4%), and palmar–plantar erythema ($n$=19, 21.5%). Most patients ($n$= 77, 87.5%) required corticosteroid therapy. JAK inhibitors were used in 29 patients (33%); response was complete in 3, partial in 21, and absent in 5, however 2 patients developed severe infections. At the last follow-up, 47 patients (53.4%) had a PedSDI score of at least 1, indicating substantial cumulative organ damage. Seven patients died during follow-up.
**Conclusion**
This nationwide monogenic lupus cohort demonstrates atypical neurological and vasculopathy/vasculitis-related manifestations and a high burden of cumulative organ damage.

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**Disclosure of interest**
None declared.

## PT043

### Interferon-ALPHA and neopterin in cerebrospinal fluid: significant diagnostic and activity biomarkers for neuropsychiatric juvenile lupus

O. Bouvard[1], V. Bondet[2], R. Lhote[3], M. Labouret[4], L. Seabra[5], A. Alimi[6], M. Aubart[7], B. Bader-Meunier[1], C. Ballot-Schmit[8], J.-F. Benoist[9], G. Dingulu[4], C. Dumaine[4], A. Faye[4], A. Felix[4], B. Fournier[1], M.-L. Frémond[1], V. Hentgen[10], R. Khellaf[11], T. Kwon[12], A.-S. L'Honneur[13], J. Maquet[14], U. Meinzer[4], A.-S. Parentelli[15], C. Parmentier[16], P. Quartier[1], F. Renaldo[17], C. Vinit[4], O. Walter[14], P. Ellul[18], D. Duffy[2], Y. J. Crow[5], I. Melki[19,*] on behalf of French ASCLEPIOS network 'Alliance for SystemiC Lupus: Evaluation of Paediatric neuropsychiatric Involvement, Outcomes and Subtypes'
[1]Immunology and Rheumatology Unit - Necker hospital, APHP; [2]Pasteur Institute, Paris; [3]Clinical research unit - Avicenne, APHP, Bobigny; [4]General Paediatrics and Paediatric Internal Medicine - Robert Debré hospital, APHP; [5]Imagine Institute; [6]Haematology depatment - Trousseau hospital; [7]Paediatric neurology department - Necker hospital, APHP, Paris; [8]Paediatrics, CH Besançon, Besançon; [9]Metabolic Biochemistry - Necker hospital, APHP, Paris; [10]General Paediatrics - CEREMAIA - Versailles hospital, Versailles, Versailles; [11]Paediatrics, CHU Reims, Reims; [12]Paediatric Nephrology - Robert Debré hospital; [13]Virology - Cochin hospital, APHP, Paris; [14]Internal Medicine, CHU Toulouse, Toulouse; [15]Paediatrics, CHU Reunion, La Reunion; [16]Paediatric nephrology - Trousseau hospital; [17]Paediatric Neurology - Trousseau hospital; [18]Child Psychiatry - Robert Debré Hospital; [19]General Paediatrics Rheumatology Unit - Trousseau Hospital, APHP, Paris, France
**Correspondence:** I. Melki
*Pediatric Rheumatology 2026*, **24(S1):**PT043

**Introduction**
Juvenile systemic lupus erythematosus (j-SLE) can be complicated by neuropsychiatric involvement (j-NPSLE), with a severe prognosis, and no validated biomarker.
**Objectives**
This study aimed to evaluate IFN-α and neopterin in cerebrospinal fluid (CSF) as potential j-NPSLE diagnostic and activity biomarkers.
**Methods**
This multicentre retrospective study included patients with j-SLE, who met the EULAR/ACR 2019 criteria and underwent a lumbar puncture with SIMOA-based determination of IFN-α in the CSF from 2012 to 2026. The j-NPSLE status, active or inactive, was defined by a multidisciplinary paediatric team according to the ACR 1999 nomenclature and the DSM-5. Serum and CSF IFN-α Simoa assays, CSF neopterin, as well as clinical, radiological and therapeutic data were collected. Intergroup comparisons and longitudinal analyses were performed.
**Results**
A total of 67 patients presenting with neuropsychiatric manifestations were included: 49 j-NPSLE and 18 SLE controls without neuropsychiatric involvement retained, for a total of 104 CSF samples. During the first active episode, j-NPSLE patients had significantly higher levels of CSF IFN-α (458 vs. 42.26 fg/mL; $p = 0.0019$), serum IFN-α (5,423.9 vs. 968.6 fg/mL; $p = 0.0148$) and neopterin in the CSF (55 vs. 23.4 nmol/L; $p = 0.0033$). In the inactive phase in j-NPSLE patients, these rates were significantly lower: CSF IFN-α 31.5 fg/mL ($p = 0.0068$) and CSF neopterin 32.6 nmol/L ($p = 0.002$), but were not significant for serum IFN-α 1645.0 fg/mL ($p = 0.1953$). In the year following specific treatment, IFN-α and neopterin levels decreased significantly in the CSF, in contrast to serum IFN-α levels ($p = 0.055$), which suggests that CSF biomarkers are more specific.
**Conclusion**
The SIMOA-IFN-α in CSF and neopterin are promising biomarkers for the diagnosis and monitoring j-NPSLE. These results also support the involvement of the type I interferon pathway in the physiopathology of j-NPSLE and suggest potential targeted therapeutic approaches.
**Trial registration identifying number**
All samples were collected with informed signed consent by the parent or legal representant. The study was approved by the "Comité de protection des personnes Ile de France II" and the French advisory committee on data processing in medical research (ID-RCB/EUDRACT: 2014-A01017-40 and biocollection under the code DC2023-5445).

**Disclosure of interest**
None declared.

## PT044

### Investigation of inflammatory cell death PANoptosis pathway activation in patients with juvenile systemic lupus erythematosus

A. Safarova[1], S. Ozdemir Cicek[2,3,*], E. Cevikyigit[4], V. Cinar[4], A. Pac Kisaarslan[2,5], Z. Hamurcu[4,5], M. H. Poyrazoğlu[2,5]
[1]Pediatrics; [2]Pediatric Rheumatology; [3]Department of Molecular Biology and Genetics, Gevher Nesibe Genome and Stem Cell Institute; [4]Medical Biology; [5]Genome and Stem Cell Center (GENKÖK), Erciyes University Faculty of Medicine, Kayseri, Türkiye
**Correspondence:** S. Ozdemir Cicek
*Pediatric Rheumatology 2026*, **24(S1):**PT044

**Introduction**
Juvenile systemic lupus erythematosus (jSLE) is a multisystemic autoimmune disease characterized by inflammation triggered by autoantibodies and immune complexes. Programmed cell death induced by activation of the innate immune response has been reported to play a significant role in the development of autoimmune diseases. PANoptosis, a form of inflammatory cell death involving the coordinated activation of pyroptosis, necroptosis, and apoptosis pathways, has been proposed as one of the key regulators of inflammatory cell death in autoimmune disorders.
**Objectives**
In this study, we aimed to investigate the serum levels of key proteins involved in the PANoptosis pathway and to evaluate their associations with clinical findings in patients with jSLE.
**Methods**
A total of 29 patients with jSLE followed at the Erciyes University Pediatric Rheumatology Clinic and 31 age- and sex-matched healthy controls were included in this study. Demographic, clinical, and laboratory data were obtained from patient records. Peripheral blood samples were collected from patients and healthy volunteers, and the protein levels of NLRP3, TNFAIP3, CASP1, PARP1, GSDMD, CASP8, MLKL, ZBP1, RIPK1, and RIPK3, which are considered molecular key regulators of PANoptosis, were measured using the ELISA method.
**Results**
Of the 29 patients with jSLE, 86.2%($n$=25) were female, and the median age was 16 years (min–max:7–19). The median disease duration was 43 months (1–108). Median SLEDAI scores were 11(3–37) at diagnosis and 2(0–16) at the time of the study. Compared with healthy controls, patients with jSLE had significantly higher levels of the pyroptosis- and necroptosis-related proteins NLRP3, CASP1, GSDMD, PARP1, ZBP1, RIPK3, RIPK1, and MLKL (all $p<0.05$), whereas no increase was detected in the apoptosis-related proteins TNFAIP3 and CASP8. Strong and statistically significant positive correlations were identified among the PANoptosis pathway biomarkers evaluated in our study. No significant associations were identified between PANoptosis-related biomarkers and renal, cutaneous, gastrointestinal, hematological, or musculoskeletal involvement. Anti-dsDNA levels showed significant positive correlations with CASP1, NLRP3, PARP1, ZBP1, RIPK1, GSDMD, CASP8, and MLKL levels. The strongest correlation was observed between anti-dsDNA and ZBP1 levels (Spearman's rho: 0.815, $p<0.001$).
**Conclusion**
Our study demonstrated marked activation of the pyroptosis and necroptosis pathways within the PANoptosis pathway in patients with jSLE, whereas no concomitant increase was observed in the apoptosis pathway. These findings suggest that, in the chronic interferon-dominant inflammatory milieu of jSLE, CASP8-mediated apoptosis may be suppressed, leading to a shift in cell death programming toward the more immunogenic necroptotic and pyroptotic pathways.

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**Disclosure of interest**
None declared.

## PT046

### Improving damage assessment in childhood-onset SLE: insights from item generation and reduction in the revised damage index

H. I. Brunner[1,*], S. M. Al-Mayouf[2], S. A. Appenzeller[3], M. S. Klein-Gitelman[4], D. M. Levy[5], T. Miyamae[6], D. S. Grewal[7], E. Smith[8], A. Migowa[9], T. Avcin[10], J. Blair[11], S. I. Swauger[12], J. Park[13], J. I. Buie[14], B. Kundakci [15], C. A. A. Silva[16], I. Bruce[17] on behalf of for the SDI Revision Literature Review Groups, SDI Revision Delphi Panel, SDI Revision Item Reduction Committee, SDI Revision Clinical Domain Groups.
[1]University of Cincinnati, Cincinnati Children's Hospital, Cincinnati, USA; [2]King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia; [3]Pediatrics, Universidade Estadual de Campinas, Campinas, Brazil; [4]Pediatrics, Northwestern University and Lurie Children's Hospital, Chicago, USA; [5]Pediatrics, Hospital for Sick Children and University of Toronto, Toronto, Canada; [6]Pediatric Rheumatology, Tokyo Women's Medical University, Tokyo, Japan; [7]Rheumatology, Children's Hospital at Westmead, Sydney, Australia; [8]Pediatric Rheumatology, University of Glasgow School of Infection and Immunity, Glasgow, UK; [9]Pediatrics, The Aga Khan University Hospital, Nairobi, Kenya; [10]Allergology, Rheumatology and Clinical Immunology, University of Ljubljana, Ljubljana, Slovenia; [11]Pediatrics, Alder Hey Children's NHS Foundation Trust, Liverpool, UK; [12]Pediatrics, Cincinnati Children's Hospital, Cincinnati, USA; [13]Pediatrics, Hospital for Sick Children and University of Toronto, Liverpool, UK; [14]Lupus Foundation of America, New York, USA; [15]Rheumatology, University of Manchester, Manchester, UK; [16]Pediatrics, Universidade de São Paulo, São Paulo, Brazil; [17]Medicine, University of Belfast, Belfast, UK
**Correspondence:** H. I. Brunner
*Pediatric Rheumatology 2026*, **24(S1):**PT046

**Introduction**
The Systemic Lupus International Collaborating Clinics/ American College of Rheumatology Damage Index for Systemic Lupus Erythematosus (SDI) is a robust instrument, but has recognised limitations.
**Objectives**
To describe the generation and reduction of revised SDI items to define a modern construct of damage that address current gaps in childhood-onset SLE (cSLE) damage assessment.
**Methods**
Item generation involved a literature review and Delphi exercises of 146 international SLE experts and patient representatives from 35 countries. Paediatric-specific items were proposed and iteratively evaluated through Delphi rounds and expert review, including paediatric experts. Item reduction was undertaken through two additional Delphi rounds, with removal of redundant or duplicate items, those not reflecting damage or were excessively rare, or lacked feasibility of measurement. Experts assigned to clinical domain groups refined the remaining items and, where appropriate, proposed severity gradations.
**Results**
The literature review identified a *Growth & Development* domain relevant to cSLE. During item generation, paediatric-specific items included growth failure, non-traumatic long bone fractures in children, delayed puberty, reduced final height, disordered puberty sequences (e.g. reaching menarche without/with minimal/ less than expected growth spurt, breast or pubic hair development, and limb leg discrepancy. All items were evaluated through the Delphi exercise, followed by review by 14 pediatric experts. Limb length discrepancy was removed due to rarity and limited relevance to the current damage construct. Remaining items were clustered into **(A)** growth failure/reduced final height and **(B)** Delayed puberty and disordered puberty. Pubertal delay is common in chronic paediatric disease, and is associated with reduced bone mineral density, reduced final height (item A), and psychological distress. The first part is captured by item A and the latter is not part of the SDI damage construct. Pubertal arrest (primary, failure to progress) is exceedingly rare in cSLE and thought to be better represented other SDI domains (gonadal dysfunction, osteoporosis). Thus the organ specific group decided to remove item B. Item A was retained and the group defined it as "*Final height lower than the mid-parental range or less or equal to 2SD under the mean race/sex-adjusted height as per national growth charts or the WHO growth chart when measured first at age 18 years and again at or after age 20 years*."
**Conclusion**
The revised SDI incorporates developmentally relevant considerations and refines paediatric damage assessment, addressing key limitations of the original SDI to capture organ damage across the life course.

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**Disclosure of interest**
None declared.

## PT047

### Integrating domains to define patient-level response in a novel multi-domain outcome measure for SLE clinical trials across the lifespan: results of an international consensus study

H. I. Brunner[1,*], K. Connelly[2], R. Koehlmeyer[3], Y. Hao[3], S. S. Kamphuis[4], S. Appenzeller[5], A. A. Aggarwal[6], L. Arkin[7], E. A. Ogbu[1], S. Wenderfer[8], S. D. Marks[9], C. Scott[10], A. Migowa[11], J. I. Buie[12], J. Maller[13], D. Tremarias[14], N. Black[15], S. Nagamori[16], S. Gydesen [14], E. F. Morand[3] on behalf of on behalf of the TRM-SLE Taskforce
[1]Pediatrics, Cincinnati Children's Hospital & University of Cincinnati, Cincinnati, USA; [2]Monash University School of Clinical Sciences at Monash Health & Monash Medical Centre, Mebourne; [3]Medicine, Monash University School of Clinical Sciences at Monash Health & Monash Medical Centre, Clayton, Australia; [4]Pediatrics, Sophia Children's Hospital, Erasmus University Medical Center, Rotterdam, Netherlands; [5]Medicine & Pediatrics, Universidade Estadual de Campinas, Campinas, Brazil; [6]Clinical Immunology & Rheumatology, Sanjay Gandhi PG Institute of Medical Sciences, Lucknow, India; [7]Dermatology, School of Medicine and Public Health, Madison, USA; [8]Pediatrics, BC Children's & University of British Columbia, Vancouver, Canada; [9]Pediatric Nephrology, GOSH, London, UK; [10]Pediatrics, Children's Hospital of Eastern Ontario, Ottawa, Canada; [11]Pediatrics, The Aga Khan University Hospital, Nairobi, Kenya; [12]The Lupus Foundation of America, New YOrk; [13]Genentech, Inc, South San Francisco, USA; [14]Lupus Europe, Brussels, Belgium; [15]Public Health, UT Southwestern, Houston, USA; [16]National Collagen Disease organization, Tokyo, Japan
**Correspondence:** H. I. Brunner
*Pediatric Rheumatology 2026*, **24(S1):**PReS26-ABS-1061

**Introduction**
Treatment Response Measure for Systemic Lupus Erythematosus (TRM-SLE) is a novel multi-domain clinical outcome assessment for use within a primary efficacy endpoint in SLE randomised clinical trials and includes adolescents (aged 12+ years) and adults. The domains included in TRM-SLE are: mucocutaneous (rash, alopecia, mucosal ulcers), arthritis, haemolytic anaemia, nephropathy, serositis and thrombocytopenia, with thresholds for entry, clinically meaningful and complete response defined for each domain.
**Objectives**
To reach consensus of how to integrate domain-level responses when classifying overall treatment responders within a novel SLE clinical trial endpoint, while considering the potential for worsening in some domains.
**Methods**
An international Taskforce comprising 83 adult/pediatric lupus clinical experts, 27 patient/patient organisation representatives, 43 other experts including industry partners, and regulatory specialists participated in Delphi and nominal group technique (NGT) exercises to reach consensus on domain integration methods and 'no worsening' definitions. Pediatric experts participated at all levels of this methodological work. For *domain integration*: : **(1)** candidate methods were nominated via an online survey and shortlisted via an online Delphi process, considering five measurement characteristics: 'meaningfulness', 'appropriateness', 'feasibility', 'sensitivity', and 'specificity.' Consensus was defined

as a score ≥ 7 (1-9 scale) for >3 characteristics by ≥70% of respondents. **(2)** Shortlisted integration methods were then ranked via nominal group technique (NGT) in online surveys supported by structured discussions. *To define 'no worsening'*, a similar nomination process followed by NGT ranking was used.

**Results**

Six of 17 candidate domain integration methods achieved the pre-set threshold for consensus. After two rounds of NGT discussion and ranking, an ordinal multi-component method received the highest ranking, incorporating (i) Complete response (TRM-100): clinically meaningful improvement in all domains active at baseline; (ii) Partial response (TRM-1): clinically meaningful improvement in >1 domain active at baseline; and (iii) No response. Alternative binary integration methods (TRM-100 alone, and TRM-50: clinically meaningful improvement in at least half the domains active at baseline). For 'no worsening', of the 17 nominations the highest-ranking measure to define 'no worsening' was based Physician Global Assessment (PGA, 0-3) [3], defined as 'No ≥10% (0.3) from baseline'.

**Conclusion**

Consensus was reached for a method to integrate responses across multiple active TRM-SLE domains, alongside a harmonised 'no worsening' definition, to support classification of patient-level response in a TRM-SLE-based trial endpoint. Importantly, this lifecourse framework, spanning pediatric and adult rheumatology, provides a foundation for more consistent outcome assessment in future SLE trials that include adolescents. Further validation is underway.

**Trial registration identifying number**

The TRM-SLE project is an academia-industry-patient collaboration funded by a multi-company consortium: Abbvie, Amgen, AstraZeneca, Biogen, Bristol-Myers Squibb, Eli Lilly, Genentech, GlaxoSmithKline, Janssen, Merck KGaA, Novartis, Takeda and UCB.

**Disclosure of interest**

None declared.

## PT048

### Lupus anticoagulant as an independent predictor of chronic damage accrual in childhood-onset systemic lupus erythematosus

G. Olivieri[1,*], V. Messia[1], E. Marasco[1], C. Bracaglia[1], F. De Benedetti[1]
[1]Division of Rheumatology, ERN-RITA Centre, IRCCS Bambino Gesù Children's Hospital, Rome, Italy
**Correspondence:** G. Olivieri
*Pediatric Rheumatology 2026*, **24(S1):**PT048

**Introduction**

Childhood-onset Systemic Lupus Erythematosus (cSLE) is characterized by a more aggressive clinical course and a higher rate of organ damage compared to adult-onset disease.

**Objectives**

The aim of this study was to evaluate the prevalence and kinetics of chronic damage accrual in a longitudinal cohort of cSLE patients and to identify predictors of chronic organ damage.

**Methods**

This study analysed a cSLE cohort (SLICC 2012 criteria) followed in a single tertiary paediatric rheumatology centre in Italy between 2012 and 2026. Clinical and laboratory data, including SLICC/ACR Damage Index (SDI) scores, were collected at baseline and >3 times annually. Statistical analysis was performed using R software. The probability of remaining damage-free (SDI = 0) over time was estimated using Kaplan-Meier survival curves. The time to achieve complete clinical remission (cCR-0) was compared between patients with and without damage using the log-rank test. Predictors of damage were identified through univariate logistic regression, with significant variables entering a multivariate model adjusted for disease duration.

**Results**

The cohort ($n$=40; 75% female; 88% Caucasian) had a median age at onset of 13.5 years (IQR 12.2–15.4). Patients were followed for a median of 3.71 years (IQR 2.0–6.44). At the last visit, the median SDI score was 0 (IQR 0–1). Chronic damage (SDI$^3$ 1) was accrued by 17 patients (42.5%), with a mean SDI of 0.8 ±1.26 and a maximum score of 5. Analysis of damage-free survival revealed an early and progressive decline in the probability of remaining damage-free. While 90% of patients remained damage-free within the first year of disease, this probability decreased significantly to 64.4% (95% CI: 50.5%–82.2%) at the 2-year mark ($n$=21 at risk). By 5 years, the damage-free survival rate settled at 53.4%. Notably, patients who remained damage-free (SDI = 0) achieved complete clinical remission (cCR-0) significantly faster and in a higher proportion compared to those who developed damage ($p$ = 0.047). Comparative analysis identified Lupus Anticoagulant (LAC) as the primary driver of chronic damage. LAC prevalence was significantly higher in patients with damage compared to those without ($p$=0.030), and its cumulative probability over time was markedly greater in the damage group ($p$=0.015). Other markers, including anti-dsDNA, hypocomplementemia, and baseline SLEDAI-2K, showed no significant association. In univariate analysis, LAC was the sole predictor of damage (OR 5.93; 95% CI: 1.27–27.71; $p$=0.0238). This independence was confirmed in multivariate analysis adjusted for disease duration, where LAC remained a robust predictor (OR 5.42; 95% CI: 1.13–25.89; $p$=0.0342). Notably, LAC showed a strong, selective association with the cardiovascular damage domain (OR 16.00; $p$=0.0204). Despite a low prevalence of cardiovascular events (12%), Fisher's Exact Test confirmed a significant pathological link ($p$=0.015; OR 14.58).

**Conclusion**

This study highlights that chronic damage is a frequent and early occurrence in cSLE, with nearly half of the cohort accruing damage within the first five years of disease. LAC emerged as the primary independent predictor of damage, outperforming traditional biomarkers and proving essential for risk stratification. These results support the inclusion of LAC as a critical biomarker for risk stratification.

**Disclosure of interest**

G. Olivieri: None declared, V. Messia: None declared, E. Marasco: None declared, C. Bracaglia Consultant with: SOBI, Novartis, F. De Benedetti Consultant with: Abbvie, SOBI, Novimmune, Novartis, Roche, Pfizer.

## PT049

### Real-world experience with B-cell targeted therapies in juvenile SLE: an interim analysis from the PReS Lupus Working Party cohort

G. Kavrul Kayaalp[1,*], N. Akay[2], M. O. Erkan[3], M. C. Felix Rodrigues[4], N. Kifer[5], H. Ilgaz Tüzen[6], S. Ayduran[7], E. Altıok Özçelik[8], N. Gürbüz[9], O. D. Tosun[10], H. M. Kaçmaz[11], G. Sümer[12], S. Şener[13], R. İşgüder[13], O. Akgün[1], N. Şahin[14], H. E. Sönmez[14], S. Demir[10], A. Balat[11], G. Aksu[9], Z. Ekici Tekin[8], S. Türkuçar[7], S. Yüksel[7], S. S. Kılıç[12], B. Kasap Demir[6], M. Jelusic[5], A. Rodrigues Fonseca[4], S. Şahin[2], S. Ozen[3], O. Kasapcopur[2], N. Aktay Ayaz[1] on behalf of PReS Lupus Working Party
[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine; [2]Pediatric Rheumatology, Istanbul University-Cerrahpaşa Faculty of Medicine, Istanbul; [3]Pediatric Rheumatology, Hacettepe University Faculty of Medicine, Ankara, Türkiye; [4]Pediatric Rheumatology, Federal University of Rio de Janeiro, Rio de Janeiro, Brazil; [5]Pediatric Rheumatology, University of Zagreb School of Medicine, Zagreb, Croatia; [6]Pediatric Rheumatology, Izmir City Hospital, Izmir; [7]Pediatric Rheumatology, Pamukkale University Faculty of Medicine, Denizli; [8]Pediatric Rheumatology, Bilkent City Hospital, Ankara; [9]Pediatric Rheumatology, Ege University Faculty of Medicine, Izmir; [10]Pediatric Rheumatology, Eskişehir Osmangazi University Faculty of Medicine, Eskişehir; [11]Pediatric Rheumatology, Gaziantep University Faculty of Medicine, Gaziantep; [12]Pediatric Rheumatology, Uludağ University Faculty of Medicine, Bursa; [13]Pediatric Rheumatology, Adana City Hospital, Adana; [14]Pediatric Rheumatology, Kocaeli University Faculty of Medicine, Kocaeli, Türkiye
**Correspondence:** G. Kavrul Kayaalp
*Pediatric Rheumatology 2026*, **24(S1):**PT049

**Introduction**

B-cell targeted therapies are increasingly used in juvenile systemic lupus erythematosus (jSLE), particularly in patients with severe or refractory disease. However, real-world pediatric data regarding patient selection, treatment response and safety remain limited.

**Objectives**

To evaluate the clinical characteristics, indications, treatment response and safety of B-cell targeted therapies in a real-world jSLE cohort.

**Methods**

This multinational, multicenter cohort study within the PReS Lupus Working Party included patients with jSLE with disease onset before 18 years of age. Data were collected retrospectively from medical records. Patients receiving B-cell targeted therapy were compared with randomly selected patients from participating centers not exposed to B-cell targeted therapy. Clinical phenotype, treatment indications, disease activity, outcomes and adverse events were analyzed.

**Results**
A total of 319 patients with jSLE were included. Median (IQR) age at disease onset was 12.0 (4.4) years, and median follow-up duration was 50 (50) months. Eighty-two patients (25.7%) received B-cell targeted therapy, corresponding to 86 treatment courses; 84 courses (97.7%) were rituximab and 2 (2.3%) were belimumab. Median (IQR) time from diagnosis to treatment initiation was 17 (45) months. Compared with untreated patients, those receiving B-cell targeted therapy had higher cumulative frequencies of renal involvement (56.1% vs. 40.9%, $p$=0.017), neurological involvement (23.2% vs. 11.8%, $p$=0.012), hemolytic anemia (54.9% vs. 32.7%, $p$<0.001), thrombocytopenia (47.6% vs. 31.2%, $p$=0.008), leukopenia (73.2% vs. 40.9%, $p$<0.001), low complement (91.5% vs. 72.6%, $p$<0.001) and anti-dsDNA positivity (91.5% vs. 75.9%, $p$=0.002). The leading indications were renal ($n$=43, 50.0%), hematological ($n$=34, 39.5%), and neurological involvement ($n$=15, 17.4%). Twenty patients (24.4%) received treatment due to multisystem involvement. Prior treatments included pulse methylprednisolone (83.7%), mycophenolate mofetil (64.0%), cyclophosphamide (51.2%), and intravenous immunoglobulin (53.5%). Median SLEDAI-2K decreased from 12.0 at treatment initiation to 4.0 at 6 and 12 months ($p$<0.001). At 12 months, 39.5% achieved LLDAS and 50.0% achieved on-treatment remission, while flares occurred in 30.2%. At last visit, 58.2% were in on-treatment remission and 1.2% in off-treatment remission. Adverse events occurred in 14.0%, most commonly infections (7.0%).
**Conclusion**
In this large multinational real-world jSLE cohort, B-cell targeted therapies—predominantly rituximab—were mainly used in patients with severe organ involvement and high immunological activity. Treatment was associated with sustained reduction in disease activity, with acceptable safety. These findings support B-cell targeted therapies as an effective and tolerable option for selected patients with severe or refractory jSLE.
**Disclosure of interest**
None declared.

## PT050

### Taking a life course approach to SLE trials: creation of the Adolescent–Adult Treatment Response Measure (TRM SLE)

E. M. Smith[1,2,*], K. Connelly[3], R. Koelmeyer[3], Y. Hao[3], S. Kamphuis[4], S. Appenzeller[5], A. Aggarwal[6], L. Arkin[7], E. A. Ogbu [8], S. E. Wenderfer[9], S. D. Marks[10], C. Scott[11], A. Migowa[12], J. Buie[13], J. Maller[14], D. Tremarias[15], N. Black[16], S. Nagamori[17], S. Gydesen[18], H. I. Brunner[19], E. F. Morand[3] on behalf of on behalf of the TRM-SLE Taskforce
[1]School of Infection and Immunity, University of Glasgow; [2]Paediatric Rheumatology, Royal Hospital for Children, Glasgow, UK; [3]School of Clinical Sciences, Monash University, Melbourne, Australia; [4]Paediatric Rheumatology, Sophia Children's Hospital, Erasmus MC, Rotterdam, Netherlands; [5]Faculty of Medical Sciences, State University of Campinas (UNICAMP), Campinas, Brazil; [6]Clinical Immunology and Rheumatology, Sanjay Gandhi Postgraduate Institute, Lucknow, India; [7]Department of Dermatology, University of Wisconsin - Madison, Madison; [8]Division of Rheumatology, Cincinnati Children's Hospital Medical Center/University of Cincinnati, Cincinnati, USA; [9]Division of Nephrology, University of British Columbia, British Columbia, Canada; [10]Paediatric Nephrology and Transplantation, Great Ormond Street Hospital for Children NHS Foundation Trust, London, UK; [11]Paediatric Rheumatology, Children's Hospital of Eastern Ontario, Ottowa, Canada; [12]Paediatric Rheumatology, Aga Khan University, Nairobi, Kenya; [13]Lupus Foundation of America, Washington; [14]Rheumatology & Nephrology, Genentech, San Francisco, USA; [15]Lupus UK, Cardiff, UK; [16]Lupus TRM patient partner, Utah, USA; [17]Japan Connective Tissue Diseases Association, Tokyo, Japan; [18]Lupus Europe, Copenhagen, Denmark; [19]Paediatric Rheumatology, Cincinnati Children's Hospital Medical Center/University of Cincinnati, Cincinnati, USA
**Correspondence:** E. M. Smith
*Pediatric Rheumatology 2026*, **24(S1):**PT050

**Introduction**
Legacy treatment response measures (e.g. SRI–4) were developed in adults but are often used as primary endpoints in paediatric SLE trials, despite lacking paediatric validation. In adult trials, these legacy measures are thought to have contributed to inconsistent or negative outcomes. A life–course appropriate measure incorporating paediatric perspectives, yet suitable for adult SLE trials, is needed.
**Objectives**
To develop a novel, fit–for–purpose clinical outcome assessment (COA) for SLE trials, applicable to individuals aged ≥12 years, that uses continuous, domainspecific measures to capture clinically meaningful treatment benefit and support regulatory approval of new therapies.
**Methods**
COA was developed through a multi–stage consensus process involving paediatric and adult lupus clinical experts, patients with life–course lupus experience, industry partners, and regulatory specialists. Targeted literature review informed protocol development. Structured discussion defined the conceptual framework, measurement goals and context of use. Modified Delphi methods identified key disease activity domains, while domain–specific working groups (including paediatric/adult rheumatologists), applied nominal group technique (NGT) to define domains, select appropriate instruments and thresholds for clinically meaningful activity and response. Combined Delphi-NGT methods were used to integrate domain responses and define worsening.
**Results**
A total of 125 paediatric and adult lupus experts, 27 patients/representatives with life–course lupus experience, and 46 additional contributors (industry/methodological experts) participated. There was unanimous agreement that the COA should measure **"**active immune mediated disease manifestations that impact on the patient and are modifiable by therapy to reduce or control disease activity", relevant to adults and adolescents (aged≥12) with active SLE in RCTs. Eight disease domains met consensus criteria based on importance to patients, appropriateness for detecting change, representation in active SLE, and measurability in trials (rash, mucosal ulcers, alopecia, arthritis, haemolytic anaemia, thrombocytopenia, nephropathy, serositis). Five domain–specific working groups (mucocutaneous, arthritis, haematology, nephropathy and serositis), including 81 paediatric/adult rheumatologists; patients and experts, undertook parallel NGT processes supported by systematic literature reviews (21,636 papers screened; 155 included). These groups defined how response should be measured in each domain, identifying suitable instruments and numerical thresholds for study entry, minimum clinically meaningful response, and complete response.
**Conclusion**
We present the first regulatory–oriented, consensus–derived multi–domain COA for SLE with life–course applicability. Validation studies are now needed to transform treatment response assessment in SLE.
**Disclosure of interest**
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## PT051

### Accuracy of whole-body MRI in a monocentric cohort of juvenile inflammatory myositis

I. Maccora[1,*], G. Carreras[2], L. Gatti[1], E. Marrani[1], M. Mastrolia[1], S. Romano[1], I. Pagnini[1], T. Giani[1], G. Simonini[1]
[1]Rheumatology Unit, ERN ReCONNET Center; [2]Biostatistic Unit, Meyer Children's Hospital IRCCS, Florence, Italy
**Correspondence:** I. Maccora
*Pediatric Rheumatology 2026*, **24(S1):**PReS26-ABS-1279

**Introduction**
Juvenile inflammatory myositis (JIM) is a chronic severe inflammatory disease of childhood, that affects mainly muscles and skin. Accuracy of current traditional diagnostic criteria by Bohan and Peter and EULAR/ACR classification criteria in JIM is not always reliable. Whereas whole-body MRI (wbMRI) is increasingly used in clinical practice.
**Objectives**
To evaluate the diagnostic accuracy of wbMRI compared with conventional diagnostic tools in JIM.

**Table 1 (Abstract PT051)** Sensibility, specificity, positive predictive value (PPV), negative predictive value (NPV) and accuracy and comparison using as reference MRI

| | Sensibility *p* value | Specificity | *p*-value | PPV | *p*-value | PNV | *p*-value | Accuracy | *p*-value |
|---|---|---|---|---|---|---|---|---|---|
| MRI | 96.3 (87.5, 100) - | 100 (100, 100) | - | 100 (100, 100) | - | 94.44 (81.82, 100) | - | 97.73 (92.68, 100) | - |
| CK | 70.97 (54.54, 86.21) 0.0070 | 100 (100, 100) | 2.0000 | 100 (100, 100) | 2.0000 | 65.38 (45, 82.76) | 0.0070 | 81.25 (68.75, 91.67) | 0.0070 |
| AST | 74.19 (58.33, 88.01) 0.014 | 82.35 (62.5, 100) | 0.0850 | 88.46 (75, 100) | 0.0850 | 63.64 (41.67, 82.61) | 0.0050 | 77.08 (64.58, 87.5) | 0.0010 |
| LDH | 76.67 (60.61, 90.91) 0.0280 | 70.59 (50, 90.91) | 0.0140 | 82.14 (66.67, 95.83) | 0.0140 | 63.16 (40, 84.62) | 0.0100 | 74.47 (61.7, 87.23) | 0.0000 |
| Aldolase | 82.61 (66.67, 96.16) 0.1000 | 94.12 (81.25, 100) | 0.7600 | 95 (83.33, 100) | 0.7600 | 80 (61.9, 95.45) | 0.1460 | 87.5 (76.74, 97.3) | 0.0470 |
| CMAS | 82.61 (65.22, 95.83) 0.0050 | 94.12 (80, 100) | 2.0000 | 95 (83.33, 100) | 2.0000 | 80 (60, 95.24) | 0.0050 | 87.5 (76.74, 97.37) | 0.0050 |
| MMT8 | 74.19 (58.33, 89.19) 0.0100 | 100 (100, 100) | 2.0000 | 100 (100, 100) | 2.0000 | 68 (50, 85.2) | 0.0100 | 83.33 (72.92, 93.75) | 0.0100 |

**Methods**
Ten-year case control study involving consecutive patients with a diagnosis of JIM according to the Bohan and Peter and EULAR/ACR criteria and including treating physician's opinion. Patients were included if they underwent a wbMRI at the diagnosis, matched with children who received wbMRI for musculoskeletal symptoms. Demographic, clinical, laboratory and functional assessment data were correlated with MRI scores (according to Malattia et al.) using R to assess the diagnostic performance of MRI (paired cluster bootstrap approach).
**Results**
31 JIM patients (16 females, 51.6%; median age at onset 8 years, range 2–15) and 17 controls (9 females, 52.3%; median age at symptom onset 9 years, range 2–13) were included. Among patients with JIM, 28 (90.3%) presented proximal muscle weakness, 21 (67.7%) myalgia, 27 (87.1%) heliotrope rash, 24 (77.4%) Gottron papules, 6 (19.4%) dysphagia, 6 (19.4%) calcinosis, 14 (45.2%) photosensitivity, 7 (22.6%) skin ulcers, and 6 (19.4%) fever. Considering the whole cohort at baseline, elevated CK was observed in 22 patients (45.8%), AST in 26 (54.2%), LDH in 28 (59.6%), and aldolase in 20 (50%), while abnormal CMAS and MMT8 scores in 22 (45.8%) and 23 patients (47.9%), respectively. MRI findings consistent with myositis were detected in 26 patients (59.1%) in the overall cohort. MRI demonstrated the best diagnostic performance, with sensitivity of 96.3% (95% CI 87.5–100), specificity of 100% (95% CI 100–100), PPV of 100% (95% CI 100–100), NPV of 94.4% (95% CI 81.2–100), and overall accuracy of 97.7% (95% CI 92.68–100), outperforming all other evaluated tests (Table 1).
**Conclusion**
This study supports the use of MRI as a highly reliable non-invasive diagnostic tool in JIM, with potential applications also in disease monitoring.
**Disclosure of interest**
None declared.

## PT052

### A novel longitudinal protocol for monitoring calcinosis in juvenile dermatomyositis: serial ultra-low-dose whole-body CT

H. M. Natour[1,*], V. Mccarvell[2,3], A. Alkandari[1], K. Chiu[1], H. Clairman[1], I. Goh[1], E. Jones[1], K. Whitney[1], N. Shkumat[2], A. S. Doria[2], B. M. Feldman[1]
[1]Division of Rheumatology; [2]Department of Diagnostic Imaging, The Hospital for Sick Children, University of Toronto, Toronto; [3]Mount Allison University, Sackville, Canada
**Correspondence:** H. M. Natour
*Pediatric Rheumatology 2026,* **24(S1):**PT052

**Introduction**
Evaluating the overall impact of calcinosis and therapeutic response in juvenile dermatomyositis (JDM) remains challenging, as conventional monitoring relies on subjective physical examination or 2D radiography.

**Objectives**
We aimed to evaluate a recently described ultra-low-dose, whole-body CT (ULD-WBCT) protocol as a safe and objective tool for the longitudinal monitoring of the distribution and evolution of calcinosis in JDM.
**Methods**
This retrospective study included patients meeting Bohan and Peter criteria or the EULAR/ACR classification criteria for JDM at The Hospital for Sick Children (January 2000–January 2026) who developed calcinosis based on physical examination or previous imaging studies. Inclusion required a minimum of two serial non-contrast ULD-WBCT scans. Imaging data were analyzed to determine regional distribution, identify morphological subtypes, and assess longitudinal changes in the extent of calcinosis.
**Results**
Out of 147 patients currently followed at the SickKids JDM clinic, 29 (19.7%) have a history of calcinosis. We included three patients from this cohort who met the criteria of having at least two serial ULD-WBCT scans (two patients with three serial scans each and one with two scans.). The median age at diagnosis was 7.8 years (IQR 7.2–9.9), and two patients were anti-NXP2 positive. The median duration from JDM diagnosis to calcinosis onset was 17 months (IQR 8.5–22). Serial imaging revealed progressive increases in the size and distribution of deposits in all cases, documenting deep myofascial migration and spinal involvement not detectable by physical examination. These objective findings prompted treatment intensification in all three cases, including escalation to anifrolumab. The ULD protocol utilized a median effective dose of ~0.73 mSv, representing an approximately 90% reduction compared to standard protocols.
**Conclusion**
The ULD-WBCT protocol is a safe, objective, and superior tool for monitoring calcinosis trajectory in pediatric JDM. By identifying subclinical disease distribution and progression over time, it facilitates more informed and timely therapeutic transitions in refractory cases.

**References**

1. Cervantes BA, Rider LG, Chen MY, et al. Development of a computed tomography calcium scoring technique for assessing calcinosis distribution, pattern and burden in dermatomyositis. Rheumatology. 2023;62(5):1189-1196.

**Disclosure of interest**
None declared.

T053

## Metric properties and longitudinal performance of the Physician Global Assessment as single disease active measure in juvenile dermatomyositis

G. Filocamo[1,*], B. Feldman[2], H. Clairman[2], H. Natour[2], S. La Bella[3], A. Marino[4], M. Cattalini[5], F. Minoia[6], B. Magnusson[7], F. Zulian[8], T. Avcin[9], A. Ravelli[10], N. Ruperto[3] on behalf of Pediatric Rheumatology International Trial Organization

[1]Hospital for Sick Children and University of Milano (Italy); [2]Hospital for Sick Children, Toronto, Canada; [3] Reumatologia Pediatrica, Fondazione IRCCS San Gerardo dei Tintori, Monza; [4]ASST G. Pini-CTO, Milano; [5]University of Brescia and ASST Spedali Civili di Brescia, Brescia; [6]Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milano, Italy; [7]Karolinska University Hospital, Stockholm, Sweden; [8]University of Padua, Padua, Italy; [9]University of Ljubljana, Ljubljana, Slovenia; [10]IRCCS G Gaslini, Genova, Italy

**Correspondence:** G. Filocamo

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### Introduction

Accurate and feasible outcome measures are essential for assessing disease activity in juvenile dermatomyositis (JDM). Many commonly used instruments are time–consuming, require formal training, and rely on multiple quantitative components, limiting their feasibility in clinical settings. Consequently, their use is inconsistent across centers, leading to incomplete data capture in large registries, hampering longitudinal monitoring, and ultimately reducing the applicability of treat to target strategies. The Physician Global Assessment (PhGA) VAS is widely used in clinical practice and research; however, its metric properties as a standalone measure have not been comprehensively evaluated.

### Objectives

To assess the construct validity, responsiveness and discriminative ability of the PhGA as a single measure of disease activity in JDM.

### Methods

We performed a post hoc analysis of the PRINTO trial [1]. Construct validity was evaluated through exploratory factor analysis (EFA) and convergent validity, using Spearman's correlations with core set measures, DAS, and JDMAI (version comprising Skin VAS and MMT) at baseline visits. Responsiveness to change between baseline and the 6–month follow-up visit was assessed using (a) standardized response mean (SRM) and (b) relative efficiency (RE), compared with other outcome measures. Longitudinal performance was evaluated using linear mixed-effects models (LMMs). Discriminative ability with respect to treatment response was assessed using longitudinal mixed-effects models.

### Results

The JDM PRINTO trial included 139 children with newly diagnosed JDM from 54 centres, with follow-up visits up to 4 years. At baseline: median age 7.6 years, median age at onset 7.4 years, DAS total score 13/20, and JDMAI 22.5/40. At EFA, PhGA loaded strongly on the principal disease activity factor (loading 0.72), which accounted for 41.1% of the total variance, supporting its alignment with the underlying construct of disease activity. PhGA demonstrated moderate correlations with DAS (ρ = 0.60) and JDMAI (ρ = 0.65). PhGA correlated with all core set measures similarly to DAS and JDMAI, except for MMT and CMAS, where PhGA showed stronger correlations (PhGA/MMT ρ = -0.58; PhGA/CMAS ρ = -0.60). JDMAI showed a stronger correlation with cutaneous VAS (ρ = 0.73) compared to PhGA and DAS (ρ = 0.48 and 0.47, respectively). PhGA exhibited high responsiveness, comparable to DAS and JDMAI, and greater than laboratory markers. RE analyses confirmed that PhGA performed similarly to composite indices and better than laboratory and other outcome measures (Table 1). In longitudinal models, PhGA showed a significant improvement over time (β = -0.132 SD per visit, $p<0.001$), with trajectories comparable to DAS and JDMAI. PhGA, JDMAI and DAS demonstrated significant improvement over time; however, only PhGA showed a statistically significant visit-by-treatment interaction.

**Table 1 (Abstract T053)**

| Variable | SRM | Mean change | SD change | RE* |
|---|---|---|---|---|
| DAS total | -2.09 | -9.27 | 4.42 | 0.84 |
| PhGA VAS | -1.92 | -4.80 | 2.50 | 1.00 |
| JDMAI | -1.76 | -9.20 | 5.22 | 1.25 |
| ParGA VAS | -1.47 | -4.15 | 2.82 | 1.77 |

*RE values <1 indicate higher efficiency of the comparator compared with PhGA VAS

### Conclusion

PhGA VAS demonstrates good construct validity, high responsiveness, and robust longitudinal performance in JDM, with a greater sensitivity in discriminating differential treatment responses compared with JDMAI and DAS. These findings support its use as a simple and reliable measure of disease activity, particularly in settings where comprehensive composite indices may not be feasible.

### References

1. Ruperto N et al. Lancet 2016;387:671–8.

### Disclosure of interest

None declared.

PT054

## Pattern and predictors of mortality in juvenile dermatomyositis: a long term experience from a tertiary care centre in North India

S. Sarkar[1,*], P. Vignesh[1], A. Dod[1], R. K. Pilania[1], D. Suri[1], A. rawat[1], A. Gupta[1], S. Singh[1]

[1]Paediatrics, PGIMER Chandigarh, Chandigarh, India

**Correspondence:** S. Sarkar

*Pediatric Rheumatology 2026*, **24(S1):**PT054

### Introduction

Juvenile Dermatomyositis (JDM) is an autoimmune inflammatory myopathy in which severe pulmonary and vasculopathy continue to contribute to mortality despite advances in immunosuppressive therapy.

### Objectives

To identify potentially modifiable predictors associated with mortality in Juvenile Dermatomyositis.To evaluate clinical, laboratory, and antibody-related factors associated with poor outcomes.

### Methods

This retrospective observational study included 212 children with JDM managed over a 30-year period. Comparative analysis between survivors and non-survivors was performed using sPSS. Odds ratios (OR) calculated for significant predictors. ROC analysis and Cox proportional hazards regression were performed for laboratory cut-offs and independent predictors of mortality.

### Results

A total of 212 children with Juvenile Dermatomyositis (JDM) were evaluated , 22 patients (10.4%) died. Females constituted 59.1% (13/22) [male: female 1:1.4]. Mean age at onset was 7.1±3.5 years (2–16 years), mean delay in diagnosis was 3.2±1.8 months (2 weeks–8 months), was significantly associated with mortality ($p$=0.041, OR 2.87). Mortality ranged from 11.7% (8/68) during 1990–2010 to 9.09% (14/154) during 2010–2025. Myopathic JDM was observed in 81.8% (18/22), overlap syndrome was present in 18.2% (4/22). Nailfold capillary abnormalities were identified in 100% (22/22) ($p$=0.03, OR 3.8). Among antibody-tested patients ($n$=15), anti-MDA5 positivity was observed in 46.7% (7/15), while anti-NXP2 positivity was identified in 33.3% (5/15). RPILD/ILD accounted for 40.9% (9/22) of deaths, aspiration pneumonia (22.7%, 5/22), gastrointestinal vasculopathy/GI bleed (18.2%, 4/22). On multivariate Cox regression, RPILD/ILD (adjusted HR 3.21, $p$=0.004), anti-MDA5 positivity (adjusted HR 2.94, $p$=0.018), and ferritin >780 ng/mL (adjusted HR 2.76, $p$=0.026) independently predicted mortality. Ferritin demonstrated the best ROC performance for mortality prediction (AUC 0.84, sensitivity 81%, specificity 78%). ROC analysis demonstrated that ferritin had the best predictive performance for mortality [AUC 0.84, $p<0.001$] at a cut-off >780 ng/mL (81% sensitivity, 78% specificity). AST >140 U/L [AUC 0.75, $p$=0.003], CK-NAC >850 U/L [AUC 0.74, $p$=0.005], and LDH >920 U/L [AUC 0.72, $p$=0.008] also predicted mortality, while ALT >95 U/L showed lower predictive utility [AUC 0.66, $p$=0.041].

### Conclusion

Mortality in JDM remains driven predominantly by pulmonary and vasculopathic phenotypes, particularly RPILD and gastrointestinal vasculopathy. Delayed diagnosis, anti-MDA5 positivity, NFC abnormalities, severe myopathy, and elevated ferritin emerged as important predictors of poor outcome, highlighting potentially modifiable targets for early aggressive intervention.

### Disclosure of interest

None declared.

## PT055

### Tofacitinib for calcinosis cutis associated with juvenile dermatomyositis: an open-label single-arm study (TOFCAL study)

N. K. Bagri[1,*], P. Jain [1], S. L. Mohan[2], S. Kaushik[3], R. Deepak[3], D. Mishra[4], M. A. Khan[4], V. Gupta[5], P. Naranje[2], A. V. Ramanan[6], R. Lodha[1], M. Jana[2] on behalf of Pragati Jain1, Supraja Laguduva Mohan2, Shivani Kaushik3, Rakesh Deepak3, Dibyanshu Mishra4, MA Khan4, Vishal Gupta5, Priyanka Naranje2, A V Ramanan6, Rakesh Lodha1 ,Manisha Jana2, 1Department of Pediatrics, All India Institute of Medical Sciences, New D
[1]Pediatrics; [2]Radiodiagnosis and Interventional Radiology; [3]Transplant Immunology and Immunogenetics; [4]Biostatistics; [5]Dermatology, All India Institute of Medical Sciences, New Delhi, India; [6]Paediatric Rheumatology, Bristol Royal Hospital for children and Royal National Hospital for Rheumatic diseases, Bath, UK
**Correspondence:** N. K. Bagri
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**Introduction**
Calcinosis cutis (CC) is a form of dystrophic calcification and a disabling complication affecting nearly 20% and 40% of children with JDM. The management of calcinosis cutis is often refractory to various therapeutic agents. Interferons (IFNs) might play a pathogenic role in JDM and calcinosis cutis. Blocking IFN signaling as it utilizes the JAK-STAT pathway, with a JAK inhibitor, might be a therapeutic option for calcinosis cutis, as shown in a few anecdotal reports. This open-label single-arm study aimed to study the efficacy of tofacitinib in calcinosis cutis associated with JDM.
**Objectives**
Primary
To study the effect of tofacitinib on the burden of CC in children (2–18 years) with JDM, as assessed by Agatston score using a low-dose whole body CT scan (WBCT) at 24 ± 2 weeks follow-up.
Secondary
To evaluate the effect on CDASI, CMAS scales, steroid usage, IFN-α and β levels, and adverse events.
**Methods**
We conducted an open-label, single-arm study at Pediatric Rheumatology and Clinical Immunology services, Department of Pediatrics, All India Institute of Medical Sciences, New Delhi, India, from March 2024 to November 2025. Children between 2 and 18 years of age who were diagnosed with JDM / Juvenile Dermatomyositis-Systemic Scleroderma (JDM-SSc) overlap and calcinosis cutis were screened. Children with JDM and clinical/radiological evidence of CC were enrolled for this study. Those who received cyclophosphamide or rituximab in the preceding 6 months, on concomitant topical tacrolimus, with active tuberculosis, or hematological abnormalities were excluded. The study drug tofacitinib was administered orally as per the standard weight-based doses. Concomitant use of stable doses of steroids, methotrexate, and/or mycophenolate mofetil and/or IVIG was permitted.
**Results**
Twenty children (11 boys) with a mean (SD) age of 10.4 (3.6 years) completed the study. Eighteen children had CC associated with JDM, while two had JDM/SScl overlap. Median (IQR) duration of CC in the study population was 31 (19.5–54)31 months. The mean (SD) duration of follow-up for the study participants was 29.9 ± 1.4 weeks. There was a significant reduction in the Agatston score, median (IQR) at follow-up compared to baseline 4007(1616-55515) vs. 6349(3765-65255) $p = 0.012$, with a moderate effect size (d-robust -0.52). No serious adverse events were noted during the study period. None of the study participants experienced new calcifications, ulcerative lesions, or worsening of disease.
**Conclusion**: Tofacitinib, when added to standard therapy, was effective in reducing the Agatston score in children with calcinosis cutis associated with JDM. No major safety signals were observed during the study period.
**Declaration**: The abstract has been presented at the Global Conference on Myositis, 2026. The manuscript is currently under review.

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**Trial registration identifying number**
Clinical Trial Registry of India (REF/2023/12/076915).
**Disclosure of interest**
None declared.

## PT056

### Comparative transcriptomics of SIGLEC-1+ monocytes in (juvenile) dermatomyositis and systemic Autoimmune diseases reveals shared interferon activity, mitochondrial dysfunction, but also disease-specific effector programs

J. W. N. Marsden[1,*], S. R. Veldkamp[1], W. Armbrust[2], J. M. van den Berg[3], P. C. E. Hissink Muller[4], S. Kamphuis[5], E. Schatorjé[6], A. van der Kooi[7], J. Raaphorst[7], A. van Royen-Kerkhof[8], M. H. A. Jansen[8], F. van Wijk[1]
[1]Center for Translational Immunology, University Medical Center Utrecht, Utrecht; [2]Beatrix Children's Hospital, University Medical Center Groningen, Groningen; [3]Emma Children's Hospital, Amsterdam Medical Center, Utrecht; [4]Willem Alexander Children's Hospital, Leiden University Medical Center, Leiden; [5]Sophia Children's Hospital, Erasmus Medical Center, Rotterdam; [6]Amalia Children's Hospital, Radboud University Medical Center, Nijmegen; [7]Neurology, Amsterdam Medical Center, Amsterdam; [8]Wilhelmina Children's Hospital, University Medical Center Utrecht, Utrecht, Netherlands
**Correspondence:** J. W. N. Marsden
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**Introduction**
SIGLEC-1 expression on monocytes reflects type I Interferon (IFN) and disease activity in Juvenile and adult Dermatomyositis ((J)DM) and other autoimmune diseases.[1,2] We hypothesized that these monocytes represent pathogenic effector cells. To elucidate this, we performed bulk RNA sequencing of SIGLEC-1+ and SIGLEC-1− monocytes in a multi-disease cohort.
**Objectives**
Define the SIGLEC-1 signature in JDM monocytes during active disease and remission. Compare SIGLEC-1+ monocytes across IFN-driven autoimmune diseases.
**Methods**
The cohort included 12 JDM patients (12 onset – 9 follow-up), 6 DM patients, 8 patients with active juvenile Systemic Lupus Erythematosus (jSLE) or juvenile Mixed Connective Tissue Disease (jMCTD), and 11 healthy controls (HCs). RNA-sequencing was performed on sorted blood CD14+ SIGLEC-1+ and SIGLEC-1- monocytes. This study was approved by the medical ethics committee of the University Medical Center Utrecht (METC no. 15–191 and 07–025) and by the medical ethics committee of the Amsterdam University Medical Center (METC no. 2016_326). All patients and/or their parents gave written informed consent in accordance with the Declaration of Helsinki.
**Results**
Differential expression analysis of SIGLEC-1+ versus SIGLEC-1− monocytes in JDM at onset identified a program consistent with *CXCL10*-mediated IFN effector activation. In parallel, mitochondrial electron transport chain (ETC) complex I genes were downregulated. Comparison of JDM onset versus remission confirmed downregulation of complex I genes alongside IFN-driven activation in active disease. Notably, JDM remission samples showed normalization of inflammatory signatures, while ETC alterations persisted, with sustained downregulation of complex I genes and compensatory upregulation of complex III and IV genes. Across IFN-mediated diseases, SIGLEC-1+ monocytes shared a core program with upregulation of IFN-associated and activation-related genes, including *IFI27* and *ISG15*, alongside downregulation of ETC genes (*MT-ND1/3/5*, *MT-ATP6/8*). Contrasting the *CXCL10*-mediated IFN profile in JDM, SIGLEC-1+ monocytes in adult DM displayed chronic NF-κB-driven inflammation via *MAP3K8*/*TIM-3*, whereas in jSLE/jMCTD they were enriched for nucleic acid-sensing and TLR7-mediated IFN pathways.
**Conclusion**
JDM SIGLEC-1+ monocytes are characterized by an IFN-driven effector and tissue-homing program accompanied by ETC alterations involv-

ing complex I genes. While inflammatory signatures normalize during remission, persistent mitochondrial dysregulation suggests metabolic reprogramming despite clinical quiescence, potentially reflecting residual immune dysfunction or altered fitness. Across IFN-mediated diseases, SIGLEC-1+ monocytes share an IFN-associated activation profile but display distinct effector programs, supporting a context-dependent pathogenic role.

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**Disclosure of interest**
None declared.

## PT057

### Single-center experience of refractory cases of juvenile idiopathic inflammatory myopathies treated with JAK inhibitors

O. Kostareva[1], I. Nikishina[1,*], S. Arsenyeva[1], V. Matkava[1], A. Shapovalenko[1]
[1]V.A. Nasonova Scientific Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** I. Nikishina
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**Introduction**
Juvenile Idiopathic Inflammatory Myopathies (JIIM) are rare and heterogeneous autoimmune disorders. Treatment remains challenging due to the lack of approved targeted therapies for severe and refractory cases. Given their broad immunomodulatory effects, Janus kinase inhibitors (JAKi) emerge as a promising therapeutic strategy.
**Objectives**
We aimed to evaluate the effectiveness and safety of JAKi in a real-world pediatric JIIM cohort.
**Methods**
We analyzed a single-center cohort of 69 patients with JIIM (2010–2025). Within this group, 36 patients required biologic or targeted therapy, including 14 patients (15 prescriptions) treated with JAKi (tofacitinib or upadacitinib). We assessed clinical manifestations, laboratory markers (CK, transaminases, LDH), instrumental data, previous treatment history, and clinical outcomes.
**Results**
The cohort included 14 patients: 8 (57%) with JDM, 5 (36%) with overlap syndrome, and 1 (7%) with JPM. Median age at onset was 8.9 years (range 1.3–16.5). The mean duration of disease before the diagnosis verification was 7 months (1; 22). JAKi were initiated at a median disease duration of 52 months. Clinical features at baseline included myositis (100%), skin lesions (86%), arthritis (79%), dysphagia (29%), and calcinosis (43%) manifested on average 4.5 years after disease onset. Interstitial lung involvement was detected in 2 pts with overlap-syndrome. High ANA positivity was observed in 79% of pts. Levels of CK, ALT, AST and LDH were elevated in 80% of pts. All patients were pre-treated with glucocorticoids and methotrexate; 86% received IVIG. JAKi were used as first-line targeted therapy in 57% of cases, from them with overlap-syndrom – 3 pts, JDM with calcinosis – 3 pts, JPM and JDM – 1 pts each. 13 pts continue tofacitinib or upadacitinib, with median treatment duration of 19 months. One patient with refractory JDM and multiple calcinosis discontinued tofacitinib after 9 months because of insufficient efficacy and was successfully switched to upadacitinib. Under JAKi therapy we observed improvement of skin, muscle and articular manifestations, favorable dynamics of calcinosis in some cases, and steroid-sparing effect. Tolerance was good.
**Conclusion**
JAK inhibitors demonstrate good therapeutic potential in JIIM, including overlap syndromes and refractory cases. JAKi seem to be effective in controlling skin, muscle, and joint manifestations, while providing clinical benefits in managing calcinosis and interstitial lung involvement. Our expeirence suggest that JAKi are a viable option both as first-line targeted therapy and as a switching strategy for patients failing conventional treatments.
**Disclosure of interest**
None declared.

## PT058

### Assessment of clinical and laboratory markers of metabolic dysfunction and lipodystrophy in patients with juvenile dermatomyositis

V. Pandiarajan[1,*], A. S. Raghuvanshi[1], H. Kaur[1], J. Kabeerdos[1], N. Sachdeva[2], A. Rawat[1], A. Gupta[1]
[1]Pediatrics; [2]Endocrinology, Postgraduate Institute of Medical Education and Research, Chandigarh, India
**Correspondence:** V. Pandiarajan
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**Introduction**
Juvenile dermatomyositis (JDMS) is an autoimmune inflammatory myopathy strongly associated with cardiovascular and metabolic comorbidities, including lipid abnormalities and lipodystrophy. Understanding the relationship between adipokines, such as adiponectin, and metabolic health is crucial for managing long-term outcomes in these patients.
**Objectives**
The primary aim was to assess the clinical and laboratory markers of metabolic dysfunction and lipid profile changes in North Indian children with JDMS. Specific objectives included evaluating clinical markers such as body mass index (BMI) and lipodystrophy, assessing skinfold thickness, and measuring biochemical markers, including fasting blood glucose, HOMA-IR, fasting lipid profile, and serum adiponectin levels, compared with healthy controls.
**Methods**
This observational study, conducted at a tertiary-care referral centre in North India, involved 45 children with JDMS and 45 age- and gender-matched healthy controls. Comprehensive anthropometric measurements were recorded, including height, weight, BMI Z-scores, and triceps/subscapular skinfold thickness. Laboratory analysis included measuring fasting plasma glucose, serum insulin by ECLIA, lipid profiles, and serum adiponectin levels by ELISA. Statistical analysis was performed using R software, employing Wilcoxon rank-sum tests and Spearman's correlation.
**Results**
The study cohort of 45 children with juvenile dermatomyositis (JDMS) had a mean age at enrolment of 12.6 ± 4.8 years. The median age at diagnosis was 6.0 years (IQR 3.3–10.0), following a median delay in diagnosis of 6 months. Clinical lipodystrophy is evident in 51% of patients at enrolment. Median serum adiponectin levels were significantly lower in the JDMS group (7 µg/mL) compared to controls (16 µg/mL; $p < 0.001$). A significant positive correlation was observed between BMI Z-score and HOMA-IR ($\rho = 0.601$, $p < 0.001$), suggesting that increased relative body mass is a driver of insulin resistance in this population. Bivariate analysis revealed no statistically significant association between specific myositis autoantibodies and clinical lipodystrophy at enrolment. Additionally, multivariable logistic regression identified age at enrolment and duration of corticosteroid therapy as independent factors associated with higher insulin resistance.
**Conclusion**
Metabolic disturbances, insulin resistance, and adipose tissue abnormalities are prevalent in children with JDMS and are likely driven by cumulative disease burden and treatment exposure. Routine metabolic monitoring and strategies to minimise prolonged corticosteroid use may help mitigate long-term cardiometabolic risk.
**Disclosure of interest**
None declared.

PT059

**Juvenile dermatomyositis across diagnostic eras: treatment evolution and long-term outcomes in a 25-year cohort**

M. O. Erkan[1,*], H. Ercan Emreol[1], M. Besrek[2], Y. Bayindir[1], S. Oz Yildiz[3], V. Cam[1], D. Unal[1], O. Necipoglu Banak[1], H. D. Dara Kar[1], S. Ozkan[1], B. Talim[4], A. E. Yildiz[5], O. Basaran[1], Y. Bilginer[1], G. Haliloğlu[3], S. Ozen[1], E. Sag[1]
[1]Pediatric Rheumatology; [2]Pediatrics; [3]Pediatric Neurology; [4]Pediatrics, Pathology Unit; [5]Radiology, Hacettepe University, Ankara, Türkiye
**Correspondence:** M. O. Erkan
*Pediatric Rheumatology 2026*, **24(S1):**PT059

**Introduction**
Juvenile dermatomyositis (JDM) is a rare autoimmune myopathy with heterogeneous clinical phenotypes and variable long-term outcomes. Cumulative damage remains a major concern despite advances in diagnosis and treatment.
**Objectives**
To summarize a 25-year single-centre experience of JDM and to evaluate clinical characteristics, investigations, autoantibody profiles, treatment patterns and determinants of cumulative damage.
**Methods**
We retrospectively included 104 patients fulfilling the 2017 EULAR/ACR criteria (2000–2025) with ≥6 months follow-up. Clinical features, complications, investigations (EMG, MRI, biopsy), treatments and myositis-specific/associated antibodies were recorded. Diagnostic eras were defined as 2000–2007, 2008–2014 and 2015–2025. Damage was assessed using standardised and normalised MDI extent/extended scores and MyoDAM severity.
**Results**
The cohort showed female predominance (66.3%). Mean age at onset and diagnosis were 7.7±4.1 and 8.5±4.2 years; median follow-up was 52.6 months. Skin involvement (91.3%) and proximal muscle weakness (79.8%) were the most frequent presenting features. Autoantibody frequencies were comparable to previous reports, although phenotype associations differed. Calcinosis increased from 13.5% at diagnosis to 40.4% overall; skin ulceration from 3.8% to 12.5%. Nailfold capillaroscopy abnormalities (57.5%) were associated with calcinosis ($p$=0.031). Calcinosis frequency did not differ across eras ($p$=0.535), and time-to-calcinosis curves were similar (log-rank $p$=0.235). In multivariable analyses, diagnostic delay >6 months, chronic active course, dysphagia and cutaneous ulceration were independently associated with higher damage (adjusted $R^2$: 0.416 for MDI extent; 0.403 for MDI extended; 0.449 for MyoDAM severity).
**Conclusion**
Cumulative damage was primarily driven by delayed diagnosis, persistent disease course and severe vasculopathic/systemic features, underscoring the importance of early recognition and treat-to-target escalation to prevent long-term damage.
**Disclosure of interest**
None declared.

PT060

**Clinical correlates of interstitial lung disease patterns in juvenile dermatomyositis: a single-center cohort from India**

P. Kaur[1,*], S. Chandola[2], M. Jana[2], N. Bagri[1]
[1]Pediatrics; [2]Radiodiagnosis & Interventional Radiology, All India Institute of Medical Sciences, New Delhi, New Delhi, India
**Correspondence:** P. Kaur
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**Introduction**
Visceral organ involvement in juvenile dermatomyositis (JDM) and JDM-Systemic Sclerosis (SSc) overlap is poorly and variably described, particularly the extent and severity of pulmonary manifestations, sparing the MDA5 antibody-positive disease.
**Objectives**
This study aimed to review the clinical and radiological characteristics of interstitial lung disease (ILD) in children with JDM and JDM-SSc overlap. Secondly, to compare outcomes between the two groups.
**Methods**
We retrospectively reviewed the high-resolution computed tomography (HRCT) chest of 70 children with JDM and JDM-SSc enrolled in the pediatric rheumatology clinic (2016-April 2026). A dominant ILD pattern was assigned after review. The HRCT-based Warrick score was noted for severity assessment where applicable. Data was analysed using descriptive statistics.

**Table 1 (Abstract PT060)** .

| | Entire Cohort | JDM | JDM-SSc overlap | Comparison between children with JDM and JDM-SSc overlap | |
|---|---|---|---|---|---|
| **Variable** | ***N*=18/70** | ***N*=12/57** | ***N*=6/13** | **Difference (95% CI)** | ***P* value** |
| Age at onset, years, mean (SD) | 8.5 (4) | 8.9 (4.6) | 7 (2.9) | 1.9(-0.76-4.5) | *P*=0.1597 |
| ILD, % | 25.7 | 21.1 | 46.2 | 25.1 (-0.9-51.2) | *P* = 0.0637 |
| OP pattern | 44.4 | 58.8 | 16.6 | 42.2(12.3-58.8) | ***P* = 0.0064** |
| NSIP pattern | 27.7 | 33.3 | 16.6 | 16.7(-12.3-34.1) | *P* = 0.2400 |
| UIP pattern | 11.1 | 16.6 | 33.3 | 16.7(-5.6-44.3) | *P* = 0.1750 |
| Fibrotic lung changes | 16.6 | 8.3 | 33.3 | 25(3.4-52) | ***P* = 0.0161** |
| Warrick score, median (IQR) | 3.5 (2.7–6) | 3 (2–4) | 5 (4–10) | | 0.09047 |
| Pulmonary hypertension, % | 16.6 | 16.6 | 16.6 | - | - |
| Clinical course*, % | 61.1 | 66.6 | 50 | | |
| Stabilized | 22.2 | 16.6 | 33.3 | 16.6(-10.5-42.8) | *P* = 0.2653 |
| Improved | 16.7 | 16.6 | 16.6 | 16.7(-5.6-44.3) | *P* = 0.1750 |
| Worsened | | | | - | - |

OP, organizing pneumonia; NSIP, non-specific interstitial pneumonia; UIP, usual interstitial pneumonia

Warrick score was calculated where technically feasible

*Children with no worsening of the pulmonary function test, or escalation of immunosuppression was labelled as 'stabilized'; those with improvement in pulmonary function or de-escalation of immunosuppression were labelled as 'improved'; and clinical worsening or escalation in immunomodulation or mortality were labelled as 'worsened'

**Results**
Of 70, *n*=18 (25.7%) children had ILD: *n*=12/57 (21.1%) in the JDM subgroup and *n*=6/13 (46.2%) in the JDM-SSc overlap subgroup (Table 1), with a mean age of onset at 8.5(4) years. Organizing pneumonia was the commonest ILD pattern overall and in the JDM subgroup, whereas UIP predominated in children with JDM-SSc overlap. Fibrotic (irreversible) ILD was significantly more prevalent in the overlap (33.3%) than in the JDM subgroup (8.3%). ILD stabilized in *n*=11/18 (61.1%). One mortality was noted in the JDM subgroup due to rapidly progressive ILD in a child with antisynthetase syndrome.
**Conclusion**
ILD is a prevalent morbidity in children with JDM. Those with JDM-SSc overlap had a significantly higher propensity for irreversible fibrotic lung damage than JDM alone. Although statistically insignificant, there was a trend to earlier onset and worse Warrick score in children with overlap. Clinico-radiological monitoring of these high-risk children may help in circumventing irreversible loss of pulmonary function.

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**Disclosure of interest**
None declared.

## PT061

### MIR22HG expression and role in new-onset oligoarticular juvenile idiopathic arthritis

A. La Sala[1,*], F. Briasco[1], S. Pelassa[1], C. Artale[1], F. Raggi[1], C. Rossi[1], S. M. Orsi[2], F. Orso[3,4], D. Taverna[3], M. Gattorno[1], A. Consolaro[1], M. C. Bosco[1]
[1]Unit of Rheumatology and Autoinflammatory Diseases, IRCCS G. Gaslini Institute; [2]Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova, Genova; [3]Department of Molecular Biotechnology and Health Sciences, Molecular Biotechnology Center "Guido Tarone", Torino; [4]Department of Translational Medicine, University of Piemonte Orientale, Novara, Italy
**Correspondence:** A. La Sala
*Pediatric Rheumatology 2026*, **24(S1):**PT061

**Introduction**
Oligoarticular Juvenile Idiopathic Arthritis (OJIA) is the most common pediatric rheumatic disease, causing joint damage and disability. To date, its molecular pathogenetic mechanisms are poorly understood. Recent evidence suggests a possible role of the lncRNA MIR22HG in adult arthritides, but its involvement in OJIA has not yet been investigated.
**Objectives**
This study aimed to evaluate MIR22HG expression and contribution to OJIA pathogenesis. MIR22HG modulation was performed in an in vitro monocytic (Mn) cell model to evaluate its role in Mn functions.
**Methods**
CD14+ Mn were isolated from PBMCs and SFMCs from 30 new-onset OJIA patients and PBMCs from 20 age- and sex-matched control children (CTR). MIR22HG expression was assessed by RT-qPCR. miRNA profiling was carried out in both CD14+ and the Mn line, THP1 in which MIR22HG was either silenced by siRNA transfection (si-TPH-1) or overexpressed through lentiviral transduction (up-THP-1). Pathway and target gene analysis of MIR22HG-regulated miRNAs were also carried out. THP-1 cell viability, apoptosis, cytokine release, and STAT3 phosphorylation were evaluated.
**Results**
CD14+ cells in PBMCs from OJIA patients showed significantly lower MIR22HG levels than from CTRs, with good discriminatory power. Comparative miRNA profiling identified two miRNAs, miR-15a-5p and miR-142, that were upregulated in both patients' CD14+ cells and in si-THP-1 cells, while downregulated in up-THP-1 cells, suggesting they may represent specific MIR22HG targets. Bioinformatic analyses linked these two miRNAs to pathways involved in cell cycle, inflammation, apoptosis, and stress response, and identified STAT3 among their target genes. Functional in vitro assays in THP1 cells demonstrated reduced viability, altered cytokine release, and changes in STAT3 phosphorylation in si-THP1, whereas opposite effects were observed in up-THP1, indicating regulation of Mn inflammatory activity by MIR22HG.
**Conclusion**
These findings suggest that MIR22HG may play a protective anti-inflammatory role by regulating Mn viability and activity. Its downregulation in OJIA may represent a mechanism promoting inflammation, supporting its potential value as an early disease biomarker and therapeutic target.
**Disclosure of interest**
None declared.

## PT062

### Comparison of first-line treatment strategies in Still disease: JIR vs CLiPS

O. Koker[1,*], R. Naddei[2], S. Azevedo[3], K. Daghor-Abbaci[4], C. Girard[5], M. Jouret[6], K. Pateras[7], F. Aguiar[8], P. Mertz[9], F. Oliveira-Ramos[10], S. Georgin-Lavialle[11], D. Foell[12], K. Theodoropoulou[13] on behalf of JIR-CLiPS Network
[1]Pediatric Rheumatology, Marmara University Faculty of Medicine, Istanbul, Türkiye; [2]Translational Medical Sciences, University of Naples Federico II, Naples, Italy; [3]Pediatric Rheumatology, Unidade Local de Saúde da Região de Aveiro and Aveiro Rheumatology Research Center, Aveiro, Portugal; [4]Rheumatology and Immunology, University of Algiers, Bab El Oued University Hospital Center, Algiers, Algeria; [5]Rheumatology, University Hospital of Geneva, Geneva, Switzerland; [6]Pediatric Nephrology, Rheumatology, Mother and Children University Hospital, Hospices Civils de Lyon, Lyon, France; [7]Statistics, Athens University of Economics and Business, Athens, Greece; [8]Rheumatology, ULS São João and Faculdade de Medicina da Universidade do Porto, Porto, Portugal; [9]Pediatrics, Sorbonne University, Versailles Hospital, Le Chesnay; French National Reference Center for Autoinflammatory Diseases, Paris, France; [10]Pediatric Rheumatology, University of Lisbon, Lisbon, Portugal; [11]Internal Medicine, Sorbonne University, Tenon Hospital, AP-HP, DMU3ID, Paris, France; [12]Pediatric Rheumatology and Immunology, University Children's Hospital Münster, Münster, Germany; [13]Pediatric Rheumatology and Immunology, Lausanne University Hospital (CHUV) and University of Lausanne (UNIL), Lausanne, Switzerland
**Correspondence:** O. Koker
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**Introduction**
Treatment strategies in pediatric Still disease (SD) have evolved substantially over recent decades, particularly with the increasing availability of biologics. However, real-life treatment patterns remain heterogeneous across centers and countries. Comparative data between physician-preference-based approaches and real-world registry cohorts are limited.
**Objectives**
To describe first-line treatment strategies in pediatric SD, compare real-life patterns from the Juvenile Inflammatory Rheumatism(JIR) cohort with Clinical Practice Strategies(CliPS) data and identify clinical and demographic factors.
**Methods**
This retrospective observational study included physician-confirmed SD patients enrolled in the JIR cohort between 2017-2024,with baseline demographic, clinical and treatment data. First-line treatment strategies were categorized as:*n***on-steroidal-antiinflammatorydrugs(NSAIDs)**, **corticosteroids±NSAIDs**,**disease-modifying-antirheumaticdrugs(DMARDs; represented by methotrexate)±corticosteroids/NSAIDs** and **biologics±DMARDs/corticosteroids/NSAIDs**. Clinical presentation was classified as systemic-,articular-,serositis-associated phenotypes. Since CliPS survey reflects more recent practice, JIR–CliPS comparisons were restricted to JIR patients registered after 2020.
**Results**
The cohort included 412 patients, with a median age at diagnosis of 7 years. Systemic manifestations were present in 78%,articular in 60%,serositis in 12.6%. Corticosteroids and biologics were the leading first-line strategies in systemic-predominant(40% and 30%) and articular-predominant phenotypes(35% and 31%), respectively. Conversely, serositis-associated was mainly treated with corticosteroids(61%), with lower use of biologics(22%). Steroid use decreased from 50 to 54% in 1990–2000 s to 19% after 2020, while DMARD use declined from 29% to 7%. Conversely, biologic use increased from 6% in 2000s to 25% in 2010s and became the leading strategy after 2020(48%). In post-2020 systemic-predominant disease, biologics were similarly dominat in JIR and CliPS(46% vs. 45%) and steroid-based strategies were comparable(33% vs. 36%). NSAIDs were reported only in JIR(21%), whereas DMARD were reported in CliPS(11.3%). In the articular-predominant phenotype, biologics were more frequent in JIR than CliPS(42% vs. 27%), while DMARD-based strategies were reported only in CliPS(42%). NSAIDs were observed only in JIR(33%), whereas corticosteroid use was comparable(25% vs. 22%).
**Conclusion**
Treatment strategies in pediatric SD have shifted substantially toward biologic-based approaches over time, although important differences persist according to phenotype and cohort characteristics. While systemic phenotypes showed broadly similar patterns between JIR and CliPS, differences were more pronounced in articular disease, particularly regarding DMARD use. Ongoing analyses will explore the influence of country, income level, and biologic drug availability on therapeutic decision-making.
**Disclosure of interest**
None declared.

## PT063

### Assessment of physical and motor development in children with juvenile idiopathic arthritis - results before and after disease compared to national population

E. M. Tanko[1], L. Plusko[1], B. Leskosek[2], N. Emersic[1,3], T. Avcin[1,3], N. Toplak[1], M. Bizjak[1,3], G. Jurak[2], M. Zajc Avramovic[1,3,*]
[1]University Children's Hospital Ljubljana; [2]Faculty of Sport, University of Ljubljana; [3]Medical faculty, University of Ljubljana, Ljubljana, Slovenia
**Correspondence:** M. Zajc Avramovic
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**Introduction**
Juvenile idiopathic arthritis (JIA) is the most common chronic pediatric rheumatic disease and may significantly affect physical and motor development. Slovenia's SLOfit national physical fitness surveillance system is one of the world's most comprehensive and longest-running child fitness databases that assesses annualy the entire Slovenian population aged 6–19 years and provides standardized assessments of anthropometry, aerobic fitness, muscular strength, coordination, flexibility, and body composition.
**Objectives**
The aim of this study was to assess the physical and motor development of children with JIA according to disease activity and compare it with the healthy Slovenian pediatric population before disease onset and during different disease activity states.
**Methods**
A retrospective longitudinal observational cohort study was conducted in patients diagnosed with JIA and treated at the Department of Allergology, Rheumatology and Clinical Immunology, University Children's Hospital Ljubljana. Data from the Slovenian national surveillance system for physical and motor development of children and youth (SLOfit/Sports Educational Chart (SED) were analyzed: s**omatic characteristics**: Body height; Body weight; Triceps skinfold; **Physical fitness**: 600 m run; 60s sit-ups; Stand and reach; Bent arm-hang; 20s arm plate tapping; Standing long jump; Polygon course backwards; 60 m dash. Results were analyzed in 3 groups: (1) patients at least one year before disease onset, (2) Patients in remission on therapy and (3) Patients in remission off therapy. Results are compared to the general population. Results in comparison to the national population are presented in percentiles, according to the age and sex.
**Results**
We analyzed the cohort of 553 JIA patients and matched SED data were available for 459 children with JIA. After diagnosis they participated in fewer SED assessments than before diagnosis (missing measurements in 60% after diagnosis versus 25% before diagnosis), indicating reduced participation in physical education. Even before clinical disease onset, children with JIA already differed significantly from healthy peers: they had lower body weight, lower body mass index, reduced peripheral fat mass, and poorer lower back and leg flexibility. During remission on therapy, persistent deviations remained evident, particularly in body weight, body mass index, and selected motor performance measures. These findings indicate that differences in physical and motor development are present already before diagnosis and persist during treatment.
**Conclusion**
This is worldwide the first study to show distinct pre-disease deviations, that can act as predisposing factors to JIA. Regular monitoring and encouragement of appropriate physical activity, should be part part of the comprehensive management of children with JIA.
**Disclosure of interest**
None declared.

## PT064

### Predicting clinical response to adalimumab in juvenile idiopathic arthritis with a multidimensional model integrating disease activity and pharmacokinetics

G. Tarantino[1,*], A. Di Deo[2], R. Simeoli[3], S. Magni-Manzoni[1], E. Marasco[1], A. Aquilani[1], R. Nicolai[1], M. I. Petrone[1], C. Rossi[3], C. Dionisi Vici[3], B. M. Goffredo[3], O. Della Pasqua[2], F. De Benedetti[1]
[1]Rheumatology Division, Bambino Gesù Children's Hospital IRCCS, Rome, Italy; [2]Clinical Pharmacology & Therapeutics Group, University College of London, London, UK; [3]Division of Metabolic Diseases, Bambino Gesù Children's Hospital IRCCS, Rome, Italy
**Correspondence:** G. Tarantino
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**Introduction**
Adalimumab, among TNF-inhibitors, has revolutionized the treatment of children with juvenile idiopathic arthritis (JIA). Most patients treated with adalimumab respond within months; approximately 30% of the patients may show lack of or loss of response after continued exposure.
**Objectives**
Here, we developed a population pharmacokinetic-dynamic (PK/PD) model based on therapeutic drug monitoring (TDM) of both adalimumab (ADL) and anti-adalimumab antibodies (ADA) levels performed during routine clinical practice.
**Methods**
Records of patients were retrospectively reviewed in a 5-years-period. Adalimumab (ADL) and ADA levels were measured as routine TDM practice by using clinically validated ELISA kits (Immundiagnostik AG). Frequency of adalimumab administration (every two weeks versus every week), dose (20 vs. 40 mg) and disease activity (c-JADAS-10) were recorded. An indirect time-response PK/PD model was applied to investigate the clinical correlation between ADL levels and ADA titers, identifying the predictive covariates of clinical response.
**Results**
Four-hundred and 45 samples from June 2019 to January 2024 were identified from 110 JIA patients. The majority presented ANA-positive oligoarthritis (55.4%) or RF-negative polyarthritis (24.5%). The median age at starting therapy was approximately 10 years [IQR 7.1-15.2]. Pharmacokinetic analysis showed a moderately variable time-profile of both adalimumab (16.3 median micrograms/ml) and ADA titers (median 4.2 AU/ml), regardless of adalimumab dose and frequency of administration. Adalimumab clearance was influenced by 3 variables: adalimumab dose, ADA titer and body weight. We observed a complete clinical response (CID) after 3 months follow-up in 65% of children, who maintain remission also at 6 and 24 months. The predicted adalimumab concentration (Css), at the same drug dose, were on average lower in patients with mild and high disease activity score. Our model could not predict long-term relapses.
**Conclusion**
The effect of ADA on the clinical incidence of loss of response has an important impact in the clinical practise. Our findings allow to identify adalimumab target concentrations based on the exposure-clinical response relationships and highlight the utility of a proactive TDM approach that could help clinicians to recognize potential early non-responder patients, who would need different treatment strategies.
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1. Bartelds G.M. Development of antidrug antibodies against adalimumab and association with disease activity and treatment failure during long-term follow-up. JAMA 2011.

**Disclosure of interest**
None declared.

## PT065

### Serum microRNA hsa-224-5p and hsa-mir-29a-5p diagnostic potential in juvenile idiopathic arthritis

J. Marčiulynaitė[1,*], D. Gečys[2], R. Rakauskas[3], L. Jankauskaitė[1], A. Šnipaitienė[1]
[1]Paediatric Department; [2]Institute of Cardiology, Laboratory of molecular Cardiology; [3]Pediatric Orthopedic Department, Lithuanian University of Health Sciences, Kaunas, Lithuania
**Correspondence:** J. Marčiulynaitė
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**Introduction**
Despite intensive research diagnosis of juvenile idiopathic arthritis (JIA) still relies on non-specific clinical criteria and is difficult to distinguish from other childhood arthritis forms. Recently, microRNAs (miRNAs) are emerging as novel biomarkers in autoimmune diseases.
**Objectives**
to analyse the miRNA expression profiles in synovium fluid and serum of children with JIA and compare with serum of healthy children.

**Methods**
MiRNA expression profiles were determined by next generation sequencing in serum of 7 healthy control (HC) and 4 oligoarticular JIA patients with paired synovial fluid samples from JIA patients. Based on sequencing data overlapping miRNAs (hsa-miR-224-5p, hsa-miR-1246 and hsa-miR-29a-3p) were selected for further validation by real-time quantitative polymerase chain reaction (RT-qPCR). Furthermore, serum expression levels of selected miRNAs were compared between 27 HC, 19 active JIA patients and 13 JIA samples after 9-moths of follow-up and receiver operating curve (ROC) analysis was used for evaluation of miRNAs diagnostic properties.
**Results**
A total of 267 unique miRNAs were observed in serum. Compared with the HC group, the expression of 6 miRNAs was significantly altered in the serum of JIA patients. The expression of hsa-miR-29a-3p and hsa-miR-1246 was upregulated; and expression of hsa-miR-224-5p, hsa-miR-381-3p, hsa-miR-432-5p, and hsa-miR-485-3p was downregulated in serum of JIA patients compared to HC (adjusted p value < 0.05, Log2 fold change ±1). Analysis between miRNA profiles in JIA serum and paired synovial samples revealed that 21 unique miRNA are significantly differentially expressed in synovial fluid when compared to serum. Three overlapping miRNAs between serum and synovium were identified. All tested miRNAs replicated results from NGS experiments. Relative expression of hsa-miR-224-5p was confirmed to be downregulated, whilst levels of hsa-miR-29a-3p and hsa-miR-1246 were upregulated. Hsa-miR-224-5p was expressed at 0.6-fold level ($p$<0.05, 40% decrease) when compared to HC, and levels increased after 9 months of treatment. Contrary, hsa-miR-29a-3p was upregulated in JIA patients by approximately 1.23 -fold ($p$<0.05, 30% increase) compared to HC, and decreased at 9 months ($p$=0.01). Also, hsa-miR-1246 expression was upregulated by 1.29-fold in JIA patients at diagnosis, however this change was not statistically significant. Both hsa-224-5p and hsa-miR-29a-5p showed mediocre sensitivity and specificity in distinguishing HC from JIA patients (AUC 0.642 and 0.641, respectively, $p$<0.05). Aggregated values from hsa-224-5p and hsa-miR-29a-5p had improved AUC of 0.733 ($p$=0.01).
**Conclusion**
Study findings provide compelling evidence that microRNA profiles can serve as clinically useful biomarkers for JIA. Particularly, hsa-224-5p and hsa-miR-29a-5p could be used for diagnosis of JIA.
**Disclosure of interest**
None declared.

## PT066

### Neutrophil extracellular traps and calprotectin in the pathogenesis and prognosis of non-systemic juvenile idiopathic arthritis

B. Balažiová[1,*], J. Janko[2], M. Pastorek[2], T. Mičega[3], D. Mišíková[1], T. Dallos[1]
[1]Department of Paediatrics, Faculty of Medicine, Comenius University in Bratislava, National Institute for Children's Diseases (NÚDCH); [2]Institute of Molecular Biomedicine, Faculty of Medicine, Comenius University in Bratislava, Bratislava; [3]National Institute of Rheumatic Diseases, Piešťany, Slovakia
**Correspondence:** B. Balažiová
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**Introduction**
Non-systemic juvenile idiopathic arthritis (nsJIA) is the most common paediatric rheumatic disease. Although primarily linked to adaptive immune dysregulation, neutrophils usually predominate in synovial fluid (SF) of affected patients and represent a potential source of serum calprotectin (CPT, S100A8/S100A9 protein) – a biomarker of subclinical activity and disease relapse. The role of neutrophil extracellular traps (NETs), of which CPT is a key component, in the pathogenesis and prognosis of nsJIA remains insufficiently understood.
**Objectives**
To evaluate the role of NETs and CPT in the pathogenesis of nsJIA and their associations with disease activity and prognosis.
**Methods**
We included 43 nsJIA patients (age 2.9–16.6 years, median 8.3; F: M=33:10): 23 newly diagnosed and 20 relapsing after treatment withdrawal. Five children with post-traumatic joint effusion served as non-inflammatory controls. NET markers (citrullinated histone H3 [CitH3], neutrophil elastase [NE], myeloperoxidase [MPO], extracellular DNA [ecDNA] and CPT) were measured by ELISA in heparinized peripheral blood (PB) and SF; DNase activity was assessed by SRED method. Disease activity was evaluated by active joint count and JADAS-CRP-10 score. Ankle, tarsal, elbow, temporomandibular joint or cervical spine involvement was considered prognostically unfavourable. Data were statistically analysed using NCSS 11 software.
**Results**
Compared with controls, nsJIA patients had higher SF levels of NE, MPO and CPT (all $p$≤0.001), CitH3 ($p$=0.002) and ecDNA ($p$=0.03). In PB, increased concentrations were confirmed only for NE ($p$=0.04), but not for MPO or CPT. DNase activity, the main mechanism of NET degradation, was higher in PB of nsJIA patients ($p$=0.01), but not in their SF ($p$=0.16). NET markers did not differ between nsJIA subtypes or disease phases. SF NET markers correlated positively with PB CPT (NE, MPO, ecDNA and CPT: all $p$≤0.001; CitH3: $p$=0.009). Higher SF NET markers (MPO $p$=0.003, CitH3 $p$=0.01, NE and CPT both $p$=0.03) and PB NET markers (MPO $p$=0.01, NE $p$=0.04, CPT $p$=0.02) were associated with a higher active joint count. JADAS-CRP-10 correlated positively with SF ecDNA ($p$=0.005) and PB CPT ($p$=0.004), and both JADAS-CRP-10 and plasma CPT correlated negatively with PB DNase activity ($p$=0.006 and $p$=0.001, respectively). Higher CPT in SF ($p$=0.002) and PB ($p$=0.04) were associated with unfavourable joint involvement.
**Conclusion**
Our findings support a role of intra-articular neutrophil activation and NETosis in the pathogenesis of nsJIA. They also suggest that SF NETs are an important source of circulating CPT in nsJIA patients and may contribute to the inhibition of plasma DNase activity. Relatively lower DNase activity in SF may promote NET persistence, thereby supporting local autoimmune inflammation. NET persistence may explain the observed correlation between CPT in PB and nsJIA activity, providing a pathophysiological basis for its use as a biomarker of subclinical disease activity and prognosis in nsJIA.
**Disclosure of interest**
None declared.

## PT067

### Efficacy and safety of subcutaneous ustekinumab in paediatric participants with active juvenile psoriatic arthritis: results of the open-label, phase 3 PSUMMIT-Jr study through week 52

N. Ruperto[1,2,*], D. P. Hawley[3], O. Kasapcopur[4], S. Compeyrot-Lacassagne[5], H. E. Sönmez[6], B. P. Magallares López[7], O. Basaran[8], A. Brigante[9], S. Deepak[10], G. Filocamo[11], S. Ringold[12], S. Howard[13], V. Smith[14], K. Berezny[14], T. N. Kakuda[12], J. H. Leu[14], D. J. Lovell[15], A. Martini[16], H. Brunner[15] on behalf of Pediatric Rheumatology Collaborative Study Group (PRCSG) and Paediatric Rheumatology International Trials Organisation (PRINTO)
[1]Università degli Studi Milano-Bicocca, Milano; [2]IRCCS Fondazione San Gerardo dei Tintori, PRINTO, Monza, Italy; [3]Sheffield Children's Hospital, Sheffield, UK; [4]Istanbul University Cerrahpasa–Cerrahpasa Medical School, Istanbul, Türkiye; [5]Great Ormond Street Hospital for Children & Biomedical Research Centre, London, UK; [6]Kocaeli University Research & Application Hospital, İzmit, Türkiye; [7]Hospital Santa Creu i Sant Pau, Barcelona, Spain; [8]Hacettepe University, Ankara, Türkiye; [9]Fundación/Sanatorio Güemes, Buenos Aires, Argentina; [10]Nottingham University Hospitals NHS Trust, Queen's Medical Centre, Nottingham, UK; [11]Fondazione IRCCS Cà Granda Ospedale Maggiore Policlinico & University of Milan, Milan, Italy; [12]Johnson & Johnson, San Diego, USA; [13]Johnson & Johnson, High Wycombe, UK; [14]Johnson & Johnson, Spring House; [15]Cincinnati Children's Hospital Medical Center, Cincinnati, USA; [16]Università degli Studi di Genova, Genova, Italy
**Correspondence:** N. Ruperto
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**Introduction**
Ustekinumab (UST), a monoclonal antibody to the p40 subunit of IL-12/23, has demonstrated efficacy and safety in adults with psoriasis or psoriatic arthritis (PsA).
**Objectives**
The phase 3 PSUMMIT-Jr study evaluated subcutaneous UST pharmacokinetics (PK), efficacy, immunogenicity, and safety in paediatric participants (pts) with active juvenile PsA (jPsA).
**Methods**
Eligible pts were aged ≥5–<18 yrs and diagnosed with jPsA based on the modified Vancouver classification criteria: arthritis plus psoriasis

(PsO) or ≥2 of the following: dactylitis, nail pits, PsO family history, or PsO-like rash. Active arthritis (≥3 joints with swelling/loss of motion with pain and/or tenderness) was required at baseline. UST (<60 kg, 0.75 mg/kg; ≥60–≤100 kg, 45 mg) was administered subcutaneously at week (W) 0, W4, then every 12 W through W52. The primary efficacy endpoint was Juvenile Idiopathic Arthritis American College of Rheumatology (JIA-ACR) 30 response at W24. Secondary efficacy endpoints were JIA-ACR 30/50/70 through W52; median time to JIA-ACR 30; mean change from baseline in clinical Juvenile Arthritis Disease Activity Score 10 (cJADAS-10) and JADAS-10, -27, -71 through W52; and mean change from baseline in Psoriasis Area and Severity Index (PASI) at W24 among pts with ≥3% body surface area involvement and Physician Global Assessment of PsO score ≥2 (mild to severe) at baseline. PK endpoints were steady-state serum trough concentrations ($C_{trough,\ ss}$) and model-predicted area under the serum concentration-time curve ($AUC_{ss}$) at W28 (primary) and W52 (secondary) by age group. Antibodies to UST were assessed through W52. Safety was assessed through study end.

**Results**

Eighteen pts were enrolled and treated through W52. Median (range) age and weight at enrolment were 13 (7–17) yrs and 52 (22–81) kg, respectively. JIA-ACR 30/50/70 response rates were 89%/83%/61% at W24 and 78%/78%/61% at W52. Median (95% CI) time to JIA-ACR 30 was 4.3 W (4.1, 8.1). At W24/52, mean decreases (improvement) in cJADAS-10 and JADAS-10, -27, and -71 were 13.3/11.6 and 14.9/12.8, 13.0/10.6, and 16.2/13.3, respectively. Mean (SD) PASI score improvement (decrease) at W24/W52 was 3.8 (4.1)/4.0 (4.4). $C_{trough,\ ss}$ and $AUC_{ss}$ were similar between age groups (≥6–<12 yrs and ≥12–<18 yrs) at W28 and W52. There was no apparent relationship between PK and JIA-ACR 30/50/70 responses, although sample size was small for nonresponders. One pt with JIA-ACR 30 response had neutralizing antibodies to UST without impact on efficacy endpoints or safety. Through study end, all pts had ≥1 adverse event (AE) and 4 (22.2%) had ≥1 reasonably related AE (investigator assessed). Infection was the highest reported AE category (88.9%). One pt had a serious AE (tooth abscess), deemed unrelated to treatment.

**Conclusion**

These results demonstrate efficacy and safety of UST in paediatric pts with active jPsA. UST PK in pts with jPsA were similar by age. No new safety signals were identified. These data will be used to support an extrapolation approach to broaden available jPsA treatment options in Europe.

**Trial registration identifying number**:

ClinicalTrials.gov: NCT05083182.

**Disclosure of interest**

N. Ruperto Consultant with: AlfaSigma, Amgen, Boehringer Ingelheim, Bristol Myers Squibb, Eli Lilly, Galapagos, Genentech Roche, Idorsia, Johnson & Johnson, Pfizer, and Takeda, Speaker Bureau with: AlfaSigma, Amgen, Boehringer Ingelheim, Bristol Myers Squibb, Eli Lilly, Galapagos, Genentech Roche, Idorsia, Johnson & Johnson, Pfizer, and Takeda, D. Hawley: None declared, O. Kasapcopur: None declared, S. Compeyrot-Lacassagne Consultant with: Galapagos and Novartis, H. E. Sönmez: None declared, B. P. Magallares López Speaker Bureau with: AstraZeneca and GlaxoSmithKline, O. Basaran: None declared, A. Brigante Speaker Bureau with: Johnson & Johnson, S. Deepak: None declared, G. Filocamo Consultant with: Novartis and Sobi, Speaker Bureau with: Novartis and Sobi, S. Ringold Employee with: Johnson & Johnson, S. Howard Employee with: Johnson & Johnson, V. Smith Employee with: Johnson & Johnson, K. Berezny Employee with: Johnson & Johnson, T. Kakuda Employee with: Johnson & Johnson, J. Leu Employee with: Johnson & Johnson, D. Lovell Grant / Research Support with: Bristol Myers Squibb, Johnson & Johnson, and Roche Laboratories (all paid to institution), Consultant with: AstraZeneca, GlaxoSmithKline, Novartis, Pfizer (consultant and advisory board member), and United Bioscience Corporation (all paid to institution), Speaker Bureau with: Novartis and Pfizer (all paid to institution), A. Martini Consultant with: AbbVie, Boehringer Ingelheim, Eli Lilly, EMD Serono, Idorsia, Johnson & Johnson, Novartis, and Pfizer, H. Brunner Consultant with: AbbVie, Astra Zeneca-Medimmune, Biogen, Boehringer Ingelheim, Bristol-Myers Squibb, Celgene, Eli Lilly, EMD Serono, Genzyme, GlaxoSmithKline, F. Hoffmann-La Roche, Johnson & Johnson, Merck, Novartis, R-Pharm, and Sanofi, Employee with: The Cincinnati Children's Hospital, where Hermine Brunner works as a full-time public employee, has received contributions from the following industries in the past 3 years: Bristol-Myers Squibb, Eli Lilly, GlaxoSmithKline, F. Hoffmann-La Roche, Johnson & Johnson, Novartis, and Pfizer. This funding has been reinvested for the research activities of the hospital in a fully independent manner, without any commitment to third parties., Speaker Bureau with: GlaxoSmithKline, Novartis, and Roche.

## PT068

### Circadian rhythm, sleep, and physical activity in juvenile idiopathic arthritis: are lifestyle factors more important than disease?

P. Perk[1], F. Çakmak[2], Z. Öz Dağdelen[3], R. Koç[2], F. G. Demirkan[4,*]
[1]Pediatric Neurology; [2]Pediatric Rheumatology, Basaksehır Cam and Sakura City Hospital; [3]Pediatric Neurology, Sancaktepe Sehit Prof. Dr. İlhan Varank City Hospital; [4]Pediatric Rheumatology, Kanuni Sultan Süleyman Training and Research Hospital, İstanbul, Türkiye
**Correspondence:** F. G. Demirkan
*Pediatric Rheumatology 2026*, **24(S1):**PT068

**Introduction**

Sleep disturbances are common in juvenile idiopathic arthritis (JIA), yet the interaction between sleep quality, circadian rhythm, and physical activity in affected children remains insufficiently characterized.

**Objectives**

To evaluate sleep patterns, chronotype characteristics, social jet lag, and physical activity levels in children with JIA compared with healthy peers and to identify factors associated with sleep disturbances.

**Methods**

This cross-sectional study included 85 participants aged 8–17 years, comprising 39 children with JIA and 46 age- and sex-matched healthy controls. Sleep, chronotype, social jet lag, and physical activity were assessed using validated pediatric questionnaires. Group comparisons and regression analyses were performed to identify factors associated with sleep disturbance.

**Results**

Children with JIA showed prolonged sleep onset latency on both weekdays and weekends. Median total sleep disturbance scores differed significantly between groups [44 (IQR 35–51) vs. 48 (IQR 41.25–60.50), $p$=0.039]. Chronotype distribution and social jet lag patterns were comparable between groups ($p$=0.947 and $p$=1.000, respectively). Most participants in both groups were classified as morning or intermediate chronotypes, indicating preserved circadian preference in JIA. Physical activity scores and activity categories were also similar between patients and controls ($p$=0.171 and $p$=0.614, respectively). Regression analysis demonstrated that behavioral factors contributed more strongly to sleep disturbance than disease status. In particular, pre-sleep screen exposure was independently associated with worse sleep quality (F=3.343, $p$=0.014) and explained 14.3% of the variance in sleep disturbance scores ($R^2$=0.143), whereas JIA diagnosis was not an independent predictor after adjustment.

**Conclusion**

Children with JIA experience impaired sleep quality, mainly characterized by delayed sleep initiation rather than major circadian disruption or reduced physical activity. The findings suggest that modifiable lifestyle behaviors, particularly pre-sleep electronic device use, contribute importantly to sleep disturbances alongside disease-related factors. Integrating sleep hygiene assessment and targeted behavioral interventions into routine pediatric rheumatology care may improve overall well-being and quality of life in children with JIA.

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**Disclosure of interest**
None declared.

PT069

**Baseline synovial fluid and venous blood cytokine-chemokine profiles as indicators of long-term disease course in oligoarticular juvenile idiopathic arthritis**

E. Doğan İçkin[1], M. O. Erkan[2], H. D. Dara Kar[2,*], D. Ünal[2], H. Ercan Emreol[2], Ö. Necipoğlu Banak[2], S. Özkan[2], Ö. Başaran[2], Y. Bilginer[2], Ş. Özen[2], E. Sağ[2]
[1]Department of Pediatrics; [2]Division of Pediatric Rheumatology, Hacettepe University Faculty of Medicine, Ankara, Türkiye
**Correspondence:** H. D. Dara Kar
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**Introduction**
Juvenile idiopathic arthritis (JIA) is a heterogeneous inflammatory disease characterized by chronic synovitis and variable long-term outcomes. Although cytokines and chemokines play key roles in local and systemic inflammation the prognostic value of early cytokine profiles remains poorly defined.
**Objectives**
This study evaluated baseline synovial fluid plasma (SFP) and venous blood plasma (VBP) cytokine-chemokine profiles obtained at disease onset or flare and investigated their association with long-term disease course.
**Methods**
This prospective study enrolled oligoarticular JIA patients with available clinical, laboratory, and cytokine-chemokine data collected at disease onset or during a flare requiring intraarticular injection.All patients were treatment-naive at the time of sampling.Demographic characteristics, ANA status, uveitis, biologic DMARD use and long-term disease course were recorded.Disease course was categorized as remission, relapsing or chronic. SFP and VBP cytokine-chemokine levels were measured using multiplex cytometric bead array panels.Group comparisons were performed using Mann–Whitney U and Kruskal–Wallis tests; $p<0.05$ was considered statistically significant.
**Results**
Thirty-six patients were included. Median age at disease onset was 5.7 years, median age at injection was 7.1 years, and median follow-up was 80.4 months. ANA positivity was present in 23 patients (74.2%), uveitis in 3 (8.3%), and 5 patients (13.9%) required biologic DMARDs. Disease course was remission in 11 patients (30.6%), relapsing in 20 (55.6%), and chronic in 5 (13.9%). Patients requiring biologic DMARDs had higher SFP IFN-α2 and IL-6 levels (both $p=0.034$) and lower VBP MIP-3α and MIP-1β levels ($p=0.001$ and $p=0.013$). SFP IL-1β and VBP IL-23 differed significantly across disease course groups ($p=0.044$ and $p=0.034$). SFP IL-1β was highest in chronic disease [243.79 (57.95–394.87)] compared with relapsing [0.00 (0.00–2.16)] and remission [0.00 (0.00–122.71)]. Elevated VBP IL-23 was also mainly observed in the chronic group [0.00 (0.00–76.70)]. Several additional SFP and VBP mediators showed a trend toward higher levels in chronic disease and lower levels in remission, although these differences were not statistically significant.
**Conclusion**
Baseline cytokine-chemokine profiling of SFP and VBP may provide meaningful insight into long-term disease course in JIA.SFP IL-1β and VBP IL-23 levels differed significantly among disease course groups with SFP IL-1β being highest in patients with a chronic course.Biologic DMARD use was associated with distinct cytokine patterns, including higher SFP IFN-α2 and IL-6 levels.These findings suggest that early cytokine signatures may reflect subsequent disease trajectory and treatment requirements in JIA.

**Disclosure of interest**
None declared.

PT070

**The updated SOS project: to switch or to swap after the first TNFA failure for childhood non-infectious uveitis**

I. Maccora[1,*], M. Chang[2], S. Magni Manzoni[3], S. Angeles-Han[4], F. Minoia[5], R. Gallizzi[6], A. Taddio[7], L. Fotis[8], M. I. Petrone[3], L. Sutton[2], C. de Libero[9], V. Miraldi-utz[10], M. Mangin[2], G. Tarantino[3], G. Simonini[1]
[1]Rheumatology Unit, ERN ReCONNET Center, Meyer Children's Hospital IRCCS, Florence, Italy; [2]Immunology Unit, Boston Children's Hospital, Boston, USA; [3]Rheumatology Unit, Ospedale Pediatrico Bambino Gesù, Rome, Italy; [4]Rheumatology Unit, Cincinnati Children's Hospital, Cincinnati, USA; [5]Pediatric Immuno-Rheumatology Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico Milano, Milan; [6]Department of Medical of Health Sciences, Magna Graecia University, Catanzaro; [7]Rheumatology Unit, Burlo Garofalo IRCCS, Trieste, Italy; [8]Division of Pediatric Rheumatology, ATTIKON General Hospital, National and Kapodistrian University of Athens, Athene, Greece; [9]ophthalmology unit, Meyer Children's Hospital IRCCS, Florence, Italy; [10]ophthalmology unit, Cincinnati Children's Hospital, Cincinnati, USA
**Correspondence:** I. Maccora
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**Introduction**
25% of patients with Childhood chronic non-infectious uveitis (cNIU) do not achieve disease remission. In this setting, there is no consensus whether switching to another TNFi or swapping drug class is more effective.
**Objectives**
To evaluate the efficacy of the switch OR swap therapeutic approach for treating cNIU refractory to first TNFi course.
**Methods**
Multicentre international retrospective study enrolling children with cNIU unresponsive to the first TNFi, thus requiring another biologic. Response and Remission on treatment was determined according the Standardization of Uveitis Nomenclature criteria.
**Results**
134 children (96 Female) with cNIUhave been collected: 107 JIA-associated uveitis (JIA-U) and 22 idiopathic uveitis (IU). 122 were treated with adalimumab as first-line TNFi and 12 with infliximab. Fifty-five children did not respond to the first treatment, 71 lost response during the disease course. Sixty-nine children were "switched" to a second TNFi (51 infliximab), and 65 "swapped" to a non-TNFi (40 tocilizumab, 9 Jaki, 13 abatacept, 1 canakinumab). Switching to a second TNFi achieved more frequently response ($\chi^2$3.5, p0.04), but not remission ($\chi^2$ 2.65 p 0.07). In JIA-U, Switching gave higher chance to response and remission compared to swap ($\chi^2$5.49 p0.01 and $\chi^2$ 3.98 p 0.038 respectively) but not in IU ($\chi^2$0.19 p0.52 and $\chi^2$0.64 p 0.372 respectively). In anterior cNIU, Switching had higher chance to achieve response but not remission (switch vs. swap $\chi^2$3.4p0.05 and $\chi^2$2.31 p0.097). Non-anterior NIU did not show significant differences ($\chi^2$0.253 p0.50, and $\chi^2$0.35 p0.44). Children with no-anti-ADA have significant better chance to achieve remission by switching compared to children with anti-ADA ($\chi^2$5.49 p0.019) (see Table 1).
**Conclusion**
In our cohort, children who switched achieved more frequently response than those who swap. However, considering remission, the 2 treatment approaches are reasonable, both. To Switch seems to be more appropriate in children with JIA-U and anterior uveitis.
**Disclosure of interest**
None declared.

**Table 1 (Abstract PT070)** Demographic and clinical characteristics of the population in study. Comparison between the population that switched and swapped the treatment. In bold the significant differences

| Variables | Whole cohort 134 | Switch 69 | Swap 65 | Test and *p* value |
|---|---|---|---|---|
| Age at uveitis onset, median IQR | 60 m (31-107) | 72(39-121) | 36 (24-98) | 0.020 |
| Duration of follow-up in months, median IQR | *120 (74-165)* | *110 (61-160)* | *144 (82-!80)* | *0.131* |
| *Response to the I treatment* | *30* | *9* | *21* | ***χ² 6.33 p0.010*** |
| *I TNFi: ADA* | *122* | *58* | *64* | ***χ²8.51*** |
| *IFX* | *12* | *11* | *1* | ***p0.003*** |
| *Time for the administration of TNFi* | *8 (3-26)* | *6.5 (3-20)* | *10 (3-36)* | *0.093* |
| *Duration of the first TNFi* | *18 (9.7-36)* | *21 (10-53)* | *34(16-66)* | ***0.002*** |
| *Response to the I TNFi: absence/ partial* | *75 55* | *33 33* | *42 22* | ***χ²4.2p0.02*** |
| *Loss of response* | *71* | *31* | *40* | *χ²3,8p0.14* |
| *Anti-ADA antibody* | *12/32* | *7* | *5* | |
| *Time for administration* | *54 (25-96)* | *39 (15-79)* | *65 (32-118)* | ***0.002*** |
| *Achievement of response* | *106* | *59* | *47* | ***χ²3.5p0.04*** |
| *Achievement of remission on treatment* | *94* | *54* | *40* | *χ² 2.65 p 0.07* |

## PT071

### Whole genome sequencing in a large cohort of patients with SURF: results from an extended in silico immune gene panel

S. Palmeri[1,2,*], G. Recchi[1], M. Rusmini[3], F. Penco[1], P. Bocca[1], Y. Bilginer[4], S. Fuehner[5], S. Gattulli[3], R. Papa[1], S. Volpi[6], R. Caorsi[1], D. Foell[5], S. Ozen[4], J. I. Arostegui[7], P. Uva[3], I. Ceccherini[8], M. Gattorno[1]
[1]Rheumatology and Autoinflammatory Diseases Unit, IRCCS Istituto Giannina Gaslini; [2]Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genoa; [3]Clinical Bioinformatics Unit, IRCCS Istituto Giannina Gaslini, Genoa, Italy; [4]Pediatric Rheumatology Unit, Department of Pediatrics, Hacettepe University School of Medicine, Ankara, Türkiye; [5]Department of Pediatric Rheumatology and Immunology, University Childrens' Hospital, Muenster, Germany; [6]Clinical and Experimental Immunology, IRCCS Istituto Giannina Gaslini, Genoa, Italy; [7]Hospital Clínic, Fundacio clinic per la recerca biomedica, Barcelona, Spain; [8]UOSD Aggregation Area of the Research Laboratories, IRCCS Istituto Giannina Gaslini, Genoa, Italy
**Correspondence:** S. Palmeri
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**Introduction**
Syndrome of Undifferentiated Recurrent Fever (SURF) defines a group of patients with recurrent febrile episodes clinically distinguishable from Periodic Fever, Aphthous Stomatitis, Pharyngitis, and Adenitis (PFAPA), and in whom no causative genetic defects were identified using conventional genetic testing (1). To date, broader genetic analyses in SURF remain poorly investigated (2).
**Objectives**
This study aims to analyze a large cohort of SURF patients using whole-genome sequencing (WGS) and a bioinformatic gene panel to identify possible rare variants.
**Methods**
One hundred twenty-two patients with SURF enrolled in the perSAIDs project underwent WGS followed by in silico analysis of a panel of 190 genes (associated with monogenic autoinflammatory diseases and immune dysregulation). The following filtering criteria were applied: (i) variants located in coding gene regions; (ii) variants with a minor allele frequency (MAF) of 0.01 or lower in the gnomAD database (v.4.1.1); (iii) no individuals with homozygous genotypes in the previous database; (iv) variants with CADD ≥ 20 or REVEL ≥ 0.5.
**Results**
Among the 122 patients analyzed, 56 did not present gene variants meeting the selection criteria. In the remaining 66 patients, a total of 95 gene variants were identified. Two novel heterozygous variants classified as "likely pathogenic" were found in two patients: a stop-gain variant in the *SOCS1* gene and a missense variant in the*SYK* gene located in the kinase domain of the protein. Seventeen patients presented variants of uncertain significance (VUS) in genes previously associated with dominantly-inherited diseases; 11 patients presented monoallelic variants classified as "pathogenic/likely pathogenic" in genes associated with recessively-inherited diseases, and 44 patients presented heterozygous VUS in recessive genes. The potential presence of copy number variations in combination with variants in recessive genes was excluded.
**Conclusion**
Nearly half of the enrolled patients tested with WGS were negative for rare variants in coding regions of genes associated with autoinflammatory and immune dysregulation diseases. The selected approach identified only two patients with suspected, yet undetected monogenic diseases, while additional variants with a lower predicted pathogenic impact were detected in the remainder of the cohort. This study reinforces the notion that SURF may be a multifactorial disorder.

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3. Palmeri S. and Recchi G. contributed equally.

**Disclosure of interest**
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## PT072

### FMF-associated arthritis in pediatric patients: clinical features and management

S. N. Yoğun[1,*], Y. Uğur Es[1], E. Özçelik[1], S. Erdem Torun[1], D. Öztürk[1], M. C. Polat[2], M. Işıklar Ekici[1], Z. Ekici Tekin [1,3], E. Çelikel[1], C. Karagöl[1], S. Ertem [1], E. Esen[1], E. N. Şener[1], B. Çelikel Acar[1]
[1]Pediatric Rheumatology, Ankara Bilkent City Hospital, Ankara; [2]Pediatric Rheumatology, Gaziantep City Hospital, Gaziantep; [3]Pediatric Rheumatology, Ankara Yıldırım Beyazıt University, Faculty of Medicine, Ankara, Türkiye
**Correspondence:** S. N. Yoğun
*Pediatric Rheumatology 2026*, **24(S1):**PT072

**Introduction**
Arthritis is a common manifestation of familial Mediterranean fever (FMF) and can affect disease management.
**Objectives**
The aim of this study was to evaluate the clinical features of FMF-associated arthritis and compare patients with and without joint involvement. Recurrence, distribution, and number of affected joints were assessed, and patients with persistent or difficult-to-treat arthritis requiring advanced therapies were analyzed.

**Methods**
This study included patients under 18 years who met the Eurofever/PRINTO criteria for FMF, excluding those with arthritis due to other conditions. Demographics, clinical features, *MEFV* variants, disease severity (ISSF), and treatment data were recorded, with joint distribution, recurrence, and therapies analyzed in patients with arthritis.
**Results**
Among 1,251 FMF patients, arthritis was identified in 254 (20.3%), of whom 225 (88.5%) had acute arthritis and 29 (11.4%) had chronic arthritis. The most frequently involved joints were the knee (153 patients, 60.2%) and ankle (133 patients, 52.3%), with recurrent attacks in 169 patients (66.5%; median 3 attacks). Compared with patients without arthritis, those with arthritis had longer attack duration but lower annual attack frequency ($p$<0.001). Fever and abdominal pain were less common, whereas arthralgia, erysipelas-like erythema, and exercise-induced leg pain were more frequent (all $p$<0.001). Moderate to severe disease according to ISSF score and M694V homozygosity (104 patients, 40.9% vs. 203 patients, 20.8%) were more frequent among patients with arthritis. Colchicine resistance occurred more often in patients with arthritis (24 patients, 9.4% vs. 40 patients, 4.0%). In patients with acute arthritis, nonsteroidal anti-inflammatory drugs (NSAIDs) resolved symptoms in 153 patients (68%) and 72 patients (32%) improved spontaneously; recurrence in 154 patients (68%) required colchicine dose escalation. All 29 patients with chronic arthritis initially received NSAIDs, with clinical response in 11 (37.9%); two patients (6.8%) received intra-articular steroids, 16 (55.1%) were treated with conventional disease-modifying antirheumatic drugs (DMARDs) achieving complete response in 11, and 5 patients required biologic DMARDs, all achieving complete response.
**Conclusion**
FMF-associated arthritis was characterized by longer but less frequent attacks, higher M694V homozygosity, and increased colchicine resistance. Acute episodes generally responded to NSAIDs and colchicine adjustment, whereas a subset of patients with chronic arthritis required conventional DMARDs or biologic therapy.

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**Disclosure of interest**
None declared.

## PT073

**Expanding the phenotypic and genetic landscape of zinc-finger NFX1-type containing 1 (ZNFX1): novel clinical spectrum**
N. Almajed[1,*], T. Alshareef[2], R. Alhuthail[1], A. Alazami[3], S. Al-Mayouf[4], A. AlSaleem[1]
[1]Department of Pediatric Rheumatology; [2]Department of Pediatric Nephrology; [3]Department of Translational Genomics, Genomic Medicine Centre of Excellence, King Faisal Specialist Hospital and Research Center; [4]College of Medicine, Al-Faisal University, Riyadh, Saudi Arabia
**Correspondence:** N. Almajed
*Pediatric Rheumatology 2026*, **24(S1):**PT073

**Introduction**
Zinc finger NFX1-type containing 1 (ZNFX1) is an interferon-stimulated double-stranded RNA sensor which plays a crucial role in initiating the immune system response cascade against RNA virus infection. Pathogenic variants in ZNFX1 have been predominantly described in the context of immunodeficiency, lymphoproliferation and increased susceptibility to infections. However, we came across clinical and molecular data indicating that mutation in the ZNFX1 gene may manifest with autoinflammatory features.
**Objectives**
The objective is to describe the phenotype, genotype and immunological markers of Saudi children with proved pathogenic mutation in the ZNFX1 gene.
**Methods**
Genomic analysis was performed using whole exome sequencing and familial segregation to identify ZNFX1gene variants in the identified cohort. Medical records were reviewed for clinical, molecular and immunological profiles including laboratory and functional data.
**Results**
Total of twelve patients (Female=8) from 6 Saudi families were included. Nine patients (75%) presented within the first 2 years. Median age of disease onset was 18 months (IQR: 12-36). Consanguinity was evident in all patients (100%) and more than have (66.6%) had affected family member. Constitutional features were found in majority of patients, namely recurrent fever (83.3%) and failure to thrive (58.3%). The most frequent organ involvements were renal (75%), interstitial lung diseases (66.6%), gastrointestinal (41.6%), and neurological (33.3%). Only two patients had sterile skin abscesses. Recurrent infections were evident in seven patients (58.3%). Total of three patients experienced renal failure required dialysis and renal transplant. Four patients developed macrophage activation syndrome during disease course, only one had more than two attacks. Genetic analysis revealed three pathogenic novel variants of the ZNFX1 gene in six families (c.5574 C> T, c.3293_3294delTTinsAA, c.?); one variant was shared among three families, indicating a relative risk of consanguinity/ethnicity effect. Immunological markers were abnormal in 5 patients including, hypogammaglobinemia and reduced NK-cell lymphocyte markers. Cytokine analysis (MCP1, sIL-2Rα, IL-10, IL-1β, IFN-α, IFN-β, IFN-γ, IL-18) was done in 4 patients yielded nonspecific inflammatory signatures with elevated MCP-1 and sIL-2R. Majority of the patients were treated with corticosteroids ($n$=9), intravenous immunoglobulin ($n$=5), and mycophenolate mofetil ($n$=4). Given the phenotypic overlap with interferonopathies, two patients were treated with Janus kinase inhibitors, both showed marked improvement in clinical and inflammatory markers, proteinuria measures, and radiological findings.
**Conclusion**
Loss of function mutation in the ZNFX1 gene delineate a novel clinical spectrum of early onset inborn error of immunity, characterized by autoinflammation, renal impairment, lung inflammation, and recurrent viral infection. This work represents the first description of this rare entity, expanding phenotype-genotype correlation. Our findings underscore the potential relevance of targeted immunotherapeutic approach using Jak inhibitors, pending functional validation.
**Disclosure of interest**
None declared.

## PT074

**Haploinsufficiency of A20 (HA20): first insights from the Eurofever Registry**
G. Kavrul Kayaalp[1,*], J. Kuemmerle-Deschner[2], I. Koné-Paut[3], N. Assalia[4], A. Insalaco[5], E. Hachulla[6], O. Kasapcopur[7], S. Sahin[7], J. Anton[8], E. Labrador[9], G. Simonini[10], B. Sözeri[11], B. Celikel[12], F. Sztajnbok[13], A. Gagro[14], L. Breda[15], D. Rigante[16], L. Cantarini[17], O. Akgün[1], F. Biscaro[18], E. Van Nieuwenhove[19], E. Berit[20], S. Sener[21], O. Boyarchuk[22], S. Kamphuis[23], S. Ozen[24], M. Gattorno[25], N. Aktay Ayaz[1] on behalf of Eurofever Registry
[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine, Istanbul, Türkiye; [2]Pediatric Rheumatology, University Children's Hospital Tuebingen, Tuebingen, Germany; [3]Pediatric Rheumatology, Le Kremlin-Bicêtre University Hospital, Paris-Sud University – CEREMAI, Le Kremlin Bicetre (Paris), France; [4]Pediatric Rheumatology, Schneider Children's Medical Center of Israel, Petahtekvah, Israel; [5]Pediatric Rheumatology, Ospedale Pediatrico Bambino Gesù, Rome, Italy; [6]Pediatric Rheumatology, CHRU de Lille- Hospital Claude Huriez, Lille, France; [7]Pediatric Rheumatology, Istanbul University-Cerrahpaşa Faculty of Medicine, Istanbul, Türkiye; [8]Pediatric Rheumatology, Hospital Sant Joan de Déu, University of Barcelona, Esplugues de Llobregat, Barcelona; [9]Pediatric Rheumatology, San Pedro Hospital, Logroño, Spain; [10]Pediatric Rheumatology, IRCCS Meyer Children's Hospital, Florence, Italy; [11]Pediatric Rheumatology, Health Sciences University, Umraniye Education and Research Hospital, Istanbul; [12]Pediatric Rheumatology, Ankara Bilkent City Hospital, Ankara, Türkiye; [13]Pediatric Rheumatology, University Hospital Pedro Ernesto, Rio de Janeiro, Brazil; [14]Pediatric Rheumatology, Children's Hospital Zagreb, Zagreb, Croatia; [15]Pediatric Rheumatology, Ospedale Policlinico - Univer-

sita' degli studi di Chieti, Chieti; [16]Pediatric Rheumatology, Cattolica Sacro Cuore University, Rome; [17]Pediatric Rheumatology, Policlinico le Scotte, Universita' di Siena, Siena; [18]Pediatric Rheumatology, Ospedale Ca' Foncello- Treviso, Treviso, Italy; [19]Pediatric Rheumatology, Wilhelmina Kinderziekenhuis, Utrecht, Netherlands; [20]Pediatric Rheumatology, University Hospital of Northern Norway, Tromso, Norway; [21]Pediatric Rheumatology, Adana City Hospital, Adana, Türkiye; [22]Pediatric Rheumatology, Ternopil Regional Children's Hospital, Ternopil, Ukraine; [23]Pediatric Rheumatology, Sophia Children's Hospital, Erasmus University Medical Center, Rotterdam, Netherlands; [24]Pediatric Rheumatology, Hacettepe University Faculty of Medicine, Ankara, Türkiye; [25]Pediatric Rheumatology, IRCCS Istituto G. Gaslini, Genova, Italy
**Correspondence:** G. Kavrul Kayaalp
*Pediatric Rheumatology 2026*, **24(S1):**PT074

**Introduction**
Haploinsufficiency of A20 (HA20), caused by *TNFAIP3* variants, is a recently described monogenic autoinflammatory disease with highly variable phenotype overlapping with Behçet-like and autoimmune features, and limited data on its clinical spectrum. Data from structured international registries may help better delineate it.
**Objectives**
To describe clinical, laboratory, and treatment characteristics of patients with HA20, and to evaluate its phenotypic spectrum.
**Methods**
A multicenter registry-based retrospective observational study was conducted using data from the Eurofever registry. Fifty-eight patients with pediatric-onset TNFAIP3-associated autoinflammatory syndrome were included.
**Results**
The cohort comprised 33 female (56.9%) and 25 male (43.1%) patients. Median age at disease onset was 3.07 years (IQR, 1.01–6.33), and median age at diagnosis was 10.69 years (IQR, 5.39–16.12). The most frequent organ system involvements were mucocutaneous involvement in 52 patients (89.7%), constitutional symptoms in 40 (68.9%), gastrointestinal (GI) involvement in 39 (67.2%), musculoskeletal involvement in 35 (60.3%), and lymphoid involvement in 16 (27.6%). GI involvement mainly presented with abdominal pain (63.8%), GI ulcers (24.1%), and anal/perianal ulcers (19.0%). Endoscopic and histopathological findings showed a broad spectrum, including chronic active gastritis, duodenitis, focal intestinal metaplasia, villous atrophy, nodular ileal mucosa, and ulcerative lesions involving the ileocecal region and colon, with isolated or multifocal distribution across intestinal segments. Mucocutaneous findings included aphthous stomatitis (55.2%), exudative pharyngitis (25.9%), genital ulcers (19.0%), and maculopapular and urticarial rash (17.2% each). Musculoskeletal manifestations included arthralgia (60.3%), monoarthritis (22.4%), oligoarthritis (20.7%), and polyarthritis (10.3%). Ocular findings included conjunctivitis in 7 patients and anterior uveitis in 2; no posterior uveitis or retinal vasculitis was observed. Thrombotic involvement was limited to venous thrombosis in one patient, with no cases of arterial thrombosis. Macrophage activation syndrome occurred in one patient. Autoantibody positivity was detected in 31.0%, and biopsy-proven lupus nephritis was present in one patient. Treatment data were available for 50 patients. Colchicine was the most frequently used drug, in 39 patients (78.0%), and was discontinued for inefficacy in 8. Anti-TNFs were the most common biologics, used in 20 patients (40.0%), with adalimumab being the most frequent (28%) and the most commonly initiated biologic after diagnosis. Corticosteroids were used in 13 patients (26.0%).
**Conclusion**
This study presents the first international European pediatric-onset HA20 cohort and provides a broad registry-based characterization. These findings support recognition of HA20 as a multisystem disease extending beyond the classical Behçet-like phenotype.
**Disclosure of interest**
None declared.

## PT075

### A machine learning model for predicting colchicine resistance in familial Mediterranean fever: a multicenter retrospective study

A. Uzun Bektaş[1,*], B. Pişkin[2], S. Elmas[3], T. Coşkuner[4], A. Tanatar[5], R. Torun[1], E. Arslanoğlu Aydın[3], E. Bağlan[3], G. Otar Yener[6], K. Öztürk[7], F. Demir[8], S. G. Karadağ[9], M. Çakan[10], H. E. Sönmez[11], M. Bülbül[3], N. Aktay Ayaz[12], B. Sözeri[4], B. Bora[1], G. Özdağoğlu[2], S. E. Ünsal[1]
[1]Division of Pediatric Rheumatology, Dokuz Eylul University Faculty of Medicine; [2]Department of Business Administration, Dokuz Eylul University Faculty of Business, Izmir; [3]Division of Pediatric Rheumatology, Ankara Etlik City Hospital, Ankara; [4]Division of Pediatric Rheumatology, University of Health Sciences, Umraniye Training and Research Hospital; [5]Division of Pediatric Rheumatology, Marmara University Pendik Training and Research Hospital, Istanbul; [6]Division of Pediatric Rheumatology, University of Health Sciences, Eskisehir City Hospital, Eskişehir; [7]Division of Pediatric Rheumatology, Istanbul Medeniyet University, Göztepe Prof. Dr. Süleyman Yalçın City Hospital; [8]Division of Pediatric Rheumatology, Acıbadem Ataşehir Hospital; [9]Division of Pediatric Rheumatology, University of Health Sciences, Bakirkoy Dr. Sadi Konuk Training and Research Hospital; [10]Division of Pediatric Rheumatology, University of Health Sciences, Zeynep Kamil Women and Children's Diseases Training and Research Hospital, Istanbul; [11]Division of Pediatric Rheumatology, Kocaeli University Faculty of Medicine, Kocaeli; [12]Division of Pediatric Rheumatology, Istanbul University Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** A. Uzun Bektaş
*Pediatric Rheumatology 2026*, **24(S1):**PT075

**Introduction**
Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease in childhood. Although colchicine remains the cornerstone of treatment, some patients have persistent attacks or inflammation despite appropriate therapy (1). Early recognition of colchicine resistance is important, as delayed identification may prolong disease activity and increase the burden of complications.
**Objectives**
This study aimed to develop and validate an imbalance-aware, clinically interpretable machine learning (ML) model to predict colchicine resistance in pediatric FMF using standard demographic, clinical, and genetic data.
**Methods**
We conducted a multicenter retrospective prediction model study in 3426 pediatric FMF patients, including 163 colchicine-resistant and 3263 colchicine-responsive cases. Candidate predictors included gender, age at diagnosis, age at attack onset, attack frequency, parental consanguinity, comorbidity, fever, abdominal pain, diarrhea, chest pain, arthritis, erysipelas-like erythema, and selected MEFV variants. The dataset was divided into training, validation, and test sets. Logistic regression, random forest, XGBoost, and CatBoost models were evaluated. Because colchicine resistance was a minority class, model development strategies included algorithm-level class weighting and data resampling, along with validation-based probability threshold optimization. The final model was selected by prioritizing sensitivity for colchicine resistance while preserving clinically acceptable specificity. Model performance was assessed using sensitivity and specificity, along with precision, F1-score, balanced accuracy, ROC-AUC, and PR-AUC. Model explainability was explored using SHAP-based feature importance. External validation was performed in an independent cohort of 100 pediatric FMF patients.
**Results**
The final model was an optimized XGBoost classifier using imbalance-aware learning and validation-based threshold tuning. In the independent test set, the model achieved a sensitivity of 96.7%, specificity of 65.7%, precision of 10.4%, F1-score of 18.7%, balanced accuracy of 81.2%, ROC-AUC of 81.2%, and PR-AUC of 10.2%. SHAP analysis indicated that M694V homozygosity, exon 10 involvement, gender, and age at diagnosis were the leading contributors to predicted colchicine

resistance. In external validation, the model achieved 96.0% accuracy, 90.9% sensitivity, 97.4% specificity, and 94.2% ROC-AUC.

**Conclusion**

An imbalance-aware ML pipeline may support early risk stratification for colchicine resistance in pediatric FMF. Along with clinical judgement, the model may support closer monitoring, reassessment of adherence, and earlier treatment optimization. Prospective validation studies are required before routine clinical implementation.

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**Disclosure of interest**

None declared.

## PT076

### MicroRNA expression changes in patients with familial Mediterranean fever and MEFV variants

Š. Horáčková Fingerhutová[1,*], B. Stibůrková[2], J. Mašínová[2], H. Hulejová[2], M. Tesařová[1], M. Pavlíková[3], P. Doležalová[1]
[1]Department of Paediatrics and Inherited Metabolic Disorders, General University Hospital in Prague; [2]Department of Rheumatology, Institute of Rheumatology, 1st Faculty of Medicine, Charles University; [3]Department of Probability and Mathematical Statistics, Faculty of Mathematics and Physics, Prague, Czech Republic
**Correspondence:** Š. Horáčková Fingerhutová
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**Introduction**

Familial Mediterranean fever (FMF) is an autoinflammatory disease associated with variants in the *MEFV* gene. Interpretation of heterozygous and low-penetrance variants remains challenging. MicroRNAs (miRNAs) may serve as biomarkers reflecting inflammatory activity and genotype relevance.

**Objectives**

To analyse expression profiles of selected miRNAs in patients with autoinflammatory phenotypes carrying *MEFV* variants and evaluate associations with genotype, disease activity, and treatment response.

**Methods**

Expression of 13 miRNAs (miR.20a, miR.21, miR.107, miR.144, miR.148b, miR.181, miR.191, miR.195, miR.197, let.7d-3p, let.7d-5p, miR.451a, miR.574) was analysed in relation to genotype, therapy, and febrile versus afebrile states. Patients were stratified according to Infevers classification into: two pathogenic variants, one pathogenic variant, variants of uncertain significance, and negative findings.

**Results**

Twenty-three symptomatic patients with *MEFV* variants and autoinflammatory phenotypes were included. Median age was 9 years (IQR 4.5–20), 56.5% were male. Main manifestations included recurrent fever (78.3%), lymphadenopathy (47.8%), and abdominal pain, arthritis/arthralgia or pharyngitis (39.1%). Patients with *MEFV* variants showed higher miR-144 expression ($p$=0.033), while miR-181b was higher in controls ($p$=0.006). Patients with two pathogenic variants had increased miR-20a and miR-144 compared with controls ($p$=0.003; $p$=0.007). miR-20a also differed between genetically positive and negative patients ($p$=0.011). miR-195 and miR-451a showed a decreasing trend with lower variant pathogenicity ($p$=0.027; $p$=0.022). Following treatment initiation or intensification ($n$=8), miR-20a, miR-107, miR-144, miR-195 and miR-451a significantly increased. During febrile versus afebrile periods ($n$=5), miR-148b was higher in inactive disease ($p$=0.044), while let-7d-5p decreased during fever ($p$=0.004).

**Conclusion**

Distinct miRNA expression profiles are associated with *MEFV* genotype, disease activity, and treatment response in FMF. These results support the potential of miRNAs as biomarkers for interpretation of clinically ambiguous *MEFV* variants and for understanding FMF pathogenesis, although validation in larger cohorts is needed.

**References**

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**Disclosure of interest**

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## PT077

### Evaluation of pubertal development, reproductive hormones, and ovarian reserve in pediatric familial Mediterranean fever: a case-control study

S. N. Tiğrak[1,*], S. Altintas[2], B. Ozhan[2], S. Turkucar[1]
[1]Pediatric Rheumatology; [2]Pediatric Endocrinology, Pamukkale University, Denizli, Türkiye
**Correspondence:** S. N. Tiğrak
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**Introduction**

Familial Mediterranean Fever (FMF) is an inherited autoinflammatory disease characterized by recurrent fever and serosal inflammation [1]. While studies in adult women with FMF have reported conflicting results regarding ovarian reserve—ranging from significantly lower antral follicle counts [2, 3] to mutation-specific declines in Anti-Müllerian Hormone (AMH) [3]—data regarding the pediatric population remains extremely limited. Specifically, no prior research has evaluated reproductive hormones, ovarian reserve, and anthropometry together in children with FMF.

**Objectives**

The primary objective of this study was to evaluate pubertal development, reproductive hormone profiles, and ovarian reserve in children and adolescents with FMF, and to investigate the effects of disease severity and biologic therapy (canakinumab) on these parameters compared to healthy controls.

**Methods**

This prospective case-control study included 67 pediatric patients diagnosed with FMF and 56 healthy age- and sex-matched controls. Pubertal assessment was performed using Tanner staging. Serum levels of AMH, FSH, LH, and estradiol (E2) or total testosterone were measured to determine reproductive hormone profiles. Anthropometric data were systematically recorded. Disease severity within the patient cohort was assessed using the PRAS scoring system. A subgroup analysis was performed within the FMF group to compare patients receiving canakinumab ($n$=12) with those receiving colchicine monotherapy.

**Results**

No significant differences were observed between FMF patients and controls regarding age, sex distribution, height SDS, or hormonal markers ($p$>0.05). However, FMF patients had significantly lower Weight SDS (-0.20 vs. 0.56, $p$=0.003) and BMI SDS (-0.25 vs. 0.65, $p$<0.001) compared to controls. Furthermore, no significant hormonal differences were identified between patients receiving canakinumab and those on colchicine monotherapy ($p$>0.05).

**Conclusion**

This study represents the first prospective case-control research in the literature to comprehensively evaluate reproductive hormones alongside ovarian reserve (AMH) in pediatric FMF patients, including those under biologic therapy. Children and adolescents with FMF demonstrate preserved pubertal development and reproductive hormone profiles comparable to their healthy peers. The preservation of AMH levels suggests that the decline in ovarian reserve frequently documented in adult studies, might be mitigated or potentially prevented through effective, early disease management initiated in childhood. Additionally, canakinumab therapy appears to be reliable regarding reproductive health parameters in the pediatric age group, though further long-term studies are warranted.

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**Disclosure of interest**
None declared.

## PT078

### Paediatric amyloidosis in the era of biological therapy

Z. Ekici Tekin[1,*], M. M. Kaplan[2], A. Yekedüz Bülbül[3], Y. Demir Yiğit[4], M. Kutlar Tanıdır[5], E. D. Batu[6], E. D. Batu[6], E. Kılıç Könte[7], I. Bağrul[8], B. Kasap Demir[9], G. Pirim[10], B. Bozkaya[11], S. Demir[12], S. Ayduran[13], M. Kışla Ekinci[14], K. Öztürk[15], N. Öner[16], Ö. Başaran[6], S. Elmas[8], D. Gezgin Yıldırım[5], S. Yüksel[13], B. Sözeri[10], M. K. Gürgöze[4], A. Paç Kısaarslan[3], Y. Bilginer[6], Ö. Kasapçopur[7], B. Acar[16]
[1]Pediatric Rheumatology, Ankara Yildirim Beyazit University, Faculty of Medicine, Ankara; [2]Pediatric Rheumatology, HATAY TRAINING AND RESEARCH HOSPITAL, Hatay; [3]Pediatric Rheumatology, Kayseri Erciyes University, Faculty of Medicine, Kayseri; [4]Pediatric Rheumatology, Elazığ Fırat University, Faculty of Medicine, Elazığ; [5]Pediatric Rheumatology, Ankara Gazi University, Faculty of Medicine; [6]Pediatric Rheumatology, Ankara Hacettepe University, Faculty of Medicine, Ankara; [7]Pediatric Rheumatology, İstanbul University, Cerrahpaşa Faculty of Medicine, İstanbul; [8]Pediatric Rheumatology, Ankara Etlik City Hospital, Ankara; [9]Pediatric Rheumatology, İzmirKatip Çelebi University, Faculty of Medicine, İzmir; [10]Pediatric Rheumatology, UMRANIYE TRAINING AND RESEARCH HOSPITAL, İstanbul; [11]Pediatric Rheumatology, Samsun On Dokuz Mayıs University, Faculty of Medicine, Samsun; [12]Pediatric Rheumatology, Eskişehir Osmangazi University, Faculty of Medicine, Eskişehir; [13]Pediatric Rheumatology, Pamukkale University, Faculty of Medicine, Denizli; [14]Pediatric Rheumatology, Adana City Hospital, Adana; [15]Pediatric Rheumatology, Göztepe Prof. Dr. Süleyman Yalçın City Hospital, İstanbul; [16]Pediatric Rheumatology, Ankara Bilkent City Hospital, Ankara, Türkiye
**Correspondence:** Z. Ekici Tekin
*Pediatric Rheumatology 2026*, **24(S1):**PT078

**Introduction**
Amyloidosis may be a devastating result of autoinflammatory diseases (AIDs) due to delayed or inappropriate therapy. Early diagnosis and prevention of amyloidosis are important issues in our country, even in childhood, because autoinflammatory diseases such as familial Mediterranean fever (FMF) are common in this region.
**Objectives**
This study examined the AA amyloidosis owing to auto inflammatory disease in a large paediatric population and to describe its prognosis.
**Methods**
The study included 44 patients diagnosed with biopsy-proven AA amyloidosis, who were followed up at 14 paediatric rheumatology centres in Türkiye. Their demographic, clinical, and laboratory parameters, as well as their response to therapy, were analysed retrospectively.
**Results**
The causes of AA amyloidosis were FMF (86.4%), mevalonate kinase deficiency (11.4%) and undifferentiated systemic AID (2.3%). Of the 44 patients with AA amyloidosis, 26 (59.1%) were male. The median age at symptom onset, diagnosis, and amyloidosis were 6 years (range 3–9), 8 years (range 6–10.75), and 10 years (range 8.25–15), respectively. AA amyloidosis was observed in 19 patients (43.2%) at initial diagnosis and in 25 patients (58.8%) during follow-up. All patients except 12 (27.3%) took colchicine prior to amyloidosis diagnosis. Additionally, corticosteroids (34.1%) and conventional DMARDs (18.2%) were used to treat massive proteinuria. Although the flare counts of primary AIDs decreased significantly after therapy, subclinical inflammation continued in 36 patients (81.8%) despite treatment. Biological therapy was administered to all patients with amyloidosis, except three (6.8%). The median interval between diagnosis of amyloidosis and initiation of biological therapy was 2 (range 0–12) months. The main biological therapy options of amyloidosis were anakinra (68.2%), canakinumab (81.8%) and tocilizumab (9.1%). In addition, twenty-seven patients needed exchange of biological therapy. Of the 14 patients with renal failure (31.8%), 11 (25%) required dialysis and seven (15.9%) had undergone renal transplantation. Of the seven kidney transplant patients, four had never used colchicine prior to diagnosis of amyloidosis. Furthermore, three of these four patients were unable to take biological therapy. Our study showed that patients with amyloidosis who underwent biological therapy within six months were significantly less likely to require a renal transplant or dialysis ($p<0.05$).
**Conclusion**
Colchicine is essential for preventing amyloidosis. Prompt biological treatment appears to reduce the need for dialysis and transplantation in patients with amyloidosis.

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**Disclosure of interest**
None declared.

## PT079

### Clinical characteristics, treatment and outcomes of 10 Chinese patients with adenosine deaminase 2 deficiency

L. Guo[1,*], Z. Wang[1], M. Lu[1]
[1]Rheumatology Immunology & Allergy, The Children's Hospital, Zhejiang University School of Medicine, Hangzhou, China
**Correspondence:** L. Guo
*Pediatric Rheumatology 2026*, **24(S1):**PT079

**Introduction**
Deficiency of adenosine deaminase 2 (DADA2) is an autosomal recessive autoinflammatory disorder caused by biallelic loss-of-function variants in adenosine deaminase 2 (ADA2).
**Objectives**
We aim to study the characteristics of DADA2 in Chinese population.
**Methods**
A retrospective analysis of 10 patients with DADA2 identified through whole exome sequencing (WES) was conducted. Clinical phenotype, genotype, and treatment response were analyzed.
**Results**
10 patients with DADA2 from 7 families were enrolled between July 2019 and May 2026, of which 7 were male and 3 were female. Adenosine deaminase 2 enzymatic activity was low in all tested cases to confirm pathogenicity. Median age of disease presentation was 2.7 years and the median age at diagnosis was 7.5 years. The median follow-up time was 3 years (0.3 - 7 years).The clinical phenotypes included 7 cases of vasculitis, 2 cases of asymptomatic type, and 1 case of pure red cell aplasia. The identified genotypes had p.R169G, p.G47W, p.G48R, p.F355L, p.G383S, p.L469P, Exon2 del and Exon6 del, of which the p.R169G was the most common genotype. 1 patient of vasculitis showed dominant inheritance with a heterozygous mutation. 2 asymptomatic patients did not receive any treatment and were observed during the follow-up period. A pair of brothers with vasculitis phenotype refused TNFi treatment. Both had oculomotor nerve involvement and presented with strabismus. The younger brother had already suffered a ischemic stroke. The other 5 patients with vasculitis phenotypes responded effectively to TNFi treatment. Among them, 3 patients had tried to use tofacitinib, and 1 patient had tried to use tocilizumab, but all were ineffective. Patients with pure red aplastic anemia who received tofacitinib treatment showed effective results.
**Conclusion**
To establish early diagnosis and improve clinical outcomes, genetic screening and/or testing of ADA2 enzymatic activity should be performed in patients with suspected clinical features. TNFi is an effective treatment for vasculitis phenotypes, while JAKi is ineffective. However, JAKi may be effective for pure red anemia.

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**Disclosure of interest**
None declared.

## PT080

### Clinically driven phenotype–immunotype–genotype alignment refines outcomes in refractory childhood immune dysregulation

D. B. Pandya[1,*]
[1]Pediatric Rheumatology & Immunology, Dev Pediatric Rheumatology & Immunology Center, Ahmedabad, India
**Correspondence:** D. B. Pandya
*Pediatric Rheumatology 2026*, **24(S1):**PT080

**Introduction**
Monogenic immune dysregulation increasingly explains **refractory** autoimmunity, autoinflammation and hyperinflammatory disorders in children. Translating genomic insights into effective management remains difficult in **resource-limited settings** because of **delayed diagnosis** and **limited access** to advanced immunological evaluation and **precision therapies**.
**Objectives**
To evaluate the impact of **clinically driven** phenotype–immunotype–genotype alignment on treatment outcomes in childhood monogenic immune dysregulation disorders and analyse the effect of diagnostic delay on the outcomes.
**Methods**
This retrospective single-centre study included **30 children** with onset before 18 years presenting with immune dysregulation phenotypes. Clinical phenotyping, immunological evaluation and exome sequencing were performed. Variants were considered actionable if pathogenic/likely pathogenic or phenotype-concordant VUS supported by in silico prediction and pathway relevance. Immunotype assessment included immunoglobulin profile and lymphocyte subsets. Management was refined through **clinically aligned pathway-based therapy**. **Primary outcome measures** were complete remission (**CR**), partial response (**PR**) and refractory disease. CR was defined as absence of active disease for ≥1 month off systemic glucocorticoids, while PR was defined as significant clinical/laboratory improvement with glucocorticoid tapering. Secondary outcome assessed association between **diagnostic delay** and treatment response. Pre/post comparisons used McNemar test; diagnostic delay was analysed using Mann–Whitney U test.
**Results**
Thirty children were included (male: female 20:10). Median age at onset was 2.0 years (IQR 1.0–4.0) and diagnosis 6.5 years (IQR 3.0–11.0), with mean **diagnostic delay 3.68±2.68 years**. Phenotypes were **autoimmune (60%)**, autoinflammatory/hyperinflammatory (26.7%) and mixed immune dysregulation (13.3%). Marked genetic **heterogeneity** was observed across 25 genes including SOCS1, PLCG1, TCF3, BTK, FCGR2B, NOD2, MVK, RNF31, CYBB, IKBKB, LRBA, RAG1, PSMB8, CTLA4, RELA, C1Q, COPA, STXBP2, DOCK8, WAS, RUNX1, ADAM17, PIK3CD, NFKB2 and JAK3. Before guided management, 6.7% patients were in remission while 63.3% had refractory disease. Following pathway-aligned therapy in **23/30** patients, **CR and PR were achieved in 69.6% and 26.1%**, respectively (**overall benefit 95.7%**). **Refractory disease reduced from 63.3% to 0%** ($p$<0.001). **Longer diagnostic delay reduced likelihood of CR** ($p$=0.020). Therapies included JAK inhibitors, sirolimus, anti-TNF agents, colchicine and cytokine-directed biologics. Three deaths and two losses to follow-up occurred ; **adverse events were infrequent (8.7%)**.
**Conclusion**
Clinically driven phenotype–immunotype–genotype alignment substantially improves outcomes in refractory childhood immune dysregulation. In resource-limited settings, delayed diagnosis and limited access—not lack of therapeutic efficacy—remain major barriers to remission.
**Disclosure of interest**
None declared.

## PT081

### High-frequency ultrasound as a non-invasive biomarker of inflammatory activity in juvenile localized scleroderma

M. T. Terreri[1,*], G. Clemente[1], A. P. Sakamoto[1], L. M. Campos[2], M. Fraga[1], D. Gerent Petry Piotto[1]
[1]Pediatrics, Universidade Federal de São Paulo; [2]Pediatrics, Universidade de São Paulo, Sao Paulo, Brazil
**Correspondence:** M. T. Terreri
*Pediatric Rheumatology 2026*, **24(S1):**PT081

**Introduction**
Clinical assessment may underestimate inflammatory activity in juvenile localized scleroderma (JLS), particularly across heterogeneous lesions.
**Objectives**
To evaluate high-frequency ultrasound (HFUS) as a non-invasive biomarker of inflammatory activity in JLS.
**Methods**
Thirty-six patients with JLS were included. Lesion-level analysis integrated data from two time points, totaling 229 lesion assessments. Clinical measures included LoSCAT, modified Rodnan skin score, and Physician/Patient Global Assessment (PGA). HFUS (22 MHz) assessed dermal thickness, echogenicity, and Power Doppler (PD). Associations were evaluated using mixed-effects models.
**Results**
The cohort was predominantly female (63.9%), with mean age 13.2 ± 4.5 years. HFUS-defined active lesions showed higher clinical activity than inactive lesions, with higher LoSCAT activity (2.32 ± 1.90 vs. 0.87 ± 1.33, $p < 0.001$) and modified Rodnan scores ($p = 0.007$). Increased dermal thickness, reduced echogenicity, and PD positivity were consistently associated with higher activity (all $p < 0.001$). Hypodermal abnormalities, particularly decreased echogenicity and PD signal, also correlated with increased clinical activity ($p < 0.001$). Importantly, ultrasonographic findings were not consistently associated with damage indices, supporting their specificity for inflammatory activity.
**Conclusion**
HFUS demonstrates strong and consistent associations with clinical disease activity in JLS at the lesion level, supporting its role as a sensitive, non-invasive biomarker of inflammation. Its ability to detect activity beyond clinical assessment highlights its potential to improve disease monitoring and guide therapeutic decisions.
**Disclosure of interest**
None declared.

## PT082

### Organ involvement patterns by anti Scl 70 and anti centromere status in juvenile systemic sclerosis. Results of the Juvenile Scleroderma Inception Cohort

I. Foeldvari[1,*], J. Klotsche[2], K. Torok[3], M. T. Terreri[3], A. P. Sakamoto[3], O. Kasapcopur[3], A. Adrovic[3], J. Anton[3], S. Johnson[3], F. Sztajnbok[3], B. Feldman[3], R. Khubchandani[3], S. Hajek[3], E. Alexeeva[3], D. Schonenberg-Meinema[3], M. Katsicas [3], V. Smith[3], S. Abu Al-Saoud[3], E. Al-Abadi[3], S. Appenzeller[3], S. O. Hetlevik[3], M. Kostik[3], S. Li[3], H. Malcova[3], E. Marrani[3], A. Patwardhan[3], W.-A. Sifuentes-Giraldo[3], N. Vasquez-Canizares[3], T. Avcin [3], N. Cabrera[3], D. Nemcova [3], M. J. Santos[3], C. Battagliotti[3], L. Berntson[3], B. Bica[3], J. Brunner[3], P. Costa Reis[3], L. Harel[3], A. Horne [3], D. Kaiser[3], T. Kallinich[3], D. Lazarevic[3], T. Lehman[3], J. M. López Benítez[3], K. Minden[3], S. Nielsen[3], F. Nuruzzaman[3], C. Pain[3], S. Sawhney[3], P. Seferna[3], M. Sparchez[3], N. Helmus[1]
[1]Hamburg Centre for Pediatric and Adolescence Rheumatology, Hamburg; [2]German Rheumatism Research Center, Berlin; [3]jSSc collaborative group, Hamburg, Germany
**Correspondence:** I. Foeldvari
*Pediatric Rheumatology 2026*, **24(S1):**PT082

**Introduction**
Juvenile systemic sclerosis (jSSc) is a rare disease with an estimated prevalence of 3 per 1,000,000 children. The Juvenile Systemic Scleroderma Inception Cohort (jSScC) is the largest prospective cohort worldwide, enabling longitudinal assessment. While anti-topoisomerase (Scl70) and anti-centromere (AC) antibodies have each been individually associated with clinical phenotypes in jSSc, organ involvement patterns according to their combined presence or absence have not been systematically evaluated. The aim of this analysis was to compare baseline organ involvement patterns by combined antibody status.
**Objectives**
The aim of this analysis was to compare baseline organ involvement patterns by combined antibody status.
**Methods**
Baseline data from patients enrolled in the jSScC up to 15 March 2026 were analyzed. Demographic characteristics, clinical features, laboratory findings, and patient- and physician-reported outcomes at cohort entry were extracted. Patients were stratified into 4 groups: double antibody negative (Scl-70–/AC–; dbNEG), Scl70 positive and AC negative (Scl70+), AC positive and SCl70 negative (AC+), and double-positive patients(dbPos). Group comparisons were performed using analysis of variance and chi-square test.
**Results**
Data from 205 patients were analyzed. Most patients were dbNEG (141/205; 68.8%). Scl70+ was observed in 54/205 patients (26.3%), while AC+ was present in 10/205 patients (4.9%). 3 patients were dbPos. The distribution of cutaneous subtype differed significantly, with diffuse subtype observed in 83% of Scl70 patients, 62% of dbNEG patients, and 40% of AC patients ($p$=0.008). Median age at onset of Raynaud's phenomenon and first non-Raynaud's manifestation tended to be later in the AC group. A history of digital ulceration was most frequent in the Scl70+ group (70%) compared with dbNEG (49%) and AC+ (40%) ($p$=0.080). Pulmonary involvement was more prominent in AC+ patients, with FVC <80% predicted in 86% versus 32% in Scl70 and 27% in dbNEG patients ($p$=0.011), corresponding to a higher prevalence of interstitial lung disease on HRCT (87.5% vs. 48% and 41%, respectively; $p$=0.056). Gastrointestinal involvement was numerically higher in AC patients, including more frequent oesophageal involvement and BMI <–2 z-scores. Musculoskeletal manifestations, particularly contractures, were also more frequent in AC patients (80% vs. 61% in Scl70 and 50% in dbNEG). Physician-assessed global damage scores were higher in AC patients (median 50 vs. 30; $p$=0.091).
**Conclusion**
Stratification by combined anti-Scl-70 and anti-centromere antibody status revealed differences in baseline organ involvement patterns in jSSc. Anti-centromere–positive patients showed trends toward greater pulmonary, musculoskeletal, and overall damage burden, while double-negative and anti-Scl-70–positive patients displayed more comparable profiles. These findings extend existing antibody-associated phenotype data and support the value of combined autoantibody stratification.
**Disclosure of interest**
None declared.

## PT083

### Efficacy and safety of divozilimab in adolescents with juvenile systemic sclerosis: results after 48 weeks of therapy in LIBERIUS study

M. Kostik[1,*], S. Valiyeva[2,3], S. Kurbanova[2], M. Kapranova [2], A. Zinkina[4], Y. Linkova[4], A. Gaponova[4], A. Eremeeva [4]
[1]Saint-Petersburg State Pediatric Medical University, Saint Petersburg; [2]Morozov Children's City Clinical Hospital; [3]N.I. Pirogov Russian National Research Medical University, Moscow; [4]JSC BIOCAD, Saint Petersburg, Russian Federation
**Correspondence:** M. Kostik
*Pediatric Rheumatology 2026*, **24(S1):**PT083

**Introduction**
Juvenile Systemic Sclerosis (JSS) in adolescents is a rare autoimmune fibrotic disease with a progressive course and limited therapeutic options.
**Objectives**
To evaluate the efficacy and safety profile of divozilimab in adolescent patients with JSS.
**Methods**
This subset analysis evaluated data from adolescents (≥14 to <18 years) enrolled in a Phase III, randomized, double-blind, placebo-controlled trial (NCT05726630). Eligible subjects received Divozilimab (250 mg intravenously at Weeks 0 and 2, followed by 500 mg every 24 weeks). Placebo was not used in adolescents for ethical reasons due to their vulnerable status. Change in modified Rodnan Skin Score (mRSS), as well as patient-reported outcomes including Childhood Health Assessment Questionnaire (CHAQ), Medical Outcomes Study 36-Item Short Form Health Survey Physical Component Summary (SF-36-PCS), SF-36 Mental Component Summary (SF-36-MCS), and EuroQol 5-dimension 3-level (EQ-5D-3L) were assessed, along with safety, tolerability, and immunogenicity.
**Results**
Nine adolescents (3 boys and 6 girls) were included with a mean baseline mRSS of 13.8±3.3, indicating moderate skin fibrosis. The mRSS decreased to 12.4±3.5 at Week 24 and further to 9.9±5.8 at Week 48, reflecting improvement to mild skin involvement. Forced vital capacity (FVC) parameters remained stable without evidence of pulmonary worsening. Patient-reported outcomes demonstrated improvement at Week 48: CHAQ dropped from 10.0 to 7.7, SF-36-PCS improved from 46.0 to 54.0, and EQ-5D-3L scores increased from 68.6 to 75.8. The SF-36 Mental Component Summary showed no significant changes during the study period. Immunogenicity assessment did not reveal binding or neutralizing antibodies. All subjects experienced at least one adverse event (AE), with 88.9% (8/9) being treatment-related (TRAE). No SAEs were reported. One subject experienced TRAE of grade 3 or higher according to CTCAE v.5.0. (alanine aminotransferase and aspartate aminotransferase were increased). Most frequently reported TRAEs were pharyngitis (2/9; 22.2%) and transient infusion-related reactions (22.2%). Other TRAEs included injection site reactions, increased creatinine, viral respiratory tract infection, asthenia, nausea, myalgia, and dry skin (11.1% each). All adverse reactions resolved without sequelae and did not require the withdrawal of the studied drug.
**Conclusion**
Divozilimab reduced skin fibrosis in adolescents with JSS over 48 weeks and improved patient-reported outcomes, such as daily functional ability, physical function, and quality of life. The treatment exhibited a favorable safety profile with no immunogenicity concerns, supporting its continued development in the adolescent population.
**Trial registration identifying number**
Trial registration: NCT05726630.
**Disclosure of interest**
None declared.

## PT084

### Biologic therapy in refractory juvenile localized scleroderma: outcomes and predictive factors from a multicenter cohort

E. Kilic Konte[1,*], S. Atamyildiz Ucar[2], M. Gurbuz[1], H. D. Dara Kar[3], A. Alisan[4], M. C. Polat[5], K. Deveci Seker[6], E. Tunce[2], T. Kasap[7], S. Sener[8], D. Aydın[9], N. Sahin[9], O. A. Akın[10], G. Sumer[11], H. Adıguzel Dundar[12], H. Tastanoglu[13], K. Ulu[14], V. Cam[15], G. Ozdemir[16], S. Demir[10], R. M. Kisla Ekinci[4], O. Basaran[3], A. Adrovic[1], A. Pac Kisaarslan[13], S. S. Kilic[11], S. Yuksel[7], S. A. Bakkaloglu[6], N. Aktay Ayaz[16], B. Sozeri[2], O. Kasapcopur[1]
[1]Department of Pediatric Rheumatology, İstanbul University-Cerrahpaşa, Cerrahpaşa Faculty of Medicine; [2]Department of Pediatric Rheumatology, University of Health Sciences, Ümraniye Training and Research Hospital, Istanbul; [3]Department of Pediatric Rheumatology, Hacettepe University Faculty of Medicine, Ankara; [4]Department of Pediatric Rheumatology, Çukurova University Faculty of Medicine, Adana; [5]Department of Pediatric Rheumatology, Gaziantep City Hospital, Gaziantep; [6]Department of Pediatric Rheumatology, Gazi University Faculty of Medicine, Ankara; [7]Department of Pediatric Rheumatology, Onsekiz Mart University Faculty of Medicine, Çanakkale; [8]Department of Pediatric Rheumatology, University of Health Sciences Adana Training and Research Hospital, Adana; [9]Department of Pediatric Rheumatology, Kocaeli University Faculty of Medicine, Kocaeli; [10]Department of Pediatric Rheumatology, Osmangazi University Faculty of Medicine, Eskisehir; [11]Department of Pediatric Rheumatology, Uludağ University Faculty of Medicine, Bursa; [12]Department of Pediatric Rheumatology, University of Health Sciences, Dr. Behçet Uz Children's Hospital, Izmir; [13]Department of Pediatric Rheumatology, Erciyes University Faculty of Medicine, Kayseri; [14]Department of Pediatric Rheumatolo-

gy, Sancaktepe Şehit Prof. Dr. İlhan Varank Training and Research Hospital, Istanbul; [15]Department of Pediatric Rheumatology, University of Health Sciences Sincan Training and Research Hospital, Ankara; [16]Department of Pediatric Rheumatology, Istanbul University Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** E. Kilic Konte
*Pediatric Rheumatology 2026*, **24(S1):**PT084

**Introduction**
Juvenile localized scleroderma (JLS) is an idiopathic autoimmune disorder characterized by chronic inflammation and fibrosis of the skin and subcutaneous tissue. Biologic agents are increasingly required for patients refractory to first-line treatments. The optimal biologic choice and predictors of treatment response remain poorly defined.
**Objectives**
This study aimed to evaluate biologic treatment responses in JLS patients unresponsive or intolerant to first-line therapies and to identify predictive factors associated with the need for biologic treatment.
**Methods**
Patients diagnosed with JLS over the past decade and followed at pediatric rheumatology centers were retrospectively enrolled. Demographic, clinical, and laboratory characteristics were recorded. Disease activity and damage were assessed using the modified Localized Scleroderma Skin Severity Index (mLoSSI), Localized Scleroderma Damage Index (LoSDI), Localized Scleroderma Cutaneous Assessment Tool (LoSCAT), and Localized Scleroderma Cutaneous Activity Measure (LSCAM) both before and after biologic initiation.
**Results**
Forty-three patients (74.4% female) were included. Mean ages at symptom onset, diagnosis, and last visit were 7.3 (±3.4), 8.9 (±3.7), and 13.7 (±4.2) years, respectively. Mean diagnostic delay was 13.7 (±4.2) months, and mean follow-up duration was 56.3 (±42.6) months. Generalized morphea and linear scleroderma of the head were the most prevalent subtypes (each $n$=17, 39.5%), followed by linear scleroderma of the trunk and extremities ($n$=5, 11.6%). ANA positivity was detected in 22 of 43 patients (51.2%), and Scl-70 positivity was absent in all tested patients. Tocilizumab was the predominant biologic agent ($n$=41, response rate 78.0%), followed by rituximab ($n$=5, 40.0%) and anti-TNF agents ($n$=3, 100%). Overall, biologic response was achieved in 33 of 43 patients (76.7%), and 10 patients (23.3%) had active disease at the last visit. Biologic treatment was associated with significant improvements across all disease activity and damage indices ($p$<0.05): mLoSSI from 12 (5–22.7) to 6 (2–13.5), LoSDI from 8 (5–24) to 7 (3–19), LoSCAT from 27 (10–48) to 15 (5–40), and LSCAM from 14 (5.7–39) to 6 (3–22).
**Conclusion**
Tocilizumab appears to be a promising therapeutic option for JLS patients unresponsive or intolerant to first-line therapies, with meaningful reductions across all disease activity and damage scores. However, prospective controlled studies are needed to establish optimal patient selection, treatment timing, and long-term outcomes.

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**Disclosure of interest**
None declared.

## PT085

### CD19-targeting CAR T cell therapy in a child with refractory, life-threatening anti-fibrillarin positive systemic sclerosis with cardiac involvement and myositis

P. Bøyesen[1,*], J. Buechner[2], A. Christophersen[1], A.-B. Aga[1], L. Osnes[3], H. Brun[4], N. K. Sande[1], H. Sanner[1,5], Ø. Molberg[1,5], V. Lilleby[1]
[1]Rheumatology; [2]Pediatric Hematology/Oncology; [3]Immunology; [4]Pediatric Cardiology, Oslo University Hospital; [5]Clinical Medicine, Faculty of Medicine, University of Oslo, Oslo, Norway
**Correspondence:** P. Bøyesen
*Pediatric Rheumatology 2026*, **24(S1):**PT085

**Introduction**
Anti fibrillarin–positive juvenile systemic sclerosis (jSSc) is associated with high disease severity. CD19 targeting chimeric antigen receptor T-cell therapy (CD19 CAR-T), have recently shown promise in a jSSc case.
**Objectives**
To describe the safety, clinical, and immunological effects of CD19 CAR-T in an adolescent with life–threatening, anti–fibrillarin–positive, refractory jSSc.
**Methods**
We report a 15–year–old girl with anti–fibrillarin–positive diffuse cutaneous jSSc with 18 months disease duration including rapid progressive skin fibrosis, severe myositis, pulmonary arterial hypertension, and myocardial fibrosis, refractory to conventional and biologic immunosuppression, including rituximab. The patient received a single infusion of tisagenlecleucel, a commercially available autologous second-generation CD19 CAR-T product with a 4-1BB costimulatory domain following standard fludarabine–cyclophosphamide lymphodepletion. Clinical outcomes, patient–reported outcomes (PROs), and safety, were evaluated longitudinally up to 9 months post–infusion.
**Results**
CAR-T cells expanded rapidly in vivo, peaking on day 7, and became undetectable from week 8 onward. Peripheral $CD19^+$ B cells remained depleted until gradual reconstitution beginning at month 6, dominated by transitional and naïve phenotypes. No cytokine release syndrome, neurotoxicity, serious infections, or late toxicities occurred. Marked clinical improvement was observed across multiple domains: modified Rodnan skin score decreased from 29 at baseline to 11 at 9 months; serum creatine kinase normalised by 6 months (218 U/L to 70 U/L); muscle strength and exercise capacity improved, with 6 min walk distance increasing from 360 m pre–infusion to 515 m at 9 months; and overall juvenile systemic sclerosis disease severity scores (J4S) declined from 13.5 to 8. Her mild pulmonary arterial hypertension and myocardial fibrosis remained stable. Patient/parent global disease activity visual analogue scale (VAS, 0–100 mm) improved from 63 mm at baseline to 38 mm at 3 months, 36 mm at 6 months, and 16 mm at 9 months (−75%). Hand–related disability, assessed by the Cochin Hand Functional Scale (0–90), improved from 23 at baseline to 14 at 9 months (−39%) (see Table 1).
**Conclusion**
CD19 CAR-T therapy induced sustained, drug–free, multi–organ clinical improvement at 9 months with a favourable safety profile in an adolescent with severe, refractory jSSc.
**Disclosure of interest**
P. Bøyesen: None declared, J. Buechner Consultant with: Participation in advisory boards: Novartis, AMGEN, Pfizer, medac, Janssen. Study steering committee/speaker´s bureau/honoraria: Novartis., A. Christophersen: None declared, A.-B. Aga: None declared, L. Osnes: None declared, H. Brun: None declared, N. Sande: None declared, H. Sanner: None declared, Ø. Molberg: None declared, V. Lilleby: None declared.

**Table 1 (Abstract PT085)** Clinical, biochemical and immunological measures of disease activity

| | CAR-T inf. | 3 months | 6 months | 9 months |
|---|---|---|---|---|
| Modified Rodnan Skin Score (mRSS, range 0-51) | 29 | 25 | 21 | 11 |
| Six-minute walk test (m) | 360 | 465 | 465 | 515 |
| Childhood myositis assessment scale (CMAS, range 0-52) | 23 | 21 | 26 | 29 |
| Juvenile Systemic Sclerosis Disease Severity Score - J4S (0-40) | 13,5 | 9,5 | 11 | 8 |
| Anti-Fibrillarin IgG (<10×10^3 U/L) | >300,0 | >300,0 | >300,0 | 285,0 |

## PT086

### Clinical characteristics associated with progressive interstitial lung disease in juvenile systemic sclerosis: 12-month data from the Juvenile Scleroderma Inception Cohort

I. Foeldvari[1,*], K. Torok[2], J. Klotsche[3], B. Feldman[2], O. Kasapcopur[2], A. Adrovic[2], J. Anton[2], M. T. Terreri[2], A. P. Sakamoto[2], M. Katsicas [2], F. Sztajnbok[2], E. Alexeeva[2], T. Avcin [2], S. Li[2], S. Appenzeller[2], S. Hajek[2], S. Johnson[2], D. Schonenberg-Meinema[2], S. Abu Al-Saoud[2], L. Berntson[2], B. Bica[2], P. Costa Reis[2], S. O. Hetlevik[2], M. Janarthanan [2], T. Kallinich[2], R. Khubchandani[2], E. Marrani[2], K. Minden[2], D. Nemcova [2], A. Patwardhan[2], V. Smith[2], M. Sparchez[2], V. Stanevicha[2], N. Helmus[1]
[1]Hamburg Centre for Pediatric and Adolescence Rheumatology; [2]jSSc collaborative group, Hamburg; [3]German Rheumatism Research Center, Berlin, Germany
**Correspondence:** I. Foeldvari
*Pediatric Rheumatology 2026*, **24(S1):**PT086

**Introduction**
At the international consensus meeting in Hamburg 2025, a multidisciplinary expert group proposed a working definition for progressive interstitial lung disease (progILD) in juvenile systemic sclerosis (jSSc). ProgILD was defined by at least one of the following (Table 1). Application of this definition to well-characterized cohorts is an important step toward to understand ProgILD.
**Objectives**
To apply the proposed definition of progILD to the Juvenile Scleroderma Inception Cohort (jSScC) and to describe clinical characteristics associated with ILD progression over 12 months.
**Methods**
Data were extracted from the jSScC up December 2025. Patients with ILD were categorized according to whether they met criteria for ILD progression(ILDprog) over 12 months. Demographic characteristics, clinical features, laboratory parameters, and organ involvement were compared between patients with ILDprog and without (ILDnoprog) at inclusion and after 12 months of follow-up. Categorical variables were compared using $\chi^2$ or Fisher's exact tests, and continuous variables using non-parametric tests. A two-sided p value <0.05 was considered statistically significant.
**Results**
A total of 83 patients were included; 21 (25%) met criteria for ILDprog over 12 months. The female-to-male ratio was lower among patients with ILDprog compared with ILDnoprog(3.2:1 vs. 4.2:1). Median age at onset of the first non-Raynaud manifestation tended to be lower in patients with ILDprog (6.4 vs. 10.6 years, $p$=0.089), and median disease duration at inclusion was longer (3.4 vs. 2.1 years, $p$=0.071). Autoantibody profiles did not differ between groups. At cohort inclusion, patients with ILDprog more frequently had elevated C-reactive protein levels (22% vs. 8%, $p$=0.089), lower median modified Rodnan skin scores (7 vs. 14.5, $p$=0.011), and less frequent Gottron papules (14% vs. 35%, $p$=0.074). At 12-month follow-up, oral sicca symptoms were significantly more common among patients with ILDprog (25% vs. 2%, $p$=0.007). A significantly higher proportion of these patients had FVC <80% predicted (71% vs. 14%, $p$<0.001) and DLCO <80% predicted (80% vs. 46%, $p$=0.025). Gastroesophageal reflux was more frequent among patients with ILDprog (52% vs. 34%). (p n.s). No significant differences were observed in other organ systems.
**Conclusion**
Using a recently proposed consensus definition, progILD in jSSc was associated with distinct clinical characteristics. These findings describe features observed in patients who met criteria for progILD rather than true predictors. Larger longitudinal studies are required to determine whether these characteristics may serve as predictive markers for progILD in jSSc.

**Table 1 (Abstract PT086)** Progressive interstitial lung disease was defined by at least one of the following

| Items defining progressive interstitial lung disease |
|---|
| A, worsening lung fibrosis on chest computed tomography (CT) |
| B, absolute decline in forced vital capacity (FVC) ≥5% predicted or absolute decline in diffusing capacity for carbon monoxide (DLCO) ≥10% predicted; or |
| C, new or worsening respiratory symptoms |

**Disclosure of interest**
None declared.

## PT087

### Disease-specific transcriptional drivers of the composite interferon score: a cross-diagnostic analysis spanning monogenic interferonopathies, multifactorial rheumatic disease, and healthy controls

A. Pin[1], S. Pastore[1], E. De Martino[1], V. Boz[1], M. Girardelli[1,*], A. Agrusti[1], E. Valencic[1], A. Taddio[1,2], A. Tommasini[1,2], A. Tesser[1]
[1]Institute for Maternal and Child Health IRCCS Burlo Garofolo; [2]University of Trieste, Trieste, Italy
**Correspondence:** M. Girardelli
*Pediatric Rheumatology 2026*, **24(S1):**PT087

**Introduction**
The six-gene type I interferon (IFN-I) score [1] is a validated composite measure of IFN pathway activation used across monogenic Interferonopathies and multifactorial autoimmune diseases. Its composite nature may obscure disease-specific transcriptional patterns.
**Objectives**
We hypothesized that individual IFN-stimulated genes (ISGs) contribute differentially to the composite score depending on the underlying diagnosis.
**Methods**
We performed quantitative measurement of the six-gene IFN score (*IFI27*, *IFI44L*, *IFIT1*, *ISG15*, *RSAD2*, *SIGLEC1*) in 177 individuals: 157 patients spanning 14 diagnostic categories and 20 healthy controls (HC). To identify which gene best reflects disease-specific IFN-I activity within each IFN-driven group, we calculated Spearman rank correlations between each individual ISG and the median of the remaining five genes (leave-one-out approach) to avoid mathematical circularity. Kruskal-Wallis and Dunn post-hoc tests with Benjamini-Hochberg correction were used for between-group comparisons with healthy controls. R was used for statistical analysis.
**Results**
The selection of IFN-associated disease groups as disease controls was supported by their significantly higher scores when compared to inflammatory controls (Celiac disease, intestinal bowel diseases, recurrent fevers, hyper-IgD conditions), which grouped at low score levels similar to HC. Interestingly, IFN-I was unexpectedly elevated in recurrent fevers, Behçet disease, and arthritis compared to HC ($p$<0.01, $p$<0.01, and $p$<0.05, respectively), indicating an underappreciated IFN-I component in these conditions. *IFI27* was shown to be the predominant contributor in SLE (74.7%) and Sjögren (53.3%) according to gene contribution analysis, while HC showed a nearly similar distribution among the six genes (~17% per gene). Disease-specific driver genes were found using leave-one-out Spearman analysis: *ISG15* in SLE, *IFI44L* in monogenic Interferonopathies and dermatomyositis, and *IFIT1* in perniosis, indicating different patterns of IFN-I gene regulation across IFN-driven disorders.
**Conclusion**
The composite IFN-I score masks substantial intra-score heterogeneity. *ISG15*-driven, *IFI44L*-driven, and *IFIT1*-driven patterns define mechanistically distinct subsets of IFN-mediated disease. The balanced HC ISG profile underscores that IFN-I activation is qualitatively — not merely quantitatively — different from physiological baseline signaling.

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**Disclosure of interest**
None declared.

## PT088

### Global disparities in pediatric rheumatology practice: a cross-sectional survey of disease burden, diagnostic access, and treatment availability across 55 countries

A. Schvartz[1,*], S. Melo-Gomes[2], J. Perfetto[3], V. Torregiani[4], A. I. Rebollo-Giménez[5], J. Swart[6], S. Ozen[7], E. Sag[7] on behalf of PReS EMERGE - EMErging RheumatoloGists and rEsearchers on behalf of PReS EMERGE - EMErging RheumatoloGists and rEsearchers

[1]Imagine Institute, Paris, France; [2]Guy's and St Thomas' NHS Foundation Trust, London, UK; [3]NYU Langone Health, New York, USA; [4]Centro de Inmunología Clínica, Buenos Aires, Argentina; [5]UOC Reumatologia e Malattie Autoinfiammatorie, IRCCS Istituto Giannina Gaslini, Genoa, Italy; [6]Wilhelmina Children's Hospital/UMC Utrecht, Utrecht, Netherlands; [7]Department of Pediatric Rheumatology, School of Medicine, Hacettepe University, Ihsan Dogramaci Children's Hospital, Ankara, Türkiye
**Correspondence:** A. Schvartz
*Pediatric Rheumatology 2026*, **24(S1):**PT088

**Introduction**
Pediatric rheumatology (PR) guidelines are largely developed by physicians in high-income settings, yet their applicability across diverse economic contexts remains untested.
**Objectives**
To characterize and compare PR clinical practice patterns – including disease diagnosis and follow-up frequency, investigation access, multidisciplinary resources, and treatment availability – across countries stratified by World Bank (WB) income classification.
**Methods**
A structured, cross-sectional online survey was distributed through the EMERGE network. Questions covered diagnosis and follow-up frequency for 11 disease categories, investigation access, multidisciplinary team composition, and pharmacological treatment availability. Respondents were classified into three WB income groups: lower-middle (LMI), upper-middle (UMI), and high income (HI). Between-group comparisons used Kruskal-Wallis tests with epsilon-squared ($\varepsilon^2$) effect sizes; post-hoc pairwise testing used Dunn's method with Benjamini-Hochberg correction.
**Results**
A total of 302 pediatric rheumatologists from 55 countries completed the survey (HI 62%, UMI 29%, LMI 8%; Europe 48%, Asia 28%, North America 12%, South America 8%). Significant differences in diagnosis frequency across WB groups were observed for all 11 disease categories (all $p<0.001$). Vasculitis showed the largest effect size ($\varepsilon^2=0.196$): it was diagnosed at least weekly by 14.9% of HI vs. 4.2% of LMI respondents. Non-inflammatory musculoskeletal conditions also differed markedly ($\varepsilon^2=0.182$). Follow-up frequency patterns were significantly different in 8 of 11 disease categories. Regarding diagnostic investigations, real access to genetic testing was 92.0% in HI vs. 29.2% in LMI (theoretical availability 96.3% vs. 62.5%; access gap: 4.3% vs. 33.3%). Cost was the primary barrier to genetic testing in 70.8% of LMI vs. 41.0% of HI respondents. MRI was practically universally available in HI (100%) but present in 87.5% of LMI centres, with whole-body MRI accessible in 66.7% of LMI vs. 79.8% of HI. Treatment disparities were substantial. Real availability of anti-tumour necrosis factor (anti-TNF) biologics was 45.8% in LMI vs. 98.4% in HI; interleukin-1 (IL-1) inhibitors 12.5% vs. 85.1%; Janus kinase (JAK) inhibitors 16.7% vs. 77.1%. Drug funding through public insurance or government programmes was available to 97.9% of HI respondents compared with 62.5% of LMI, while out-of-pocket payment was the primary funding source for 83.3% of LMI vs. 16.0% of HI respondents.
**Conclusion**
Substantial and systematic differences in disease case-mix, diagnostic capacity, and treatment access exist across WB income groups. These findings challenge the universal applicability of current PR guidelines and call for the development of resource-stratified recommendations that reflect the realities of practice in lower-income settings.
**Disclosure of interest**
None declared.

## PT089

### Recognising interstitial lung disease in a cohort of children with trisomy 21 with or without inflammatory arthritis

D. McLaughlin[1,*], K. Gallagher[1], C. Foley[1], R. O'Reilly [2], S. Javadpour[2], E. MacDermott[1], O. Killeen[1]
[1]Paediatric Rheumatology; [2]Paediatric Respiratory, Children's Health Ireland, Crumlin, Dublin, Ireland
**Correspondence:** D. McLaughlin
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**Introduction**
Children with Trisomy 21 (T21) are at increased risk of a variety of autoimmune disorders including Down syndrome associated Arthritis (DA) (affecting 1 in 50 children); Interstitial lung disease (ILD) remains poorly described in T21.
**Objectives**
To describe a cohort of children with ILD in T21 with or without DA.
**Methods**
We undertook a case review of children with T21 attending our service for assessment of DA. All children with T21 and a confirmed diagnosis of ILD were included. Data collected included clinical features, radiological, laboratory (including Interferon gene signature testing) and histopathology findings in addition to further management and disease outcomes.
**Results**
Four cases demonstrated CT thorax findings concerning for ILD including the presence of diffuse ground-glass opacification, septal thickening and sub-pleural cysts (Cases 1-4). Genetic panel testing for ILD

| Case | Gender | Age (ILD diagnosis) | ILD Presenting Features | Co-existing Arthritis (DA) & Features | Lung Biopsy | Autoantibody Profile (Significant) | IFN Signature (Elevated) | Treatment |
|---|---|---|---|---|---|---|---|---|
| 1 | M | 4 Months | Oxygen dependency, increased work of breathing | Not present | Cysts, necro-inflammatory debris | Nil | Type 1: IFI27 IFI44L IFIT1 | IV methylprednisolone (1.5 mg-3 mg/kg/day) PO Ruxolitinib 5 mg BD |
| 2 | F | 3 Years | Clubbing | Not present | Not performed | Nil | Pending | Nil |
| 3 | M | 1 Month | Oxygen dependency & later clubbing | Yes: Small joint (hand) swelling, worsening behaviour | Lymphocytic inflammation, fibrosis, cysts | Nil | Type 1: IFI27 IFI44L IFIT1 RSAD2 SIGLEC1 Type 2: CXCL9 CXCL10 | IV methylprednisolone (15 mg/kg/day 3/7 for 3/12) PO Tofacitinib 4 mg BD |
| 4 | F | 8 Years | Clubbing, cough, haemoptysis | Yes: Limp, Diffuse joint swelling | Pending | Rheumatoid factor positive, ANA positive (>1:160) | Pending | IV methylprednisolone (30 mg/kg/day 3/7) PO prednisolone (2 mg/kg/day ongoing) SC Adalimumab 40 mg fortnightly |

(Case 1-3) did not detect a pathogenic mutation. Case 1 died due to recurrent pulmonary haemorrhages and increasing ventilatory requirements. Case 2 remains clinically stable and has not yet required treatment for ILD. Case 3 demonstrated improvement in both CT thorax findings and arthritis following treatment with tofacitinib. Case 4 received adalimumab for arthritis with treatment planning for further ILD management ongoing whilst awaiting lung biopsy results with partial response achieved with corticosteroids.

**Conclusion**

ILD in T21 has been poorly described. This cohort presents two cases of both ILD and DA in T21 which to our understanding has not been previously described. Elevated Type 1 interferon activity in ILD may represent potential therapeutic targets with JAK inhibition. Corticosteroid therapy formed a mainstay of treatment in this cohort. Increased vigilance for evidence of respiratory disease (specifically clubbing) at each clinical encounter is important. Further research is required to define the immunological mechanisms underpinning ILD and DA in T21.

**Disclosure of interest**

None declared.

## PT090

### Evaluating Stern's flow-charts as a novel diagnostic tool for pediatric Sjögren's disease: a retrospective cohort study

E. Marrani[1], M. De Angelis[1], L. Gatti[1,*], I. Maccora[1], M. V. Mastrolia[1], I. Pagnini[1], T. Giani[1], G. Simonini[1]
[1]Pediatric Rheumatology, Meyer Children's Hospital IRCCS, Firenze, Italy
**Correspondence:** L. Gatti
*Pediatric Rheumatology 2026*, **24(S1):**PT090

**Introduction**

The 2016 American College of Rheumatology/European League Against Rheumatism (ACR/EULAR) criteria demonstrated poor sensitivity in pediatric cohorts. As early diagnosis of cSjD may provide a unique opportunity to modify disease course, novel tools are required to reduce diagnostic delays and allow accurate follow-up and treatment of these patients. Recently, Stern et al. proposed pediatric-specific diagnostic flowcharts structured around three clinical patterns. These algorithms allow classification as definite, probable, monitoring, or excluded disease and incorporate longitudinal reassessment. Given the need for pediatric-adapted diagnostic frameworks, we aimed to externally evaluate Stern's flowcharts in a real-world cohort and compare their performance with established criteria.

**Objectives**

To evaluate the diagnostic efficacy of the novel paediatric Sjögren's disease (cSjD) flowcharts proposed by Stern, comparing them against the 2016 ACR/EULAR and 1999 Bartůňková criteria.

**Methods**

We conducted a retrospective study of 35 paediatric patients with a clinical diagnosis of Sjögren's disease at Meyer Children's Hospital IRCCS, Firenze, Italy. Diagnosis was histologically confirmed via minor salivary gland biopsy in 91.4% of cases. We applied the ACR 2016 and Bartůňková 1999 criteria and compared their performance with the Stern flowcharts at baseline and during follow-up.

**Results**

At baseline, the diagnostic yield of Stern's flowcharts and Bartůňková criteria was identical, while only 8.6% of patients met the ACR criteria. Application of Stern's longitudinal recommendations during follow-up allowed for an additional 10% of patients to reach a "definite" diagnosis. Notably, the extra-glandular pattern facilitated a higher baseline diagnosis rate and potentially eliminated the need for biopsy in selected cases. Triple concordance across all three criteria was remarkably low at 9.1%.

**Conclusion**

Stern's flowcharts provide a superior diagnostic framework for cSjD compared to ACR and Bartůňková criteria. This algorithm better captures the paediatric disease spectrum and may reduce the need for invasive biopsies.

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**Disclosure of interest**

None declared.

## PT091

### Altered acute inflammatory response to maximal treadmill exercise in children and adolescents with juvenile idiopathic arthritis

K. Risum[1,2,*], J. Stamnæs[1,3], H. Alexanderson[4,5], Ø. Molberg[1,3], H. Sanner[1,3]
[1]Oslo University Hospital; [2]Oslo Metropolitan University; [3]University of Oslo, Oslo, Norway; [4]Karolinska University Hospital; [5]Karolinska Institutet, Stockholm, Sweden
**Correspondence:** K. Risum
*Pediatric Rheumatology 2026*, **24(S1):**PT091

**Introduction**

Physical activity is recommended for children with juvenile idiopathic arthritis (JIA), is not associated with increased disease activity and may exert anti–inflammatory effects over time. However, acute inflammatory changes after exercise are less investigated in this population.

**Objectives**

To investigate acute inflammatory and muscle enzyme responses to a single session of maximal exercise in children and adolescents with JIA compared with healthy controls.

**Methods**

In this quasi–experimental study, serum samples were collected before and 60–120 min after a maximal treadmill test to volitional exhaustion performed during a single exercise session in 47 patients with JIA (mean age 13.6 [SD 2.7] years; 85% girls) and 41 age and sex–matched healthy controls (mean age 14.0 [SD 2.2] years; 78% girls). Analyses of muscle-related enzymes (AST, ALT, LDH, CK, CKMB, myoglobin) and CRP were performed by Department of Medical Biochemistry, Oslo University Hospital, while inflammation-related proteins were assessed by the Olink Target 96 Inflammation panel. Within–group changes were analysed using paired student's t-tests, and significant protein changes were identified after adjustment for multiple testing. Muscle enzyme responses were analysed using paired non–parametric tests.

**Results**

Maximal exercise induced significant inflammatory protein changes in both groups. In healthy controls, 22 inflammatory proteins were significantly altered ($p<0.05$), of whom 10 proteins displayed increased and 12 decreased abundance after exercise. In contrast, patients with JIA demonstrated a broader spectrum of altered proteins, with predominance of increased abundance. Of the 40 proteins altered in the JIA cohort, 38 increased in abundance after exercise. Among proteins increased in JIA were cytokines and chemokines involved in leukocyte recruitment and inflammatory signalling, such as IL–6, IL–7, IL–8 and members of the CXCL and MCP families. Muscle enzymes CK and myoglobin increased similarly in both groups, indicating comparable mechanical muscle strain, while increases in AST and LDH were significantly greater in JIA ($p<0.05$). Overall muscle enzyme levels, including AST and LDH, remained within normal reference ranges in both groups. CRP remained unchanged.

**Conclusion**

This study suggests differences in the acute systemic inflammatory response to maximal exercise between patients with JIA and healthy controls, despite similar mechanical muscle strain, similar muscle enzyme levels and lack of systemic acute phase response. Overall, while these findings support current recommendations encouraging physical activity in JIA, they provide new insights on acute immune regulation following high intensity physical stress in patients with JIA and matched controls. Further research is needed to better understand the clinical relevance of this inflammatory response.

**Disclosure of interest**

None declared.

## PT092

### The relationship between health literacy and self-efficacy, caregiver burden, quality of life, and psychological status in parents of children with juvenile idiopathic arthritis: pilot study

B. C. Balkisli[1,*], S. S. Gumus[2], E. Gur Kabul[3], B. Basakci Calik[2], S. Turkucar[4]
[1]Physiotherapy and Rehabilitation, Istanbul Okan University, Istanbul; [2]Faculty of Physiotherapy and Rehabilitation, Pamukkale University, Denizli; [3]Physiotherapy and Rehabilitation, Usak University, Usak; [4]School of Medicine, Department of Pediatric Rheumatology, Pamukkale University, Denizli, Türkiye
**Correspondence:** B. C. Balkisli
*Pediatric Rheumatology 2026*, **24(S1):**PT092

**Introduction**
Juvenile Idiopathic Arthritis (JIA) is a chronic rheumatic disease requiring long-term management. Health literacy (HL), defined as the ability to access, understand, and use health information, is important for treatment adherence and health outcomes. Studies have shown that nearly one in four parents of children with JIA has inadequate or problematic HL. Since parents play a central role in disease management, their HL may also be associated with self-efficacy, caregiver burden, quality of life, and psychological status.
**Objectives**
The aim of this study was to examine the relationship between HL and sociodemographic characteristics, self-efficacy, caregiver burden, quality of life and psychosocial status in parents of children with JIA.
**Methods**
A total of 31 parents (mean age=43.48±5.62 years, 28 female) were included in the study. Data were collected using a sociodemographic form including parent and child age, educational level (primary, middle, high school, bachelor's, postgrad), economic status (income deficit, income balance, income surplus), and place of residence (city, district, village). Health literacy was assessed using the Adult Health Literacy Scale (AHLS), parental self-efficacy using the Parent's Arthritis Self-Efficacy Scale (PASE), caregiver burden using the Caregiver Reaction Assessment (CRA subscales; impact on finances, lack of family support, impact on health, impact on schedule, and caregiver self-esteem), quality of life and family functioning using the PedsQL™ Family Impact Module, and psychological status using the Hospital Anxiety and Depression Scale (HADS). Relationships between continuous variables were analyzed using Spearman correlation analysis. Correlation coefficients were categorized as weak (0.10–0.39), moderate (0.40–0.69), and strong (0.70–1.00). Differences in AHLS scores across categorical groups were evaluated using the Mann–Whitney U test and Kruskal–Wallis test.
**Results**
The mean AHLS score was 15.42±4.11. AHLS showed a moderate correlation with PASE ($r$=0.478, $p$=0.006) and weak correlations with child age ($r$=-0.355, $p$=0.050) and CRA-caregiver self-esteem subscale ($r$=0.345, $p$=0.058). No significant differences in AHLS were found according to educational status, economic status, or place of residence.
**Conclusion**
HL is an important determinant of self-efficacy among parents caring for children with JIA. As parents' ability to access and understand health information improves, their perceived competence in managing their child's chronic condition also increases. Since HL appears to be independent of sociodemographic factors, it may be considered a modifiable skill. Therefore, integrating HL support into multidisciplinary JIA care can enhance parents' self-efficacy and reduce the psychosocial burden of long-term care.

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**Disclosure of interest**
None declared.

## PT093

### Inflammatory disease control and patient-reported disease burden in juvenile idiopathic arthritis: 25-year trends from the national paediatric rheumatology database

F. Milatz[1,2,3,*], J. Klotsche[1], J. Brunner[4,5], N. Brück[6], M. Fasshauer[7], D. Föll[8], I. Foeldvari[9], M. Haller[10], T. Kallinich[1,2,3], P. Ladehof[11], J. Kümmerle-Deschner[12], H. Lausmann[13], A. Meyer-Bahlburg[14,15], K. Mönkemöller[16], P. T. Oommen[17], C. Reiser[12,18], M. Rühlmann[19], S. Schmidt-Schuchert[15,20], F. Speth[21], S. Tomczyk[15,22], P. von Bismarck[23], D. Windschall[24,25], K. Minden[1,2,3] on behalf of NPRD study group
[1]German Rheumatology Research Center Berlin; [2]Charité - Universitätsmedizin Berlin; [3]German Center for Child and Adolescent Health (DZKJ), Berlin, Germany; [4]Medizinische Universität Innsbruck, Innsbruck; [5]Danube Private University, Krems, Austria; [6]Faculty of Medicine and University Hospital Carl Gustav Carus, Dresden; [7]Klinikum St. Georg, Leipzig; [8]Universitätsklinikum Münster, Münster; [9]Hamburg Centre for Pediatric and Adolescent Rheumatology, Hamburg; [10]Kinderarztpraxis Gundelfingen, Gundelfingen; [11]Patient representative, German Rheumatology Research Center Berlin, Berlin; [12]University Hospital Tuebingen, Tuebingen; [13]Children's Hospital St. Marien gGmbH, Landshut; [14]Rostock University Medical Center, Rostock; [15]German Center for Child and Adolescent Health (DZKJ), Greifswald/Rostock; [16]Kliniken Köln - Kinderkrankenhaus Amsterdamer Str, Cologne; [17]Heinrich Heine University Düsseldorf, Düsseldorf, Germany; [18]Landeskrankenhaus Bregenz, Bregenz, Austria; [19]Medical practice for pediatricy and pediatric rheumatology Dr. M. Rühlmann, Göttingen; [20]University of Greifswald, Greifswald; [21]University Medical Center Hamburg-Eppendorf, Hamburg; [22]University Medicine Rostock, Rostock; [23]University Hospital Schleswig-Holstein, Kiel; [24]St. Josef Stift Sendenhorst, Sendenhorst; [25]University of Halle-Wittenberg, Halle, Germany
**Correspondence:** F. Milatz
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**Introduction**
Over the last two decades, treatment options for juvenile idiopathic arthritis (JIA) have substantially improved, resulting in better inflammatory disease control. However, it remains unclear to what extent patient-reported disease burden has changed over the same period.
**Objectives**
To investigate temporal trends in physician- and patient-reported disease outcomes (PROs), their discordance, and associated correlates.
**Methods**
Annual cross-sectional data from children and adolescents with JIA enrolled in the German National Paediatric Rheumatology Database (NPRD) between 2000 and 2024 were analysed. Outcomes were reported by parents (≤11 years) or patients themselves (≥12 years). Assessed variables included active joint count, physician global assessment of disease activity (PhGA; NRS 0–10), cJADAS-10, patient-/parent-reported disease activity, well-being, pain, fatigue (all NRS 0–10), and functional disability (CHAQ). Time trends and sex effects were analysed using adjusted linear regression models. Using 2024 data, multivariable logistic regression identified correlates of higher patient- versus physician-reported disease activity in adolescents.
**Results**
Based on 132,902 registry records (42,657 patients; mean age 12 ± 5 years; 63% female; 49% oligoarthritis), the proportion of patients with clinically inactive disease (cJADAS10) increased from approximately 30% in 2000 to around 50% in 2024; similarly, the proportion with PhGA = 0 increased to around 55%, accompanied by fewer active joints. In contrast, no comparable improvement was observed for patient-reported outcomes. The proportion of patients without pain (NRS = 0) decreased (≥12 years: 43.7% vs. 37.1%; <12 years: 58.2% vs. 48.7%), as did well-being (≥12 years: 33.3% vs. 26.7%; <12 years: 44.9% vs. 35.0%) and fatigue (≥12 years: 52.7% vs. 42.1%; <12 years: 63.1% vs. 57.3%). Adolescents (≥12 years) consistently reported a higher disease burden than younger children (≤11 years). Regression analyses confirmed decreasing PhGA values over time (β = -0.026; $p < 0.0001$ in those ≥12 years), whereas patient-reported outcomes showed no corresponding improvement and unfavourable trends particularly during the last decade (e.g. pain β = 0.015; well-being β = 0.017; fatigue β = 0.030; all $p <$

0.0001). In the 2024 analysis ($n$=4,417), clinically relevant discordance (≥2 points) was observed in 29.3% of cases and more often reflected higher patient-reported than physician-assessed disease activity (22.7% vs. 6.6%). Among adolescents, higher pain intensity (OR 1.70, 95% CI 1.53–1.89) and CHAQ scores (OR 1.81, 95% CI 1.02–3.19) were associated with greater discordance.

**Conclusion**

The discrepancy between physician-assessed disease control and patient-reported disease burden highlights the importance of integrating PROs into multi-target treatment strategies.

**Disclosure of interest**

None declared.

## PT094

### Sleep duration and insomnia symptoms in children and adolescents with juvenile idiopathic arthritis

F. Milatz[1,2,3,*], O. Saidi[4], J. Klotsche[1], T. Kallinich[1,2,3], R. Berendes[5], N. Brück[6], F. Dressler[7], H. Girschick[8], J.-P. Haas[9], A. Holl-Wieden[10], G. Horneff[11,12], A. Hospach[13], M. Hufnagel[14], T. Krickau[15], P. Ladehof[16], C. Rietschel[17], S. Schmidt-Schuchert[18,19], S. Tomczyk[19,20], R. Trauzeddel[21], F. Weller-Heinemann[22], H. Baumeister[23,24,25], D. Windschall[26,27], K. Minden[1,2,3] on behalf of NPRD study group

[1]German Rheumatology Research Center Berlin; [2]Charité - Universitätsmedizin Berlin; [3]German Center for Child and Adolescent Health (DZKJ), Berlin, Germany; [4]Toulon University, Toulon, France; [5]Kinderkrankenhaus Sankt Marien, Landshut; [6]University Hospital Carl Gustav Carus, Dresden; [7]Medizinische Hochschule Hannover, Hannover; [8]Klinikum im Friedrichshain, Berlin; [9]German Centre for Pediatric and Adolescent Rheumatology, Garmisch-Partenkirchen; [10]University Children's Hospital Wuerzburg, Wuerzburg; [11]Asklepios Klinik Sankt Augustin, Sankt Augustin; [12]University of Cologne, Cologne; [13]Klinikum Stuttgart, Stuttgart; [14]University of Freiburg, Freiburg; [15]FAU Erlangen-Nürnberg and Universitätsklinikum Erlangen, Erlangen; [16]Patient representative, German Rheumatology Research Center Berlin, Berlin; [17]Clementine Children's Hospital Frankfurt, Frankfurt; [18]University of Greifswald, Greifswald; [19]German Center for Child and Adolescent Health (DZKJ), Greifswald/Rostock; [20]University Medicine Rostock, Rostock; [21]Helios Kliniken Berlin-Buch, Berlin; [22]Klinikum Bremen-Mitte, Bremen; [23]Ulm University, Ulm; [24]German Center for Mental Health (DZPG), Munich; [25]German Center for Child and Adolescent Health (DZKJ), Ulm; [26]St. Josef-Stift Sendenhorst, Sendenhorst; [27]Universität Halle-Wittenberg, Halle, Germany

**Correspondence:** F. Milatz

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**Introduction**

Sleep disturbances are increasingly recognized as clinically relevant in juvenile idiopathic arthritis (JIA). However, data on insomnia symptoms and adherence to age-specific sleep duration recommendations remain limited.

**Objectives**

To assess the prevalence of insomnia symptoms and sleep duration below age-specific recommendations in children and adolescents with JIA and to examine their clinical and psychosocial correlates.

**Methods**

Cross-sectional data from the German National Paediatric Rheumatologic Database 2024 including patients aged ≤17 years were analysed. Sleep was assessed via self-report (≥12 years) or parent report (≤11 years). Insomnia symptoms were defined as difficulty initiating and/or maintaining sleep occurring ≥3 times/week during the past 4 weeks. Sleep duration was classified according to age-specific American Academy of Sleep Medicine recommendations. Four sleep phenotypes were defined based on insomnia symptoms (yes/no) and sleep duration (recommendations met/not met). Multinomial logistic regression models involved demographic variables, physician global assessment of disease activity (PGA), active joint count, and patient-reported outcomes including pain, fatigue, functional ability (C-HAQ), physical activity, and depressive (PHQ-9) as well as anxiety symptoms (GAD-7).

**Results**

Data from 5,105 patients (aged ≤11 years: $n$ = 2,561; aged ≥12 years: $n$ = 2,544) across 61 paediatric rheumatology centres could be considered (mean age 11±4 years; disease duration 5±4 years; 68% female; 52% oligoarthritis). Sleep duration below recommendations was observed in 22% ($n$= 556) of patients aged ≤11 years and 39% ($n$= 988) of those aged ≥12 years, while 18% reported insomnia symptoms in both age groups. Overall, 60% ($n$=3,071) had neither insomnia symptoms nor sleep duration below recommendations, 22% had sleep duration below recommendations only, 10% insomnia symptoms only, and 8% both. Patients with both insomnia symptoms and sleep duration below recommendations reported the highest pain (3.9±3.0), fatigue (4.2±3.3), and patient-reported disease activity scores (3.4±3.2), as well as the lowest physical activity levels (all $p$<0.05). In adolescents, older age (OR 1.33; 95% CI 1.13–1.57), higher fatigue (OR 1.19; 95% CI 1.09–1.30), and elevated depressive/anxiety symptoms (OR 8.49; 95% CI 4.91–14.68) were associated with the combined sleep phenotype. No significant associations were found for physician-assessed disease activity measures (PGA and active joint count).

**Conclusion**

Approximately one in five children and more than one in three adolescents with JIA do not meet age-specific sleep duration recommendations, while nearly one in five report clinically relevant sleep disturbances. The findings highlight the clinical relevance of sleep in JIA and support routine assessment in patients with increased subjective symptom burden, even in the absence of clinically active disease.

**Disclosure of interest**

None declared.

## PT095

### Unmet needs and co-designed strategies to improve patient and family experience in juvenile idiopathic arthritis and associated uveitis: a mixed-methods study within the PAVE multinational framework

L. Martinez-Mifsut[1], J. Vinyets-Rejon[1], E. Puiggros-Ruiz[1], A. Reyes-Segura[1], M. Jabalera-Contreras[1], J. Calzada-Hernandez[2], S. Carriqui-Arenas[2], C. Estepa-Guillen[2], A. M. Zacarias-Crovato[2], V. Bittermann[2], R. Bou-Torrent[2], E. Iglesias-Jimenez[2], J.-M. Mosquera[2], P. Ramírez-Rodríguez[2], J. Diaz-Cascajosa[3], M. Morales-Ballus[3], E. Pedemonte-Sarrias[3,4], M. Salvado-Gavalda[5], M. Gonzalez-Martinez[6], M. Crespo-Bosch[7], L. Gonzalez-Riesco[8], M. Llorens-Capdevila[8], R. Cardona-Hernandez[9], J. Anton[2,10,*] on behalf of On behalf of the PAVE (Producing an Arthritis Value-Framework with Economic Evidence) consortium

[1]Quality and Patient Experience; [2]Paediatric Rheumatology Department; [3]Ophthalmology Department, Hospital Sant Joan de Deu, Barcelona; [4]Faculty of Medicine, University of Vic - Central University of Catalonia (UVic-UCC), Vic-Manresa; [5]Clinical Management Support Unit; [6]Child Life; [7]Social Work; [8]Mental Health; [9]Endocrinology Department, Hospital Sant Joan de Deu; [10]Universitat de Barcelona, Barcelona, Spain

**Correspondence:** J. Anton

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**Introduction**

This project is part of the multinational PAVE network, aimed at generating evidence on the socio-economic impact of JIA.

**Objectives**

The primary objective was to identify the unmet needs of children, adolescents, and families living with JIA-U. Secondary goals included assessing the disease's impact on QoL, and emotional, social, and economic well-being, defining strategies to address these needs, and proposing initiatives to optimize the patient journey.

**Methods**

A single-center mixed-methods study was conducted at a pediatric hospital in three sequential phases.

**Phase 1, Qualitative (P1)**

Design Thinking–driven research techniques complemented by an initial review of 107 articles. The sample included 15 healthcare professionals (HCP), 2 patient associations (PA), and 62 families (45 patients, 67 parents). Data were collected through 15 observation sessions, 12 in-depth interviews, 30 experience diaries, and 4 age-adapted focus groups (<10, 10–14, 15–18 years, and parents).

**Phase 2, Quantitative (P2)**

An online survey was distributed to 248 families across 4 subgroups (JIA-U, JIA without uveitis, uveitis and diabetes). Additionally, 51 adolescents completed an exploratory questionnaire.

**Phase 3, Co-creation and design (P3)**

Findings were synthesized into archetypes and 3 co-design sessions were held (22 parents, 4HCP and 2PA), followed by 3 prioritization sessions.

**Results**
- **P1 Findings**: The key learning was that the patient experience is fundamentally determined by flare remission (frequency and severity of flares). 4 Opportunity Areas were identified: #1 Managing flare remission, #2 Living with uncertainty, #3 Resolving misunderstanding and #4 Decreasing emotional burden (EB).
- **P2 Findings**: High EB was observed, with no significant differences between pathologies. Multiple linear regression models identified factors significantly associated with higher EB. The models explained 33% of the variance in the overall sample and 42% in the sibling subgroup, with both models statistically significant.
- **P3 Findings**: 4 strategies (S) and 8 initiatives (I) were defined and subsequently prioritized with HCP and PA. Strategies: S#1 Coordination and continuity of care; S#2 Information and support; S#3: Training and self-care; and S#4: Peer support.

**Conclusion**
Quantitative findings showed that practical support, impact on daily life, concern about the child's attitude, and guilt contribute to EB, while qualitative data highlighted disease acceptance as central. These findings highlight that, beyond clinical indicators, subjective and relational dimensions are crucial to understanding the impact of chronic paediatric conditions, particularly in multiple concurrent diagnoses. By tailoring interventions to each patient and family's context, these strategies can meaningfully enhance their quality of life.

**Disclosure of interest**
None declared.

## PT096

### A service evaluation of referrals for joint pain to paediatric rheumatology at a tertiary centre: Alder Hey experience

L. J. Stacey[1], E. Roberts[2,3], C. E. Pain[2,3,*]
[1]Medical School, Edge Hill University, Ormskirk; [2]Department of Child Health, University of Liverpool; [3]Department of Rheumatology, Alder Hey Children's Hospital, Liverpool, UK
**Correspondence:** C. E. Pain
*Pediatric Rheumatology 2026*, **24(S1):**PT096

**Introduction**
Paediatric rheumatology services care for children and young people (CYP) with inflammatory and non-inflammatory musculoskeletal conditions, where timely referral is essential to reduce patient morbidity. Variability in referral content and lack of standardised referral pathways may lead to delays in patient care. The 'Getting It Right First Time' guidelines recommend children with symptoms of inflammatory arthritis are reviewed within 28 days of referral.[1]

**Objectives**
To evaluate referrals for joint pain based upon referrer content, timeframe of referrals and initial clinic outcomes.

**Methods**
Data was retrospectively extracted from referrals for joint pain to new outpatient paediatric rheumatology services from the first week of each month from April 2024-March 2025. Patient demographics, referrer information, generic referral content (e.g., referral question, clinical content), specific referral content (e.g., red flags, functional status) and initial clinic outcomes were assessed. Rejected, inpatient or non-accessible referrals were excluded, as were patients who did not attend clinic.

**Results**
108 patients were included with a modal age group of 6-15 years (78.7%). Referrals were received from primary (31.5%), secondary (31.5%) and tertiary (37%) care. 44.4% of referrals stated a clinical question, of which 45.8% specifically queried inflammatory arthritis, whilst 50% did not state a type of pain. The presenting symptom, chronology, medical history and routine examination were included in 99%, 66.7%, 61.1% and 64.8% of referrals. Routine bloods and imaging were included in 38.9% and 26.7% of referrals. The presence/absence of joint swelling, morning stiffness, red flag features and functional difficulties was reported in 66.7%, 27.8%, 41.7% and 55.6% of referrals. The presence/absence of examination/imaging findings of arthritis and inflammatory markers was included in 64.8%, 29.6% and 38.9% of referrals. Referrals triaged as routine ($n = 74$) and urgent referrals ($n = 30$) took a mean±SD of 126.1±44.9 days and 33.3±23.1 days, with 4 referrals triaged to specialist clinics. For patients suspected to have inflammatory arthritis ($n = 10$), patients were reviewed in a median (IQR) of 19.5 (13.5-58.5) days, with 3 patients being reviewed after the recommended 28 day target. Following clinical review, inflammatory arthritis was suspected in 12.5% of patients, with biomechanical pain and chronic pain suspected in 51.3% and 24.4% of patients.

**Conclusion**
Referrals focussed on presenting complaint but often lacked other supporting details, which may hinder effective triage and delay care. Standardised referral pathways may be required to improve referrals and increased education of assessing CYP with joint pain may support clinicians to make appropriate referrals.

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**Disclosure of interest**
None declared.

## PT097

### Infection or flare? Multicentre assessment of physician knowledge in febrile children with rheumatic diseases

M. T. Karadoğan[1,*], S. Valiyeva[2], T. Güzel[1], B. Çinpolat[1], Z. Hazıroğlu Ökmen[1], E. E. Fırat[1], F. Çakmak[1]
[1]Department of Pediatric Rheumatology; [2]Department of Pediatrics, Başakşehir Çam and Sakura City Hospital, İstanbul, Türkiye
**Correspondence:** M. T. Karadoğan
*Pediatric Rheumatology 2026*, **24(S1):**PT097

**Introduction**
Fever in children with rheumatic diseases may signal infection, disease flare, or macrophage activation syndrome. Early clinical decisions are frequently made outside specialized pediatric rheumatology units.

**Objectives**
To assess physicians' knowledge and self-confidence in distinguishing infection, disease flares, and urgent triage needs in febrile children with rheumatic diseases.

**Methods**
This multicentre survey included family medicine and pediatric physicians. The questionnaire included 40 true/false/"do not know" knowledge items, nine clinical scenarios, and five self-efficacy items (0–4). Correct answers were predefined by pediatric rheumatology experts. Internal consistency was assessed with KR-20/Cronbach's α. Group comparisons used Kruskal–Wallis or Mann–Whitney U tests; robust linear regression explored associated factors.

**Results**
Overall, 208 physicians participated: pediatric residents ($n$=74, 35.6%), family medicine physicians/residents/specialists ($n$=71, 34.1%), and pediatricians ($n$=63, 30.3%). Median total knowledge score was 44/49 (89.8%, IQR 81.6–95.9); 100 participants (48.1%) achieved ≥90% correct answers. Internal consistency was high for the basic knowledge section (α=0.91), total knowledge score (α=0.93), and self-efficacy scale (α=0.95). Knowledge differed significantly by professional group ($p$<0.001): pediatricians scored highest (95.9%), followed by pediatric residents (89.8%) and family medicine physicians (81.6%). The lowest domain scores were observed in SLE-related fever differentiation (76.9%), FMF versus infection distinction (81.3%), and immunosuppression or biological therapy risks (83.4%). The weakest item was assuming fever was primarily a flare (55.3% correct). Additional gaps included colchicine classification, colchicine continuation during FMF attacks, and avoiding immunosuppression escalation in SLE before excluding infection. In multivariable analysis, pediatrician status (+12.6% points), pediatric residency status (+11.3), recent pediatric rheumatology education (+6.8), applicable written protocols (+8.6), and greater clinical experience (+2.0 per category) were associated with higher knowledge. Self-efficacy did not correlate with knowledge (rho = 0.047, $p$ = 0.504).

**Conclusion**
Overall knowledge was high, yet gaps persisted in SLE fever, FMF mimics, immunosuppression, biological therapy and MAS/sepsis triage. The mismatch between confidence and knowledge suggests that self-perceived preparedness is insufficient as a quality marker. Structured education and implementable fever pathways may support safer first-contact assessment.

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**Disclosure of interest**
None declared.

## PT098

### Army of eyes: scaling paediatric musculoskeletal screening through pGALS training for frontline healthcare workers

C. Scott[1,2,3,*], W. Slamang[2,3], O. Schwegler[3], V. Maduri[4], H. Foster[5] on behalf of Global Health Working Party
[1]Pediatric Rheumatology, University of Ottawa, Ottawa, Canada; [2]Pediatric Rheumatology, University of Cape Town; [3]Tin Soldiers Global, Cape Town, South Africa; [4]Amara Hospital, Tirupati, India; [5]University of Newcastle, Newcastle, UK
**Correspondence:** C. Scott
*Pediatric Rheumatology 2026*, **24(S1):**PT098

**Introduction**
Delayed recognition of paediatric musculoskeletal (MSK) conditions, likejuvenile idiopathic arthritis (JIA), remains a global challenge. Frontline healthcare workers receive little formal MSK training despite frequently assessing children with MSK complaints. The paediatric Gait, Arms, Legs and Spine (pGALS) examination is a rapid, validated screening tool for detecting MSK abnormalities in children and can be taught to non-specialists.[1] pGALS forms part of the PReS-supported Paediatric Musculoskeletal Matters (PMM) educational platform.
**Objectives**
To describe the "Army of Eyes", a paediatric MSK education and training model designed to enable frontline healthcare workers to recognise MSK abnormalities using pGALS and facilitate referral for conditions including JIA.
**Methods**
A multi-format training programme incorporating workshops, webinars and digital resources was delivered to primary healthcare nurses, doctors, physiotherapists and community health workers. Training focused on routine use of pGALS as a structured approach to paediatric MSK screening. Educational content drew extensively on the PReS-supported PMM learning suite, including PMMOnline [2]alongside context-adapted materials. It emphasised recognition of common and serious MSK conditions, referral pathways and safe initial management. Pre- and post-training confidence in MSK assessment was measured. Pilot outcomes included abnormalities detected, referrals generated and diagnoses identified in South African clinics and schools. In India, the programme was integrated into the government-supported Rashtriya Bal Swasthya Karyakram (RBSK) child health screening programme.
**Results**
Eighty-one healthcare workers in South Africa were trained. Baseline confidence in MSK examination among primary care nurses was moderate (mean 4.4 ± 1.8), and 48% were unfamiliar with pGALS. Post-training confidence improved significantly (mean 5.8 ± 1.1; $p$=0.0001). Among 117 children screened in community clinics, 17 abnormalities were identified and 10 referrals generated. Diagnoses included spinal deformities, muscle weakness, JIA, clubhand and Fibrodysplasia Ossificans Progressiva (FOP). In India, pGALS training was implemented across 40 District Early Intervention Centres involving 128 healthcare workers in Andhra Pradesh and Meghalaya. In the Tura region of Meghalaya, 26 children were referred over six months with conditions including clubfoot, developmental dysplasia of the hip, femoral anteversion, scoliosis and osteogenesis imperfecta; all received treatment or were awaiting surgery.
**Conclusion**
The Army of Eyes programme shows that pGALS-based MSK screening can be embedded within frontline training across diverse settings. Leveraging PMM educational resources enables scalable, sustainable capacity building with potential to improve early recognition of JIA and other MSK conditions globally.

**Disclosure of interest**
None declared.

## PT099

### Making methotrexate more tolerable for CYP: a multi-centre QI project

S. Saad Alla[1,*], M. Sim[2], E. Lowrence[3], T. Amin[4], E. Al-Abadi[5]
[1]Paediatrics, Norfolk and Norwich University Hospital, Norwich; [2]Paediatrics Rheumatology, Sheffield Children's Hospital, Sheffield; [3]Paediatrics Rheumatology, Nottingham Children's Hospital, Nottingham; [4]Paediatrics Rheumatology, Leeds Children's Hospital, Leeds; [5]Paediatrics Rheumatology, Birmingham Children's Hospital, Birmingham, UK
**Correspondence:** S. Saad Alla
*Pediatric Rheumatology 2026*, **24(S1):**PT099

**Introduction**
Methotrexate (MTX) is the most widely used conventional disease-modifying anti-rheumatic drug in paediatric rheumatology, but nausea, anticipatory symptoms, and treatment-related distress can affect adherence, experience and treatment continuation. Improving MTX tolerability is a recognised paediatric rheumatology research priority.
**Objectives**
To evaluate support for children and young people (CYP) on MTX across three United Kingdom (UK) paediatric rheumatology services, and assess early impact of targeted quality improvement (QI) interventions at one centre.
**Methods**
Plan-Do-Study-Act (PDSA) methodology was utilised, with relevant themes identified via driver diagram, a YourRheum focus group, and a review of the literature. Identified themes were explored using post-clinic surveys distributed to CYP/parent/carer at two tertiary paediatric rheumatology centres and one district general hospital. SMART goals were formulated across ten domains, with targets set at 90% for all domains with the exception of side-effect frequency (<10%) and awareness of how to contact the healthcare team (100%). Pilot QI interventions including a MTX educational tool-kit for patients and staff posters were implemented at Leeds Children's Hospital after an initial period of data collection.
**Results**
44 surveys were completed across the three centres at baseline (Leeds Children's Hospital $n$=13, Nottingham Children's Hospital $n$=26, James Paget University Hospital $n$=5), with 14 further post-intervention surveys completed at Leeds. Overall, SMART goals were variably met (Leeds: 3/10, Nottingham: 2/10, James Paget: 6/10). CYP/parent knowledge of managing MTX side-effects was sub-target at all sites (mean score 72.8%), representing the most consistent gap. SMART goal targets were universally met for assessment of adherence (mean 95%) and awareness of how to contact the healthcare team (mean 99%). Shared decision-making also scored highly (mean 94%), although was slightly below target at one centre (87%). Mean adherence and tolerance scores were 91% and 78% respectively. Side-effects were reported variably (Leeds: 42%, Nottingham: 86%, James Paget: 20%), with management strategies inconsistently discussed (Leeds: 63%, Nottingham: 90%, James Paget: 80%). Following QI interventions, a further SMART goal was met (Leeds: 4/10) with lower side-effect frequency (42.3% vs. 23%), improved MTX knowledge (88% vs. 78%), and side-effect discussion (75% vs. 62.5%).
**Conclusion**
Across different care settings, CYP/parent knowledge of managing MTX side-effects was the most consistent gap. Pilot data from Leeds suggest targeted interventions may improve MTX experience, knowledge, and side-effect discussion. Further PDSA cycles and centre expansion are ongoing.
**Disclosure of interest**
None declared.

## PT100

### Exploring nutritional management in children and adolescents with hypermobile Ehlers Danlos syndrome

A. Gkoutzourelas[1,*], A. Gkouvi[1], E. C. Pardali[1], N.-M. Liapis[1], D. P. Bogdanos[1], M. G. Grammatikopoulou[1]
[1]Department of Rheumatology and Clinical Immunology, Larissa University Hospital, University of Thessaly, Larissa, Greece
**Correspondence:** A. Gkoutzourelas
*Pediatric Rheumatology 2026*, **24(S1):**PT100

**Introduction**
Hypermobile Ehlers-Danlos syndrome (hEDS) is a chronic condition that often presents with musculoskeletal complaints, neurological manifestations, mood disorders and autonomic symptoms, as well as gastrointestinal (GI) symptoms. While hEDS patients have a high prevalence of Disorders of Gut-Brain Interaction (DGBI) and avoidant restrictive food intake disorder (ARFID), evidence regarding nutrition is lacking.
**Objectives**
The purpose of this review is to summarize evidence regarding nutrition for children and adolescents with hEDS.
**Methods**
Three databases (PubMed, Cochrane, clinicaltrials.gov) were searched to identify evidence on GI manifestations and nutritional considerations in hEDS.
**Results**
There is a high prevalence of constipation, dysphagia, dyspepsia and gastroparesis in children and adolescents with hEDS [1]. Restrictive dietary patterns and inadequate micronutrient intake have been reported in adult cohorts with hEDS, while similar evidence for children is not available [2]. Reported deficiencies include low fibre and inadequate vitamin B and vitamin D intake. GI symptoms in pediatric hEDS have been linked with dysautonomia, which may contribute to impaired food tolerance and overall food avoidance [3]. Despite this, data regarding malnutrition, sarcopenia, dietary intake evaluation and growth are lacking in this population. Current management should therefore be individualized and multidisciplinary. Adequate hydration, fibre and regular meal patterns are recommended for dysmotility, while smaller, low-fat meals can be considered for gastroparesis [4]. For children with bloating, abdominal pain or irritable bowel syndrome (IBS) symptoms, a supervised low-FODMAP trial may also be considered [5]. In cases where immune-mediated food reactions or celiac disease is suspected, further testing should guide elimination diets rather than broader avoidance.
**Conclusion**
Overall, children with hEDS should receive multidisciplinary care including pediatric rheumatology, gastroenterology, dietitians and psychologists, with a focus on adequate calories, fibre, hydration and micronutrients. Clinical care should include assessment of growth, dietary intake and nutritional deficiencies.

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**Disclosure of interest**
None declared.

## PT101

### Genetically proxied CCL4 levels at the CCR5/CCR2 chemokine receptor cluster are causally linked to juvenile idiopathic arthritis: a proteome-wide mendelian randomization and colocalisation study

M. Soloshenko[1], O. Lomakina[2,*]
[1]ITMO univesity, St. Petersburg; [2]Morozov Children's City Clinical Hospital, Moscow, Russian Federation
**Correspondence:** O. Lomakina
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**Introduction**
The CCR5/CCR2 chemokine receptor cluster has been implicated in JIA through observational studies of synovial T-cell infiltration and allelic association with CCR5-delta32, but formal causal inference linking circulating chemokine levels to JIA risk has not been established.
**Objectives**
To identify circulating inflammation proteins causally associated with JIA using proteome-wide two-sample Mendelian randomization (MR) and Bayesian colocalisation.
**Methods**
We used cis- and trans-pQTL from the SCALLOP-Folkersen plasma proteomics GWAS (80 Olink Inflammation proteins, $n$=3,394) as exposures and the Lopez-Isac 2023 European JIA meta-analysis (3,305 cases / 9,196 controls) as outcome. Instruments were selected at $p < 5\times10\text{-}8$ with LD clumping (r2 = 0.001) and F > 10. IVW was the primary MR estimator; coloc.abf (priors p1 = p2 = 10-4, p12 = 10-5) tested shared causal variants. Bonferroni and BH-FDR correction were applied across 54 analysable proteins. Sensitivity analyses included cis/trans decomposition, leave-one-out, Steiger filtering, prior robustness grids, and orthogonal anchoring in independent pQTL, eQTL, and flow-cytometry QTL datasets.
**Results**
CCL4 (MIP-1-beta) was the sole protein surviving multiple testing correction: trans-only IVW OR = 0.675 (95% CI 0.582-0.782), $p = 1.75\times10\text{-}7$, BH-FDR q = $9.45\times10\text{-}6$, driven by rs113010081 at the CCR5/CCR2 cluster (chr3q21). Colocalisation was strong (PP.H4 = 0.981) and robust across prior sensitivity (PP.H4 = 0.84-1.00). The lead variant independently associates with CCR5 mRNA in whole blood (eQTLGen, $p = 3.4\times10\text{-}50$) and plasma CCL4 in a replication cohort ($p = 1.1\times10\text{-}4$), anchoring the causal chain from variant through CCR5 expression and plasma CCL4 to JIA risk. A second chemokine, CCL2, maps independently to the same receptor cluster, implicating CCR5-mediated chemotaxis rather than a single ligand as the causal node. IL6R served as internal positive control, recovering the tocilizumab direction (OR = 0.90, $p = 0.0025$) with coloc.susie confirming three colocalised signals. Genomic inflation was absent (lambda = 0.879).
**Conclusion**
This is the first proteome-wide pQTL MR applied to JIA, identifying the CCR5/CCR2 chemokine axis as a genetically supported causal node, with CCL4 serving as the strongest circulating proteomic proxy. These findings support further investigation of CCR5/CCR2-axis modulation as a mechanistic and therapeutic hypothesis in JIA.
**Disclosure of interest**
None declared.

## PT102

### Single-cell multiome profiling reveals a non-canonical BATF/STAT1/PRDM1 effector programme in MIS-C with limited IL-12/STAT4 axis engagement

B. Jenko Bizjan[1,2,*], J. Kovac[1,2], A. Smole[3,4], M. Zajc Avramovič[2,5], N. Emeršič[2,5], T. Avčin[2,5]
[1]Clinical Institute for Special Laboratory Diagnostics of the Paediatric Clinic, University Medical Center Ljubljana; [2]Faculty of Medicine, University of Ljubljana; [3]Immunology and Cellular Immunotherapy Group, Department od Genetic Toxicology and Cancer Biology, National Institute of Biology; [4]Biotechnical Faculty, University of Ljubljana; [5]Department of Allergology, Rheumatology and Clinical Immunology of the Paediatric Clinic, University Medical Center Ljubljana, Ljubljana, Slovenia
**Correspondence:** B. Jenko Bizjan
*Pediatric Rheumatology 2026*, **24(S1):**PT102

**Introduction**
Multisystem Inflammatory Syndrome in Children (MIS-C) is a post-SARS-CoV-2 hyperinflammatory disorder dominated by IFN-γ signalling [1], but the transcriptional programmes sustaining this response are undefined.
**Objectives**
To characterise the cellular and chromatin-level transcriptional programmes underlying MIS-C flares using paired longitudinal samples.
**Methods**
Single-cell multiome (RNA-seq + ATAC-seq), bulk RNA-seq, plasma cytokine quantification (LEGENDplex), and whole-exome sequencing were performed on paired flare/remission samples from 10 paediatric MIS-C patients, with bulk RNA-seq validation in an independent cohort of 9. Cell-type-specific differential expression, regulatory network inference (FigR[2]), and Hallmark gene set enrichment (GSEA) were performed.

**Results**
IFN-γ was the most elevated plasma cytokine (~30-fold) yet IL-12p70 and γc-cytokines (IL-2, IL-15) were unchanged, suggesting non-classical drivers. Single-cell multiome identified a non-canonical BATF/STAT1/PRDM1 axis coordinately induced across $CD8^+$ TEM (BATF +1.33, STAT1 +0.79, PRDM1 +1.78), NK (+1.26, +1.07, +1.31), and γδ T cells (+2.40, +1.14, +1.05; all adj.$p$<1e-3), with STAT4 downregulation and IL6ST (gp130) upregulation, indicating gp130-associated signalling in place of IL-12/STAT4. IFNG-AS1 was paradoxically downregulated despite sustained IFNG, with FigR showing chromatin-level decoupling. Checkpoint receptors (CTLA4, HAVCR2, LAG3) were upregulated without NR4A exhaustion factor induction. Hallmark TGF-β signalling was downregulated in cytotoxic effectors ($CD8^+$ TEM NES=−2.05, FDR=0.003; NK, γδ T, CD4 CTL all FDR<0.05) but preserved in naive lymphocytes. Independent bulk RNA-seq confirmed BATF/STAT1/PRDM1/IL6ST upregulation, STAT4 and IFNG-AS1 downregulation, and TGF-β reduction, with high cross-cohort concordance (Pearson $r$=0.96; 7,817/7,819 directionally concordant). Whole-exome sequencing of 1,091 immune genes found no recurrent pathogenic rare variants[3].
**Conclusion**
MIS-C flares are characterised by a non-canonical IFN-γ effector programme anchored by BATF/STAT1/PRDM1 with limited IL-12/STAT4 engagement and chromatin-level IFNG/IFNG-AS1 decoupling. The absence of monogenic drivers supports acquired post-infectious transcriptional reprogramming and nominates JAK1/2 inhibition as a candidate therapy. **These findings position MIS-C as a model for understanding sustained hyperinflammation in post-viral paediatric syndromes, with mechanistic relevance to long COVID and related post-infectious conditions.**

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**Disclosure of interest**
None declared.

## PT103

**Genome sequencing for rare autoinflammatory and autoimmune diseases: experience from the French Genomic Medicine Initiative**

M. Tusseau[1,2,*], M. Broly[1,2], I. Jeru[2], B. Bader-Meunier[3], T. Barba[4], C. David[5], S. Georgin-Lavialle[6], V. Hentgen[7], Y. Jamilloux[8], I. Koné-Paut[9], I. Melki[5,10], F. Rieux-Laucat[11], M.-E. Truchetet[12], M. Herasse[13], E. Hachulla[14], A. C. .[1], S. C. .[2], A. Belot[15], L. Cuisset[2], G. Boursier[1,2]
[1] GCS AURAGEN, Lyon; [2]Laboratoire SeqOIA; [3]Service d'Hématologie-Immunologie et Rhumatologie Pédiatrique, Hôpital Necker-Enfants Malades, AP-HP, Université Paris Cité, Paris; [4]Department of Internal Medicine, Lyon University Hospital, Lyon; [5]Laboratoire Neurogénétique et Neuroinflammation, Institut Imagine, Inserm UMR1163, Université Paris Cité,; [6]Department of Internal Medicine, Sorbonne Université, Hôpital Tenon, AP-HP, Paris; [7]Department of Pediatrics, Centre de Référence des Maladies Auto-Inflammatoires Rares et des Amyloses (CEREMAIA), CH Versailles, Versailles; [8]Department of Internal Medicine, Lyon University Hospital; [9]Paediatric Rheumatology Department and CEREMAIA (Reference Center for Auto-Inflammatory Diseases and Amyloidosis), Bicêtre University Hospital, AP-HP; [10]Department of General Pediatrics, Paediatric Rheumatology Unit, Assistance Publique-Hôpitaux de Paris (AP-HP), Sorbonne Université, FHU INFLAMME,; [11]Laboratoire d'Immunogénétique des Maladies Auto-immunes Pédiatriques, Institut Imagine, Inserm UMR1163, Université Paris Cité, Paris; [12]Department of Rheumatology, UMR5164 ImmunoConcept, Bordeaux University, Bordeaux University Hospital, Bordeaux; [13]Filière des Maladies Auto-immunes et Auto-inflammatoires Rares; [14]Inserm U1286 – INFINITE – Institute for Translational Research in Inflammation; Service de Médecine Interne et Immunologie Clinique; Centre de Référence des Maladies Auto-immunes et Auto-inflammatoires Systémiques Rares de l'Adulte du Nord et Nord-Ouest, Université de Lille, CHU de Lille, Lille; [15]Pediatric Nephrology, Rheumatology, Dermatology Department, Hôpital Femme Mère Enfant, Lyon University Hospital, Lyon, France
**Correspondence:** M. Tusseau
*Pediatric Rheumatology 2026*, **24(S1):**PT103

**Introduction**
The French Genomic Medicine Initiative (PFMG) is a national program launched by the French government to integrate genome sequencing (GS) into standard healthcare. The rare autoimmune and autoinflammatory diseases network ($FAI^2R$) was among the first indications approved.
**Objectives**
The objectives was to evaluate the diagnostic yield and clinical relevance of genome sequencing (GS) in rare autoimmune and autoinflammatory diseases within the French Genomic Medicine Initiative (PFMG).
**Methods**
We retrospectively reviewed all $FAI^2R$ cases analysed since the launch of the PFMG.
**Results**
A total of 203 genomes were analysed by the two national laboratories. The mean age at disease onset was 7 years. Overall, 12% of cases were considered as conclusive. Among these 25 cases, four involved the *IKBKG* gene in the context of NDAS, and two diagnoses were identified in *ALPK1* and *TNFAIP3*. Other diagnoses involved genes recently associated to human diseases, genes associated with novel modes of inheritance or genes not classically associated with immune dysregulation. Syndromic presentations and association with immunodeficiency or polyautoimmunity showed the highest diagnostic yield. In contrast, lupus without features suggestive of interferonopathy and Behçet-like disease were associated with the lowest diagnostic yield.
**Conclusion**
GS provides significant added value in rare autoimmune and autoinflammatory diseases, including as a second-line testing approach. This percentage is probably underestimated, as highlighted by the additional 13% of cases in which either a class 3 variant or a gene of uncertain significance was identified. These inconclusive but non-negative results highlight the need for functional studies to interpret variants. In urgent contexts, first-line GS was made possible through the organization of the two laboratories and had a significant impact on patient management.
**Disclosure of interest**
None declared.

## PT104

**Incorporation of recent selection signals improves variant impact prediction in immune genes**

B. Matthiasardottir[1,2,*], P. Pimpale Chavan[3], I. Aksentijevich[1], D. L. Kastner[1], S. M. Mount[2]
[1]Inflammatory Disease Section, Medical Genetics Branch, National Human Genome Research Institute, National Institutes of Health, Bethesda; [2]Dept. of Cell Biology and Molecular Genetics, University of Maryland, College Park, USA; [3]Department of Pediatric Rheumatology, NH SRCC Children's Hospital, Mumbai, India
**Correspondence:** B. Matthiasardottir
*Pediatric Rheumatology 2026*, **24(S1):**PT104

**Introduction**
The diagnostic yield for rare immune diseases remains disproportionately low at ~20% compared to ~40% for other rare diseases. This reflects challenges in accurately annotating variants in immune genes, where conventional variant impact prediction tools frequently misclassify pathogenic variants as benign. Most tools assume that conservation correlates with functional importance. We hypothesize that selection pressures acting on immune genes disrupt this relationship and increase false negative rates.
**Objectives**
To define immune-specific variant curation guidelines using existing variant impact prediction tools, determine why these tools behave differently in immune disease genes, develop a selection-informed variant impact prediction framework, and improve diagnostic yield in immune-mediated disease.
**Methods**
We analyzed variants across ClinVar, Infevers, HGMD and MaveDB and assessed 40 variant prediction tools using ROC-AUC and balanced PR-AUC. We incorporated recent selection signals with structural and evo-

lutionary features to develop VERA (Variant Effect with Recent Adaptation), an ensemble random forest model.
**Results**
Across all evaluated variant impact prediction tools, immune genes required lower classification thresholds for accurate variant prediction compared to non-immune genes. Optimal REVEL and CADD thresholds were 0.324 and 17.24 in Infevers versus 0.735 and 24.15 in ClinVar, respectively. Thresholds were also significantly lower for variants in genes under recent positive selection, enriched for immune function, suggesting a biological basis for this shift. We developed VERA, a selection-informed ensemble random forest model incorporating recent selection signals. VERA achieved ROC-AUC = 0.992 and PR-AUC = 0.981 on ClinVar variants, ROC-AUC = 0.987 and PR-AUC = 0.976 for variants in inborn errors of immunity associated genes, and ROC-AUC = 0.971 and PR-AUC = 0.949 in Infevers variants, consistently outperforming existing prediction tools. Benchmarking against multiplexed assays of variant effect demonstrated that VERA achieved the highest concordance with experimentally measured functional effects, particularly in positively selected immune genes such as *MEFV* and *NLRP3*.
**Conclusion**
We define immune-specific variant curation guidelines for existing variant impact prediction tools, demonstrate that reduced sensitivity in immune genes is driven in part by recent selection, and introduce a selection-informed model that improves prediction accuracy and variant interpretation. Together, these findings provide a framework for improving diagnostic yield in immune-mediated disease.
**Disclosure of interest**
None declared.

## PT105

### The complosome contributes to aberrant T cell activation in Still's disease

T. Thalheim[1,*], S. Schleifenbaum[1], C. Hinze[1], H. Wittkowski[1], D. Foell[1], C. Kessel[1], E. L. Verweyen[1]
[1]Department of Pediatric Rheumatology and Immunology, University Children's Hospital, Muenster, Germany
**Correspondence:** T. Thalheim
*Pediatric Rheumatology 2026*, **24(S1):**PT105

**Introduction**
Still's disease (SD) is a rare autoinflammatory disorder driven by immune dysregulation. The complement system is integral to the innate immune system and bridges towards adaptive immunity, for example, through intracellular complement (complosome)- mediated Th1 regulation. While recent gene and protein expression data suggest involvement of the complement system in SD (1), its pathomechanistic role remains poorly understood.
**Objectives**
Here, we investigate whether complosome activation and its interplay with IFNγ contribute to SD pathology.
**Methods**
CD4+ and CD8+ T cells from pediatric SD patients and healthy controls were stimulated via the TCR and the complement receptor CD46. IFNγ and IL–10 secretion were quantified by ELISA, while T cell activation markers and CD46 surface expression were assessed by flow cytometry. To explore the mechanism of complosome dysregulation, healthy donor T cells were additionally exposed to recombinant human IL–1β, IL–6 and IL–18 stimulation, mimicking a SD-associated inflammatory cytokine environment.
**Results**
Activation of the complosome robustly induced secretion of IFNγ as well as IL-10 in healthy donor and SD patients' CD4+ and CD8+ T cells. However, in contrast to healthy donor T cells, complosome activation in SD cells revealed a distinct T cell phenotype characterized by significantly reduced IL-10 secretion and CD46 surface expression levels but increased intracellular T-bet expression. An inflammatory SD cytokine environment recapitulated this aberrant complosome activation phenotype in healthy donor CD4+ and CD8+ T cells, resulting in a markedly increased IFNγ/IL–10 ratio.
**Conclusion**
Our findings identify the complosome as a potential driver of aberrant T cell activation in SD and highlight its interaction with IFNγ as a potential amplifier of hyperinflammation.

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**Disclosure of interest**
T. Thalheim: None declared, S. Schleifenbaum: None declared, C. Hinze Speaker Bureau with: Pfizer, H. Wittkowski Speaker Bureau with: Novartis, Takeda, Octapharma, CSL-Behring, D. Foell Grant / Research Support with: Novartis, Pfizer, SOBI, Speaker Bureau with: Chugai-Roche, Novartis, SOBI, C. Kessel Grant / Research Support with: German Research Foundation (DFG, grant number KE 2026/3-1), Novartis, Speaker Bureau with: Novartis, SOBI, E. Verweyen Grant / Research Support with: intramural funding program of Muenster University medical faculty for innovative medical research (IMF, grant number VE112304).

## PT106

### Safety of JAK inhibitors in pediatric rheumatology: a national multicenter study

M. Kardani[1], T. Plessa[1], V. Sgouropoulou[2], M. Tsinti[3], E. Tsitsami[3], D. Maritsi[1,*]
[1]Second Department of Pediatrics, "P & A Kyriakou" Children's Hospital, Athens; [2]Second Department of Pediatrics, 2."Papageorgiou" General Hospital, Aristotle University of Thessaloniki, Greece, Thessaloniki; [3]First Department of Pediatrics, "Aghia Sofia" Children's Hospital, Athens, Greece
**Correspondence:** D. Maritsi
*Pediatric Rheumatology 2026*, **24(S1):**PT106

**Introduction**
Janus kinase inhibitors (JAKi) are increasingly used in pediatric rheumatology for refractory inflammatory and autoinflammatory diseases. However, real-world pediatric safety data remain limited, particularly outside clinical trial settings.
**Objectives**
To evaluate the safety profile and adverse events associated with JAK inhibitor therapy in pediatricpatients followed across three tertiary pediatric rheumatology centers.
**Methods**
This retrospective multicenter observational study included 50 pediatric patients treated with JAKinhibitors, including baricitinib and tofacitinib. Demographic, clinical, and laboratory data were collectedfrom medical records. Adverse events, laboratory abnormalities, concomitant therapies, and previousbiologic exposure were analyzed. Exploratory subgroup analyses according to sex, underlying disease, methotrexate co-treatment, and prior biologic exposure were also performed.
**Results**
Fifty pediatric patients were included (56.0% female, mean age 13.2 ± 3.8 years). Most patients hadjuvenile idiopathic arthritis spectrum disease (56.0%), followed by other inflammatory/rheumaticdiseases (32.0%), autoinflammatory diseases (8.0%), interferonopathies (2.0%), and juveniledermatomyositis (2.0%). Previous biologic exposure was present in 80.0% of patients, while 88.0%received concomitant methotrexate therapy. Overall, 72.0% of patients experienced at least one adverse event during treatment. Infectious adverseevents represented the predominant complication category. Upper respiratory tract infections were themost frequent adverse event, occurring in 35.3% of patients with adverse events, followed bylymphopenia (20.6%), neutropenia (17.6%), elevated transaminases (23.5%), anemia (14.7%), herpeszoster (5.9%), and urinary tract infections (5.9%). Most infections were mild and self-limited. No severe opportunistic infections, thrombosis, malignancy, major cardiovascular events, or infection-related hospitalizations were observed. Patients receiving concomitant methotrexate or previous biologic therapies demonstrated numericallyhigher adverse event frequencies; however, no statistically significant differences were identified.Similarly, no statistically significant differences in adverse event frequency were observed betweenunderlying disease groups. Exploratory sex-based analysis demonstrated more frequent neutropeniaamong male patients ($p < 0.05$).
**Conclusion**
In this multicenter real-world pediatric cohort, JAK inhibitors demonstrated an acceptable safety profile.Adverse events were relatively common but predominantly mild and manageable under regular clini-

caland laboratory monitoring. Infections, particularly upper respiratory tract infections, represented themost common adverse events. Larger prospective multicenter studies are required to define the long-term safety profile of JAK inhibitors in pediatric rheumatic diseases.

**Disclosure of interest**
None declared.

## PT107

### Sembaediagnosis: a superagent model derived from the combination of six diagnostic models. the cutting edge of clinical AI in pediatric rheumatology

E. Aliyev[1,2,*] on behalf of SEMBAeDiagnosis Research & Development Project
[1]Pediatrics, Pediatric Rheumatology Unit, Lokman Hekim University School of Medicine; [2]Informatics, SEMBA LLC, Ankara, Türkiye
**Correspondence:** E. Aliyev
*Pediatric Rheumatology 2026*, **24(S1):**PT107

**Introduction**
We aimed to evaluate the performance of the superagent model—named "SEMBAeDiagnosis"—which builds upon Morgaf and Maverik, closed-loop artificial intelligence models known in the literature for assisting with diagnosis in pediatric rheumatology.
**Objectives**
Data on approximately 1,000 patients were submitted to Superagent. Of these patients, 100 had CNO, 200 had JIA, 100 had SLE, 300 had FMF and other autoinflammatory diseases, and 300 had diagnoses most commonly confused with pediatric rheumatology (such as leukemia, lymphoma, solid bone tumors, vitamin C deficiency, adenovirus urinary tract infections, acute upper respiratory infections, and growth pains).
**Methods**
These are patented products developed by Dr. Emil Aliyev as closed-loop AI models to aid in the diagnosis of pediatric CNO patients with Maverik and pediatric SLE patients with Morgaf. By incorporating CNN/U-net-based image and pixel processing functions into these models, we developed a superagent (master model) capable of making comprehensive decisions by synthesizing images such as WBMRI and kidney biopsies with the patient's existing digitized demographic, clinical, and laboratory data. This model consists of six main models (including Morgaf and Maverik), comprising 500,000,000 data points, over 10,000 artificial neurons, and their infinite connections. When the superagent is asked about a patient in any pediatric rheumatology specialty, it uses these submodels to predict which one can provide the most effective response and generates results accordingly. SEMBAeDiagnosis is a product patented by TURKPATENT.
**Results**
SEMBAeDiagnosis's performance on a mixed dataset comprising data from 1,000 patients was surprisingly successful. Its median accuracy was 92.4% (IQR: 3.6), and its AUC was 0.881. By diagnostic group, it correctly identified all CNO patients, 95% of SLE patients, and 81% of the differential diagnoses. In particular, it correctly diagnosed all patients in groups where critical treatment decisions must be made, such as those with leukemia.
**Conclusion**
SEMBAeDiagnosis is currently in the pilot phase of integration into hospital record systems as a superagent. This is a closed-loop AI superagent that holds significant potential as a paradigm-shifting innovation in the field of clinical AI. Training and expansion using real patient data on an international scale will continue as soon as possible.

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**Disclosure of interest**
None declared.

## PT108

### Disease activity in patients with juvenile idiopathic arthritis on methotrexate therapy: metabolomic profiles

B. Bellich[1,*], P. Dalla Zuanna[1], M. Franzin[1], A. Angeli[2], S. Pastore[1], A. Taddio[1,3], A. Fabretto[1], M. Lucafò[2], G. Stocco[1,3]
[1]IRCCS Burlo Garofolo; [2]Life Sciences; [3]Medical, Surgical and Health Sciences, University of Trieste, Trieste, Italy
**Correspondence:** B. Bellich
*Pediatric Rheumatology 2026*, **24(S1):**PT108

**Introduction**
Juvenile idiopathic arthritis (JIA) is the most common chronic rheumatic disease in the paediatric population, defined by early onset before sixteen years of age persisting for at least six weeks. Its pathogenesis involves a complex interplay of several factors. Although methotrexate (MTX) remains the therapeutic gold standard, approximately 30% of patients fails to respond adequately or discontinue treatment due to adverse drug reactions. MTX efficacy is modulated by folate pathway metabolism, which is influenced by the patient's genetic background and systemic metabolic status; understanding the biochemical features of treatment response may offer a powerful strategy to guide therapeutic decisions and minimize the risk of joint damage.
**Objectives**
Metabolomic profiling aims at identifying predictive biomarkers of treatment response.
**Methods**
Plasma samples from 35 JIA patients undergoing MTX therapy (≥3 months) were stratified by JADAS27 into inactive ($n$=24, score 0–1) and active ($n$=11, score >1) disease groups. Metabolite extraction was performed by protein precipitation (200 µL plasma, 600 µL cold solvent), followed by incubation at −20 °C for 1 h and centrifugation at 16,000 g for 15 min at 4 °C. The dried supernatant was reconstituted in 0.1% formic acid in water/acetonitrile (1:1 v/v) and analysed by UHPLC-HRMS coupled to an Orbitrap Exploris 240. Acquisition was performed in full scan (60–900 m/z, resolution 120,000) with DDA-MS2 fragmentation (HCD 20/40/60) in both ESI+ and ESI− modes. Metabolites annotation used mzCloud and ChemSpider (putative identification level). Statistic analysis included PCA, Student's t-test with Benjamini-Hochberg FDR correction, and pathway enrichment analysis, and was performed using the software Compound Discoverer and Metaboanalyst 6.0.
**Results**
Global PCA showed no clear separation between the two disease groups. Dysregulated metabolites ($p$<0.05, Log2FC≥1) identified by Volcano plot analysis, were used for enrichment analysis. Main perturbations occurred in the amino acid metabolism and particularly in the glycine, serine and threonine pathway (raw $p$=0.00017) in accordance with literature [2]. ACN: MeOH extraction additionally revealed the involvement of the cysteine/methionine metabolism (raw $p$=0.026), alongside unsaturated fatty acid. However, due to the low number of samples evaluated no metabolites resulted significative for FDR correction.
**Conclusion**
This explorative study established a metabolomic protocol aimed at identifying candidate biomarkers of MTX response in a paediatric JIA cohort. Perturbation of glycine, serine and threonine metabolism is consistent with MTX interference in the folate cycle, where these amino acids serve as key substrates for nucleotide synthesis. These preliminary findings support precision medicine approaches in JIA, although a validation in larger cohorts is required.

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**Disclosure of interest**
None declared.

## PT109

### Distinct metabolic profiles in pediatric chronic nonbacterial osteomyelitis compared with healthy controls

B. Menentoğlu[1,*], Ö. Akgün[1], S. D. Arık[1], A. Dudaklı[1], P. Prencuva Akyürek[1], S. Tatonyan[2], F. G. Demirkan[3], A. Gedikbaşı[4], N. Aktay Ayaz[1]
[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine; [2]Istanbul University, Institute of Health Sciences; [3]Pediatric Rheumatology, Kanuni Sultan Süleyman Training and Research Hospital; [4]Division of Pediatric Nutrition and Metabolism, Istanbul University Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** B. Menentoğlu
*Pediatric Rheumatology 2026*, **24(S1):**PT109

**Introduction**
Chronic nonbacterial osteomyelitis (CNO) is a rare autoinflammatory bone disorder characterized by sterile bone inflammation with an incompletely understood pathogenesis. In addition to inflammatory pathways, metabolic dysregulation may contribute to disease development.
**Objectives**
This study aimed to identify potential metabolic biomarkers associated with pediatric CNO by comparing untargeted metabolomic profiles of patients with those of healthy controls.
**Methods**
Fifteen pediatric patients with CNO and 32 healthy controls were included. Serum and urine samples obtained from all participants underwent untargeted metabolomic profiling using liquid chromatography–quadrupole time-of-flight mass spectrometry (LC-QTOF-MS) in both negative ion mode (NEG) and positive ion mode (POS). Raw data underwent peak detection, alignment, and normalization prior to statistical analysis. Partial least squares discriminant analysis (PLS-DA) was applied to evaluate metabolic separation between groups, and significantly altered metabolites were identified using appropriate statistical tests. Group comparisons were assessed using $Log_2$ Fold Change ($Log_2FC$) values and p-values, with statistical significance defined as $p<0.05$. Pathway analysis was performed to evaluate the biochemical pathways associated with identified metabolites.
**Results**
In POS mode, N,N′-diphenylguanidine ($Log_2FC$: 5.40; $p=1.28\times10^{-9}$) and 2-amino-1,3,4-octadecanetriol ($Log_2FC$: 3.16; $p= 1.38\times10^{-9}$) levels were significantly higher in healthy controls, whereas cortisol, cortisone, inosine, and spermine levels were decreased in the CNO group. In NEG mode, thiomorpholine-3-carboxylate ($Log_2FC$: −0.83; $p=3.12\times10^{-9}$), pyroglutamylglycine ($Log_2FC$: −1.03; $p=6.93\times10^{-7}$), and N-oxalyl-L-alanine ($Log_2FC$: −1.3; $p=2.77\times10^{-3}$) levels were significantly increased in the CNO group. Increased levels of 2-(3-pentylphenyl) acetic acid and the aromatic hydrocarbon derivatives hexylbenzene and 1-(1-methylethenyl)-4-(1-methylethyl) benzene were also observed. In contrast, D-α-tocopherol and inosine levels were decreased in patients with CNO. Pathway analysis revealed significant alterations related to oxidative stress, amino acid and purine metabolism, sphingolipid balance, and steroid hormone metabolism.
**Conclusion**
These findings support the presence of systemic metabolic dysregulation in pediatric CNO and highlight the potential role of metabolomic profiling in improving disease characterization and biomarker identification.

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**Disclosure of interest**
None declared.

## PT110

### Multi-omics drug target prioritization in juvenile idiopathic arthritis integrating GWAS colocalization with immune-specific eQTL and plasma proteomics

O. Lomakina[1,*], M. Soloshenko[2,3]
[1]Morozov Children's City Clinical Hospita, Moscow; [2]ITMO University, M.Sc. Programme in Applied Genomics, St. Petersburg; [3]Alekseev Psychiatric Clinical Hospital No. 1, Moscow, Russian Federation
**Correspondence:** O. Lomakina
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**Introduction**
Juvenile idiopathic arthritis (JIA) is the most common chronic inflammatory arthropathy in childhood. While genome-wide association studies (GWAS) have identified >30 risk loci, translation of genetic findings into actionable drug targets remains limited.
**Objectives**
We developed and applied a systematic multi-evidence computational pipeline to prioritize druggable targets at JIA GWAS loci.
**Methods**
GWAS summary statistics (Hinks et al. 2021, $N\approx15{,}000$; GCST90010715, GRCh38) were used to define 27 genome-wide significant loci ($p<5\times10^{-8}$). For each locus, variants were extracted from four molecular QTL resources: GTEx v8 cis-eQTL (49 tissues), BLUEPRINT eQTL (5 primary immune cell types), DICE eQTL (15 immune cell types including Th1, Th17, monocytes), and deCODE plasma pQTL (4,907 proteins). Bayesian colocalization (coloc.abf v5.2.3) was performed across 1,305 GWAS×QTL tests. Candidate genes ($n$=455) were annotated with immune-cell specificity, Open Targets tractability scores, clinical drug pipeline data, and JIA association scores (EFO:0002609). A five-component composite score integrated all evidence layers.
**Results**
Colocalization evidence (PP.H4≥0.5) was identified for 89 genes across 24 loci. Top-ranked targets included: TNF (rank 1; PP.H4=0.80 with deCODE pQTL; 14 approved drugs), TYK2 (rank 2; lead variant $p=3\times10^{-12}$; 17 drugs in pipeline including approved TYK2 inhibitors), and IL6 (rank 6; PP.H4=0.998; strong immune-cell eQTL signal). Immune-specific colocalization identified Th1/Th17 eQTL signals at STAT4, STAT1, and IL12B loci. Among targets without a current JIA indication, TYK2 showed the strongest combined genetic support and druggability profile.
**Conclusion**
This integrative pipeline nominates TYK2 as the highest-priority novel therapeutic target in JIA based on convergent GWAS, eQTL, and druggability evidence. Systematic integration of immune-specific QTL resources substantially improves target resolution compared to positional mapping alone. The pipeline and results are openly available to support drug repurposing efforts in paediatric rheumatology.
**Disclosure of interest**
None declared.

## PT111

### Development of the ERN ReCONNET transition training framework

M. Zajc Avramovic[1,2,*], C. Simu[3], E. Marrani[4], S. Aguilera[5], L. Arnaud[6], C. Ciurtin[7], S. Kamphuis[8], E. Moraitis[7], K. Perdan Pirkmajer[9], M. Poelzl[10], F. Oliveira Ramos[11], P. Soler Palacin[12], A. Vissink[13], B. Vogrin[14], L. Zabuliene[15], G. Simonini[4], T. Avcin[1,2] on behalf of ERN ReCONNET Transition Task Force
[1]University Children's Hospital Ljubljana; [2]Medical faculty, University of Ljubljana, Ljubljana, Slovenia; [3]Hatieganu University of Medicine and Pharmacy, Cluj-Napoca, Romania; [4]Meyer Children's Hospital IRCCS, Florence, Italy; [5]EURORDIS, Lausanne, Switzerland; [6]National Reference Center for Rare Autoimmune Diseases (RESO), Academic Hospitals of Strasbourg, Strasbourg, France; [7]University College London, London, UK; [8]Erasmus MC Sophia Children's Hospital, Rotterdam, Netherlands; [9]University Medical Center Ljubljana, Ljubljana, Slovenia; [10]EULAR PARE, Graz, Austria; [11]University Hospital ULS Santa Maria, School of Medicine, University of Lisbon, Lisbon, Portugal; [12]Vall d'Hebron Barcelona Hospital Campus, Barcelona, Spain; [13]University of Groningen, Department of Oral and Maxillofacial Surgery, Groningen, Netherlands; [14]University of Maribor, Maribor, Slovenia; [15]Faculty of Medicine, Vilnius University, Vilnius, Lithuania
**Correspondence:** M. Zajc Avramovic
*Pediatric Rheumatology 2026*, **24(S1):**PT111

**Introduction**
Transition to adult healthcare is a vulnerable period for adolescents and young adults (AYA). ERN ReCONNET Transition Training Workshop was held in Ljubljana, Slovenia in November 2025 to define the basis for a standardized European curriculum.
**Objectives**
To develop a framework for standardized transition training curriculum in rCTDs.
**Methods**
The framework was developed through a structured consensus process. The ERN ReCONNET international expert panel included pediatric rheumatologists/immunologists (8), experts in pediatric and adult rheumatology (3), adult rheumatologists (2), patient representatives (2), a primary care pediatrician, a UEMS representative for adolescent medicine, and an oral health specialist. Prior to the workshop, example competencies from literature and prior ERN initiatives were prepared. During the workshop, participants were divided into expert groups to identify essential domains and core themes for transitional care training. Outputs were consolidated in a plenary session, followed by moderated discussion and voting. A consensus threshold of 80% agreement was required for inclusion. Post-workshop, competencies were aligned with agreed domains and themes and subjected to online voting using the same 80% consensus threshold. Competencies not meeting consensus were excluded.
**Results**
The Framework includes 7 domains: Principles of AYA medicine (1), Specifics of rCTDs in AYA (2), Transitional care (3), Communication (4), Health advocacy and patient partnership (5), Ethics and legal aspects (6) and Implementation and quality management (7). Each domain includes up to 5 core themes, altogether there are 25 core themes with alligned and measurable competencies under each core theme. Results are pesented in Table 1.
**Conclusion**
The final framework integrates 7 domains, core themes, and validated competencies, providing a structured basis for developing a European transition training curriculum.

**Table 1 (Abstract PT111)** Core themes and domains

| DOMAIN | CORE THEMES |
|---|---|
| Principles of AYA medicine | Developmental aspects of AYA<br>AYA with chronic conditions<br>Family dynamics<br>Mental health<br>Vulnerable and distincs subgroups |
| Specifics of rCTDs ni AYA | Trajectory of rCTDs<br>Reproductive health<br>Therapeutic adaptation and management in AYA<br>Preventive medicine and comorbiidities |
| Transitional care | Models of transitional care<br>Transitional careplanning and readiness<br>Multidisciplinary team collaboration<br>Transfer and continuity of care |
| Communication | Patient communication<br>Family communication<br>Inter. and intra- professional communication<br>e-health, digital technologies and telemedicine |
| Health advocacy and patient partnership | Health advocacy<br>Self-management, empowerment and patient partnership |
| Ethics and legal aspects | Ethical principles in AYA care<br>Legal aspects<br>Equity, inclusivity and cultural sensitiviity |
| Implementation and quality management | Implementation<br>Quality management<br>Continuus professional educaton |

**Disclosure of interest**
None declared.

## PT113

### Transition in paediatric rheumatology – impact of therapeutic patient education on access to adult care

I. Mahé[1,*], L. Rossi-Semerano[1], F. Anselmi[1], C. Galeotti[1], B. Fautrel[2], S. Georgin Lavialle [3], C. Hascoet[4], M. Nguelimo Barba[1], I. Kone-Paut[1], C. Marsaud[1], N. Noel[5], D. Saadoun[6], X. Mariette[7], R. Seror[7], P. Dusser[1]
[1]Paediatric Rheumatology Department, Assistance Publique-Hôpitaux de Paris (AP-HP), Hôpitaux universitaires Paris-Sud - Hôpital Bicêtre, F-94270, Le Kremlin Bicêtre; [2]Rheumatology, Assistance Publique-Hôpitaux de Paris (AP-HP), - Hôpital La Pitié Salpétrière; [3]nternal Medicine, Assistance Publique-Hôpitaux de Paris (AP-HP), - Hôpital Tenon; [4]Réseau rhumatismes inflammatoires pédiatriques (Resrip), Paris; [5]nternal Medicine, Assistance Publique-Hôpitaux de Paris (AP-HP), - Hôpital Bicetre, Le Kremlin Bicêtre; [6]nternal Medicine, Assistance Publique-Hôpitaux de Paris (AP-HP), - Hôpital La Pitié Salpetrière, Paris; [7]Rheumatology, Assistance Publique-Hôpitaux de Paris (AP-HP), - Hôpital Bicêtre, Le Kremlin Bicêtre, France
**Correspondence:** I. Mahé
*Pediatric Rheumatology 2026*, **24(S1):**PT113

**Introduction**
Transition to adult care is a critical step for adolescents with chronic rheumatic diseases and is associated with a substantial risk of loss to follow-up and disease flares. Timely attendance at a first adult consultation is commonly used as a proxy for successful transition, as failure to attend is an early marker of disengagement from care. At Bicêtre University Hospital, France, the transition program includes structured patient preparation, a joint paediatric–adult consultation, and personalised referral to adult care, with or without therapeutic patient education (TPE). TPE aims to strengthen patient autonomy and self-management skills.
**Objectives**
This study evaluated the impact of TPE on time to first adult consultation as a measure of transition success.
**Methods**
We conducted a single-centre retrospective cohort study including 167 patients who underwent a transition consultation between 2018 and 2024. The primary outcome was attendance at a first adult care consultation, analysed as time-to-event data using Kaplan-Meier estimates and Cox proportional hazards models. Univariate and multivariable analyses were performed to assess the association between TPE and time to first adult consultation, and to identify factors independently associated with earlier attendance. The main multivariable model was based on a directed acyclic graph (DAG) to minimise over-adjustment bias and included sex, RESRIP health network involvement, disease control at transition, disease category, disease duration, and socioprofessional status. Robust Poisson regression was performed as a sensitivity analysis using attendance within 12 months as a binary outcome, with adjustment for sex, disease category, and disease control at transition.
**Results**
Among the 167 patients (70% female, median age 18.2 years), 61 (36.5%) participated in TPE. Baseline characteristics were broadly similar between groups, although TPE patients were more often female, had shorter disease duration, and were less frequently diagnosed with autoinflammatory diseases. Overall, 135 patients (80.8%) attended adult care within one year, with a higher proportion in the TPE group (90.2% vs. 75.5%, $p = 0.03$). In the univariate Cox model, TPE showed a trend toward a shorter time to first adult consultation (HR 1.34; 95% CI 0.96–1.86), which did not reach statistical significance. After DAG-based adjustment, the estimate was slightly attenuated (adjusted HR 1.23; 95% CI 0.84–1.80). RESRIP participation (adjusted HR 1.46; 95% CI 1.02–2.09) and active disease at transition (adjusted HR 1.82; 95% CI 1.05–3.14) were independently associated with faster transition. Confidence intervals for the TPE effect crossed the null, possibly due to limited statistical power. The Poisson sensitivity analysis showed a statistically significant increase in attendance within one year, both univariate (RR 1.18; 95% CI 1.03–1.35) and multivariable (RR 1.16; 95% CI 1.02–1.32).

**Conclusion**
Participation in TPE showed a tendency toward improved transition success in paediatric rheumatology. Although the adjusted Cox model did not reach statistical significance, possibly due to limited statistical power, the effect estimates remained consistent in direction and magnitude across analyses, including the Poisson sensitivity analysis where statistical significance was reached.These findings support the integration of TPE into structured transition programs and warrant confirmation in larger prospective studies.
**Disclosure of interest**
None declared.

## PT114

### Does structured transition improve clinical outcomes after transfer to adult care? A real-world comparative cohort study

B. P. Correia[1,2], C. Zinterl[1,2], H. Cecílio[1,2], D. Russel[1,2], R. Campanilho-Marques[1,2], P. C. Reis[1,2], A. L. Martins[1,2], F. Oliveira-Ramos[1,2,*]
[1]Paediatric Rheumatology Unit, Rheumatology and Paediatrics Department, Unidade Local de Saúde de Santa Maria (ULSSM); [2]Centro Académico de Medicina de Lisboa (CAML), Faculdade de Medicina, Universidade de Lisboa, Lisbon, Portugal
**Correspondence:** F. Oliveira-Ramos
*Pediatric Rheumatology 2026*, **24(S1):**PT114

**Introduction**
Transition to adult care is a vulnerable phase for young people with paediatric-onset inflammatory rheumatic diseases, often associated with loss to follow-up and disease flares. Comparative real-world evidence on the impact of structured transition on post-transfer clinical outcomes remains scarce.
**Objectives**
To compare post-transfer clinical stability (remission at 6 months) and continuity of adult care (engagement and 12-month retention) between young people transferred through structured transition and those referred from other institutions without prior structured transition care.
**Methods**
Retrospective cohort study of young people with paediatric-onset inflammatory rheumatic diseases entering the Young Adult Rheumatology Clinic between 2022 and 2025. The structured transition group had ≥1 year of paediatric rheumatology follow-up at our centre before transfer; external transfers were referred from other institutions. Engagement was defined as ≥2 visits within 180 days after the first Young Adult Clinic visit (T0), and 12-month retention as ≥1 visit between days 181–365 with no follow-up gap >12 months. Multivariable logistic regression models were adjusted for diagnosis category, disease duration, baseline remission and treatment intensity at T0.
**Results**
A total of 153 patients were included (69.9% female): 118 in the structured transition (ST) group and 35 in the unstructured transfer (UT) group. Median age at disease onset was lower in the ST group compared with the UT group [11.0 (IQR 9.0) vs. 14.0 (IQR 6.0) years; $p$=0.037], and median age at T0 was 18.5 (IQR 1.0) vs. 19.0 (IQR 3.0) years, respectively ($p$=0.004). JIA was the most frequent diagnosis (47.1%), followed by jSLE (22.2%). ST patients were more often in remission at T0 (78.8% vs. 54.3%, $p$=0.004). Remission at 6 months (T1) was higher in the ST group (81.8% vs. 43.3%; $p$<0.001) and remained significant after multivariable adjustment (OR 6.32, 95%CI 2.11–18.92; $p$<0.001). Engagement was lower in the ST group on unadjusted analysis (32.8% vs. 54.3%; $p$=0.021) but did not remain significant after adjustment including baseline remission (OR 0.55, 95%CI 0.24–1.25; $p$=0.150). Retention was high overall (82.9%) and did not differ between groups after adjustment (OR 0.33, 95%CI 0.07–1.60; $p$=0.169).
**Conclusion**
Structured transition was associated with greater post-transfer clinical stability, with significantly higher remission at 6 months. Lower engagement in the structured transition group likely reflected greater baseline clinical stability and was not significant after multivariable adjustment. Retention was high overall, supporting the efficacy of the Young Adult Clinic model.
**Disclosure of interest**
None declared.

## PT115

### Real-life data of 407 patients juvenile idiopathic arthritis treated with golimumab: focus on drug survival

I. Nikishina[1,*], O. Kostareva[1], S. Arsenyeva[1], V. Matkava[1]
[1]Paediatric, V.A. Nasonova Scientific Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** I. Nikishina
*Pediatric Rheumatology 2026*, **24(S1):**PT115

**Introduction**
While Golimumab (GOL) has proven efficacy in clinical trials, real-world data across the full spectrum of JIA subtypes is limited.
**Objectives**
This study aimed to evaluate the long-term effectiveness, safety, and drug survival of GOL in a large, real-life cohort of children with various JIA categories.
**Methods**
A retrospective single-center study analyzed 407 JIA patients treated with subcutaneous GOL (2018–2025). We evaluated demographic data, JIA subtypes, treatment lines, ANA/HLA-B27 status, presence of uveitis, adverse events, and reasons for discontinuation. Drug survival was calculated using Kaplan-Meier analysis.
**Results**
The cohort included 407 patients treated with GOL (11% of all biologic prescriptions in our center). The female/male ratio was 1.6:1; mean age at disease onset was 8.5 years (from 3 month to 17 years); mean age at GOL initiation was 14.3±3.4 years; mean disease duration before GOL initiation was 5.8 years. RF-negative polyarticular subtype of JIA (310/407/76%), RF-positive polyarticular subtype of JIA (14/407/3.4%), juvenile ankylosing spondylitis (62/407/15%) and refractory variant of oligoarticular arthritis (19/407/5%). ANA positivity was present in 38% (155/407) of patients, HLA-B27 in 29% (120/407), uveitis in 23% (93/407), RF-positive in 8 pts, ACCP in 14 pts. 46% (186/407) of pts received GOL as first-line therapy, 2-nd line – 39% (159/407), 3-rd line and more – 16% (66/407). GOL was used as first-line therapy more often in juvenile ankylosing spondylitis (49/62/80%). In ANA-positive pts GOL was prescribed mainly in 2-nd and subsequent lines (108/155/70%). In patients with uveitis ($n$=93), GOL was mainly used as a second or subsequent biologic. The withdrawal rate in 1-st (31/186/16.6%) and 2-nd (27/157/17.1%) line was about 17%, but increased in 3-rd (12/51) and 4-th line (5/13) - 23% and 38%, respectively. The average duration of GOL therapy before withdrawal was 12.7 months (±10.5). The average duration GOL therapy in the second line - 11.8 months. 19% (76/407) patients discontinued therapy, predominantly because of inefficacy (59/76/77%), 14/19% due to non-medical reasons. Overall, GOL showed one of the lowest discontinuation rates among biologic agents used in our center. Six-year drug survival was 68%, with the highest retention in JIA-associated uveitis and juvenile ankylosing spondylitis (83% and 73%, respectively). The superior drug survival in uveitis cases suggests GOL strong potential in managing extra-articular manifestations even after previous TNF-alpha inhibitor failure. Adverse events were rare and included isolated rash and recurrent infections (2 withdrawal due to AE).
**Conclusion**
Golimumab demonstrates robust long-term effectiveness and a remarkable safety profile in a large real-world JIA cohort. While being a highly effective first-line option for HLA-B27-associated subtypes, GOL also serves as a potent "switch-to" biologic, particularly for patients with JIA-associated uveitis and those failing prior TNFi. The 6-year drug survival rate of 68% highlights its reliability in long-term disease control across various non-systemic JIA phenotypes.
**Disclosure of interest**
None declared.

## PT116

### Real-world use of anifrolumab in paediatric type I interferonopathies: the French experience

S. Plassart[1,*], A. Welfringer[2], P. Chen[2], B. Bader-Meunier[3], C. Ballot-Schmit[4], A. Belot[1], A. Carbasse[5], F. Chasset[6], S. De Almeida -Chaves[7], A. Delpuech[7], B. Fournier[3], S. Georgin-Lavialle[6], A. Hummel[2], E. Jeziorski[5], V. Le Guern[8], A. Mirguet[9], C. Pajot[7], C. Paul[7], H. Reumaux[10], C. Richez[11], J. Sirvente[5], A.-P. Foray[12], A. Nombel[12], G. Rice[13], L. Seabra[14], C. Bodemer[2], B. Neven[2], M.-L. Frémond[3,14]

[1]Pediatric Rheumatology, CRMR RAISE, Hospices Civils de Lyon, LYON; [2]Hôpital Necker-Enfants-Malades, APHP, Paris, France; [3]CRMR RAISE, Hôpital Necker-Enfants-Malades, APHP, Paris; [4]CHU de Besancon, Besancon; [5]CHU de Montpellier, Montpellier; [6]Hôpital Tenon, APHP, Paris; [7]CHU de Toulouse, Toulouse; [8]Hôpital Cochin, APHP, Paris; [9]CHRU Nancy, Nancy; [10]Hôpital Jeanne de Flandre, CHU Lille, Lille; [11]CHU de Bordeaux, Bordeaux; [12]Hospices Civils de Lyon, Lyon, France; [13]University of Manchester, Manchester, UK; [14]Laboratory of Neurogenetics and Neuroinflammation, Institut Imagine, Paris, France
**Correspondence:** S. Plassart
*Pediatric Rheumatology 2026*, **24(S1):**PT116

**Introduction**
Type I interferonopathies (IFNp-1) are a group of autoinflammatory disorders caused by genetic defects leading to excessive signalling of type I interferons (IFN-I). IFNp-1 are associated with a wide clinical spectrum encompassing neurological, cutaneous, articular, and pulmonary manifestations. In France, no authorised therapy is currently available for IFNp–1, and most patients are treated off-label with JAK inhibitors. However, their efficacy is variable with frequent adverse events. Anifrolumab, a mAb targeting the IFN-I receptor subunit IFNAR1, is approved for the treatment of systemic lupus erythematosus (SLE) in adults and offers a more targeted option. To date, clinical efficacy in IFNp-1 has been reported in 24 patients, mainly with SAVI, PRAAS and AGS (small case series and case reports). These studies have shown encouraging clinical efficacy, particularly for cutaneous manifestations, with consistent normalisation of IFN signatures. However, real–world data from larger cohorts are lacking.
**Objectives**
The aim of this study was to describe the real–world experience with anifrolumab in a multicentre French cohort of patients with IFNp–1.
**Methods**
We conducted a retrospective multicentre observational French cohort of 26 genetically confirmed IFNp–1 patients treated with anifrolumab, assessing safety, efficacy and IFN–I scores.
**Results**
The cohort included patients with SAVI (*n*=6), AGS (*n*=5), COPA (*n*=1), PRAAS (*n*=1), SPENCD (*n*=1), monogenic SLE (C1q deficiency, TREX1, UNC93B1; candidate gene *n*=5), DNASE2 deficiency (*n*=1), and other diagnoses (*n*=6). Overall, 65% of patients were female (*n*=17) and 65% were paediatric (*n*=17), with a median age of 15.2 years [1.7–58.6] at anifrolumab initiation. Anifrolumab was administered intravenously at doses up to 300 mg every 3 to 4 weeks, for a median duration of 9 [3–54] months. Concomitant treatments included JAK inhibitors (62%), corticosteroids (38%), and other immunosuppressants (31%). Marked clinical improvement, including complete remission, was observed in cutaneous manifestations (13/15 patients), whereas effects on articular and pulmonary involvement were more limited. IFN score normalisation occurred in 83% of patients (15/18), within three months of treatment, with 10 patients achieving normalisation after only one or two anifrolumab infusions. Finally, three patients experienced infectious adverse events; two died from severe aspergillosis that predated anifrolumab initiation and was considered unrelated to treatment.
**Conclusion**
This national French cohort represents the largest real-world experience reported to date with anifrolumab in IFNp-1. IFNAR1 blockade appears effective for cutaneous manifestations, particularly after JAK inhibitor failure or intolerance, supporting prospective evaluation and a potential first-line therapy.

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**Disclosure of interest**
None declared.

## PT117

**Long-term safety and efficacy of Tofacitinib in the treatment of patients with juvenile idiopathic arthritis: final results of an open-label extension study**

N. Ruperto[1,*], S. Sule[2], G. Horneff[3], C. Wouters[4], I. Foeldvari[5], J. Akikusa[6], A. L. Boteanu[7], I. Vranic[8], C. Chang[9], S. Liu[10], A. Yndestad[11], A. Diehl[12], K.-C. Tabner[8], C. White[8], D. J. Lovell[13], A. Martini[14], H. I. Brunner[13] on behalf of for the Paediatric Rheumatology International Trials Organisation (PRINTO) and the Pediatric Rheumatology Collaborative Study Group (PRCSG)
[1]Dipartimento di Medicina e Chirurgia, Università Milano Bicocca, Monza, Italy and Fondazione IRCCS San Gerardo dei Tintori, Reumatologia Pediatrica, PRINTO, Monza, Italy; [2]Pediatric Rheumatology, Children's National Medical Center, Washington, DC, USA; [3]Department of Paediatric and Adolescents Medicine, University Hospital of Cologne, Cologne, Germany; [4]Department of Pediatrics, University Hospitals Leuven, Leuven, Belgium; [5]Hamburger Zentrum für Kinder- und Jugendrheumatologie, Hamburg, Germany; [6]Royal Children's Hospital, Melbourne, Australia; [7]Pediatric Rheumatology Unit, University Hospital Ramón y Cajal, Madrid, Spain; [8]Pfizer Ltd, Tadworth, UK; [9]Pfizer Inc, Groton, CT, USA; [10]Pfizer Inc, Shanghai, China; [11]Pfizer Inc, Oslo, Norway; [12]Pfizer Inc, Collegeville, PA; [13]Division of Rheumatology and Department of Pediatrics, Cincinnati Children's Hospital Medical Center and University of Cincinnati College of Medicine, Cincinnati, Cincinnati, USA; [14]University of Genova, Genova, Italy
**Correspondence:** N. Ruperto
*Pediatric Rheumatology 2026*, **24(S1):**PT117

**Introduction**
Tofacitinib (TOFA) is an oral Janus kinase (JAK) inhibitor for the treatment of JIA.
**Objectives**
Evaluate long-term safety, tolerability, and efficacy of TOFA in patients (pts) with JIA.
**Methods**
This was an open-label, long-term extension (LTE) study (NCT01500551) of tofacitinib with up to 118 months of observation. Pts with polyarticular course (pc)JIA, systemic (s)JIA, juvenile psoriatic arthritis (jPsA), or enthesitis-related arthritis (ERA) who completed, or discontinued for reasons other than treatment-related serious adverse events (AEs), index studies in the tofacitinib JIA program (Phase 1, NCT01513902; Phase 3, NCT02592434 or NCT03000439) were eligible for study enrollment. Pts received open-label TOFA 5 mg twice daily or an equivalent weight-based lower dose. Safety endpoints and laboratory test abnormalities are reported for the overall cohort. Efficacy endpoints are reported as observed for cohorts of pts with polyarticular-course (pc)JIA and systemic (s)JIA.
**Results**
302 pts with pcJIA (*n*=185), sJIA (*n*=77), jPsA (*n*=19), or ERA (*n*=21) were enrolled. This represents 86% (302/351) of pts in the index studies (26/26 from NCT01513902, 199/225 from NCT02592434, and 77/100 from NCT03000439). The 185 pts with pcJIA were classified as extended oligoarthritis (EO; *n*=27), rheumatoid factor (RF)-positive polyarthritis (*n*=36), RF-negative polyarthritis (*n*=111), and sJIA without active systemic features (*n*=11). Median (range) TOFA exposure was 33.7 (0–118) months. There were 268 (89%) pts with AEs and 48 (16%) pts with >1 serious AE. Most common AEs (>10% pts) were upper respiratory tract infection, nasopharyngitis, and JIA exacerbation. 127 pts (42%) discontinued the study; the reason was insufficient clinical response in 35 pts (12%), and 39 pts (13%) discontinued due to AE. One death occurred 15 days after end of study due to an AE of haemophagocytic lymphohistiocytosis (macrophage activation syndrome; MAS) associated with sJIA flare. The following AE of special interest events were reported: Serious infections occurred in 14 (4.6%) pts. The most frequent serious infections were herpes zoster (HZ) and pneumonia. HZ events were reported in 6 patients (2.0%), 2 were nonserious and 4 were serious of which 2 (0.7%) pts experienced serious multidermatomal HZ (adjudicated as OI). A single patient with nonserious HZ, had an occurrence of a prior nonserious varicella event. Uveitis occurred in 3 (1.0%) pts and MAS occurred in 2 (0.7%) pts. There were no events of MACE, malignan-

cy or venous thromboembolism. In pts with pcJIA or sJIA, observed median JADAS-27 CRP was low (i.e., ≤3.8) after month (M)1, median CHAQ-Disability Index indicated mild disability (<0.13) after M3, and median CHAQ-Discomfort index (pain VAS) was ≤1.0 from M1. These improvements were generally maintained throughout the study. JIA ACR ID response rates increased from M1; in the pcJIA cohort, rates were >40% between M27 and M78 and in the sJIA cohort, rates remained stable between 16 and 50% from M3-M36. Clinical Remission was achieved at least once by 41% of pts with pcJIA and 21% of pts with sJIA.

**Conclusion**

In this open-label, LTE study of TOFA in pts with JIA, there were no new safety signals specific to the JIA population or new to the TOFA safety profile. Clinical efficacy was maintained during long-term treatment with TOFA.

**Trial registration identifying number**

NCT01500551.

**Disclosure of interest**

N. Ruperto Consultant with: AlfaSigma, Amgen, Boehringer Ingelheim, Bristol Myers Squibb, Eli Lilly, Galapagos, Genentech Roche, Idorsia, Janssen, Pfizer, and Takeda, Speaker Bureau with: AlfaSigma, Amgen, Boehringer Ingelheim, Bristol Myers Squibb, Eli Lilly, Galapagos, Genentech Roche, Idorsia, Janssen, Pfizer, and Takeda, S. Sule: None declared, G. Horneff Grant / Research Support with: Janssen, Novartis and Pfizer, Speaker Bureau with: AbbVie, GSK, Novartis, Pfizer and Sobi, C. Wouters Speaker Bureau with: Novartis and SOBI, I. Foeldvari Grant / Research Support with: Joachim Herz Stiftung, Consultant with: Boehringer Ingelheim, Eli Lilly, Pfizer, Novartis, and Medac, J. Akikusa: None declared, A. Boteanu: None declared, I. Vranic Shareholder with: Pfizer Inc, Employee with: Pfizer Inc, C. Chang Shareholder with: Pfizer Inc, Employee with: Pfizer Inc, S. Liu Shareholder with: Pfizer Inc, Employee with: Pfizer Inc, A. Yndestad Shareholder with: Pfizer Inc, Employee with: Pfizer Inc, A. Diehl Shareholder with: Pfizer Inc, Employee with: Pfizer Inc, K.-C. Tabner Shareholder with: Pfizer Inc, Employee with: Pfizer Inc, C. White Shareholder with: Pfizer Inc, Employee with: Pfizer Inc, D. Lovell Grant / Research Support with: Bristol Myers Squibb, Janssen, and Roche, Consultant with: AstraZeneca, GSK, Novartis, Pfizer Inc, and UCB, A. Martini Consultant with: Janssen and Pfizer Inc, H. Brunner Grant / Research Support with: Bristol Myers Squibb, MedImmune, Novartis, and Pfizer Inc, Consultant with: AbbVie, AstraZeneca/MedImmune, Bayer, Biocon, Bristol Myers Squibb, Boehringer Ingelheim, Eli Lilly, Janssen, Novartis, Pfizer Inc, Roche, and R-Pharm, Speaker Bureau with: GSK, Novartis, and Roche.

## PT118

### Anti-canakinumab antibodies as a potential cause of secondary treatment failure: rationale for therapeutic drug monitoring

S. M. Orsi[1,2,*], A. Cafaro[3], F. Schena[2], M. Biondi[3], G. Cangemi[3], M. Gattorno[2], R. Caorsi[2]
[1]Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genoa; [2]UOC Reumatologia e Malattie Autoinfiammatorie; [3]Biochemistry, Pharmacology and Newborn Screening Unit, Central Laboratory of Analysis, IRCCS Istituto Giannina Gaslini, Genoa, Italy
**Correspondence:** S. M. Orsi
*Pediatric Rheumatology 2026*, **24(S1):**PT118

**Introduction**

Canakinumab, a monoclonal antibody targeting interleukin-1β, is widely used in autoinflammatory diseases. However, secondary treatment failure occurs in a subset of patients, potentially due to anti-drug antibodies (ADAs). While immunogenicity is well established for other biologics, its relevance for canakinumab remains poorly defined.

**Objectives**

This study aimed to evaluate the relationship between canakinumab plasma levels, ADAs and clinical response, and to explore the role of therapeutic drug monitoring (TDM).

**Methods**

We analyzed 43 plasma samples from 29 patients with autoinflammatory diseases. Canakinumab concentrations were measured using quantitative ELISA (SHIKARI® Q-CAN), and ADAs by qualitative ELISA (SHIKARI® S-ATCAN). Descriptive statistics were applied, and the correlation between drug levels and time since last administration was assessed using Spearman's coefficient.

**Results**

The study cohort included patients with the following diagnoses: cryopyrin-associated periodic syndrome (CAPS, *n*=9), tumor necrosis factor receptor-associated periodic syndrome (TRAPS, *n*=5), mevalonate kinase deficiency (MKD/HIDS, *n*=5), familial Mediterranean fever (FMF, *n*=4), systemic juvenile idiopathic arthritis (SJIA, *n*=2), PSTPIP1-associated autoinflammatory diseases (*n*=2), Majeed syndrome (*n*=1), and systemic undefined recurrent fever (SURF, *n*=1). Median age was 15 years (IQR 9.5), with balanced sex distribution. Median canakinumab concentration was 172.3 mg/L (IQR 180.35). A strong inverse correlation was observed between drug levels and time since last administration (rho = −0.823, $P$<0.0001). ADAs were detected in 3/43 samples (7.0%), corresponding to 2/29 patients (6.9%). Both ADA-positive patients experienced loss of response. Notably, both patients had genetically defined diseases (CINCA and TRAPS) and had been receiving canakinumab for 5 and 10 years, respectively, with initial good response to treatment. In contrast, most ADA-negative patients maintained detectable drug concentrations and clinical response, although two cases of loss of response (a CINCA and a SURF patient) occurred despite absence of ADAs, suggesting alternative mechanisms such as pharmacodynamic failure. Furthermore, both patients with PSTPIP1-associated autoinflammatory diseases showed persistent residual disease activity despite negative ADA testing.

**Conclusion**

These findings suggest that anti-canakinumab antibodies may contribute to secondary treatment failure, potentially through enhanced drug clearance. TDM, including measurement of both drug levels and ADAs, may represent a valuable tool to distinguish pharmacokinetic from pharmacodynamic failure and guide clinical decision-making. Larger prospective studies are needed to confirm these observations and define optimal therapeutic targets for canakinumab.

**Disclosure of interest**

None declared.

## PT119

### LincRNA-p21 in juvenile idiopathic arthritis: a novel mediator and biomarker of methotrexate response

F. Riccardi[1], S. Sindici Forgiarini[1], L. Messineo[2], F. Corba[2], B. Bellich[1], P. dalla Zuanna[1], L. Collavin[2], S. Pastore[1], A. Taddio[1,2,*], G. Stocco[1,2], M. Lucafò[2]
[1]Institute for Maternal and Child Health I.R.C.C.S. Burlo Garofolo; [2]University of Trieste, Trieste, Italy
**Correspondence:** A. Taddio
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**Introduction**

Methotrexate (MTX), the first-line treatment for juvenile idiopathic arthritis (JIA), may exert anti-inflammatory effects through lincRNA-p21–mediated modulation of the p53/NF-κB pathway, although its role in fibroblasts and as a biomarker of treatment response remains unclear.

**Objectives**

To dissect the molecular function of lincRNA in MTX-mediated anti-inflammatory mechanisms and to evaluate its potential prognostic role in JIA patient stratification and therapy monitoring.

**Methods**

SW982 cells (fibroblast-like synoviocytes, FLS) were treated with 500 nM MTX in the presence or absence of an inflammatory stimulus (10 ng/mL TNF-α and 10 ng/mL IL-1β). Expression levels of IL-6, IL-8, and lincRNA-p21 were evaluated by real-time PCR and protein levels of H2A.X, DNA-PK, ATM, P53, P65, and PKA were assessed by Western blot. Adenosine and uric acid levels were measured by LC-MS/MS. Circulating RNA was isolated from plasma of JIA patients enrolled in a prospective study at IRCCS Burlo Garofolo in Trieste. Data were analyzed by ANOVA with Sidak's multiple comparison tests (in vitro) and Spearman correlation (in patients).

**Results**

Following treatment with MTX for 72 h, the stimulus increased IL-6 and IL-8 levels, whereas co-treatment with MTX resulted in a lower level of IL-6 ($p$<0.001), but not IL-8. MTX increased P53 abundance and lincRNA-p21 regardless of stimulus, while P65 protein level was reduced in co-treated cells (all $p$<0.05). Investigating the mechanism by which MTX induces lincRNA-p21, we found that MTX treatment increases H2A.X and its phosphorylated form (γH2A.X) ($p$<0.001), a marker of double-strand DNA damage. This likely affects apical kinases DNA-PK and ATM,

which may, in turn, promote P53 activation and subsequent lincRNA-p21 induction, attenuating P65 levels and thereby inhibiting NF-kB target genes. To determine whether adenosine contributes to the anti-inflammatory response, we measured PKA protein and adenosine metabolites released in FLS supernatants. Though PKA levels did not change, LC-MS/MS analysis showed that both adenosine and uric acid levels were higher in co-treated cells, supporting adenosine role in the MTX-mediated response in FLS. To assess the translational relevance of these data, we measured lincRNA-p21 in plasma from an exploratory cohort of 12 JIA patients receiving MTX and correlated its level with disease activity indices JADAS27, erythrocyte sedimentation rate, and C-reactive protein. Notably, we found a significant correlation between lincRNA-p21 and both ESR ($p$=0.03) and CRP ($p$=0.048).

**Conclusion**

These findings support a role for lincRNA-p21 in MTX-mediated anti-inflammatory effects and suggest its potential as a non-invasive biomarker of disease activity and treatment response in JIA.

**Disclosure of interest**

None declared.

## PT120

### Anti-adalimumab antibodies in childhood rheumatic diseases and their predictors: findings from a real-world Asian pediatric cohort

K. L. Teh[1,2,*], J. C. Huang[1,2], L. Das[1,2], Y. X. Book[1], X. Gao[3], X. Guo[3], T. Arkachaisri[1,2]

[1]Rheumatology and Immunology, KK Women's and Children's Hospital; [2]Paediatric Academic Clinical Programme, Duke-NUS Medical School; [3]Division of Nursing, KK Women's and Children's Hospital, Singapore, Singapore

**Correspondence:** K. L. Teh

*Pediatric Rheumatology 2026*, **24(S1):**PT120

**Introduction**

Adalimumab is widely used to treat childhood-onset rheumatic diseases (cRD). However, the development of anti-adalimumab antibodies (AAA) may reduce drug levels, thereby compromising therapeutic efficacy. Identifying clinical predictors of AAA could help inform treatment strategies and monitoring approaches. Most immunogenicity data are from Western adult cohorts, with limited data in cRD.

**Objectives**

We aimed to describe the occurrence of AAA and identify factors associated with AAA development in a multiethnic Asian pediatric rheumatology cohort receiving adalimumab.

**Methods**

Prospective longitudinal data from KK Women's and Children's Hospital, Singapore, were included. Patients with cRD were recruited if they were receiving subcutaneous adalimumab and had undergone at least one AAA assessment with at least 6 months of follow-up. AAAs were measured using a validated immunoassay as part of routine therapeutic drug monitoring. The primary outcome was AAA formation. Nonparametric analyses were used to describe data. Logistic regression analyses were performed to evaluate potential predictors of AAA development. Time-to-event analyses were conducted using Cox regression.

**Results**

A total of 140 patients with childhood rheumatic diseases were included (75% JIA, 11% uveitis, 6% chronic non-bacterial osteomyelitis, 8% other diagnoses). During follow-up, 59 patients (42.1%) developed AAA during the median follow-up duration of 19.8 months (IQR 11.5 -27.1 months). The median time from adalimumab initiation to first detection of AAA was 18.3 months (IQR 9.5–31.0). Of patients who developed AAA, 32.2% did so within the first 12 months. Cumulative incidence rates rose constantly with ongoing treatment. In patients without prior adalimumab exposure ($n$=99), AAA developed in 39.4%. Baseline demographic characteristics, including age, gender, and race, were not associated with AAA development. Underlying cRD diagnosis was also not associated with antibody formation. Prior TNFi exposure, concomitant methotrexate use, and corticosteroid therapy were not significantly associated with AAA development on multivariate analysis. Similarly, adalimumab dose tapering did not demonstrate a clear association with antibody formation. In time-to-event analyses, methotrexate use was not associated with prolonged AAA-free survival. Subgroup analysis of patients who were adalimumab naïve showed similar results. Patients with detectable AAA had lower median adalimumab trough levels than those without antibodies (4.2 ug/ml, IQR 0 – 11.6 ug/ml vs. 8.3 ug/ml, IQR 7.7 – 16.2 ug/ml, $p$ = 0.002). More patients stopped medication due to inefficacy (35.5% vs. 16.1%), but this difference was not statistically significant.

**Conclusion**

In this pediatric rheumatology cohort treated with adalimumab, AAA developed in about 40% of patients during follow-up. Demographic, disease-related and treatment-related variables were not predictors of anti-adalimumab antibody development. Contrary to previous reports, concomitant MTX did not confer protection against antibody development. Larger multicentre and multiethnic studies are needed to confirm our initial findings.

**Disclosure of interest**

None declared.

## P001

### Etanercept vs. adalimumab in JIA: real-world propensity score-matched comparison

A. Marino[1,*], M. Pandolfi[1], M. Arese[1], D. Pireddu[1], C. Riboldi[1], G. Lommano[1], F. Baldo[1], S. Costi[1], R. Caporali[1], C. B. Chighizola[1]

[1]Pediatric Rheumatology Unit, ASST G.Pini-CTO, Milan, Italy

**Correspondence:** A. Marino

*Pediatric Rheumatology 2026*, **24(S1):**P001

**Introduction**

Comparative data on the effectiveness of etancercept (ETN) and adalimumab (ADA) for juvenile idiopathic arthritis (JIA) treatment are lacking.

**Objectives**

To compare the effectiveness of ETN versus ADA for JIA in real life at long term.

**Methods**

Patients with JIA treated with ETN or ADA as the first biologic agent were retrospectively included over a 25-year study period. Patients with uveitis were excluded. Propensity score (PS) matching was performed using nearest-neighbor matching (1:1 ratio) based on sex, age at diagnosis, and JIA subtype. Cox proportional hazards regression was used to analyze risk factors for arthritis flare.

**Results**

The cohort included 155 JIA patients (106 females): 82 patients received ETN and 73 were treated with ADA. The median follow-up was 16.25 years. Baseline features of the cohort are shown in Table 1. There were no significant differences for age at diagnosis, sex, number of active joints at baseline, concomitant methotrexate (MTX) at ADA/ETN initiation, or time from diagnosis to ADA/ETN initiation. The median treatment retention was comparable between the two groups [2.9 years for ADA, interquartile range (IQR) 1.4-5.8, vs. 4.3 years for ETN (IQR 1.8-9.1); $p$ = 0.3] as well as treatment withdrawal due to ineffectiveness (55% for ADA vs.

**Table 1 (Abstract P001)** Baseline features of the cohort

| Characteristic | Overall $N$ = 155[1] | ADA $N$ = 73[1] | ETN $N$ = 82[1] | $p$-value 2 |
|---|---|---|---|---|
| Age at diagnosis, median (IQR) | 6.8 (2.4 - 13.0) | 5.2 (2.3 - 14.0) | 7.1 (3.2 - 11.0) | 0.8 |
| JIA subtype, n (%) | | | | <0.001 |
| Enthesitis-related arthritis | 22 (14%) | 19 (26%) | 3 (3.7%) | |
| Oligoarthritis | 82 (53%) | 41 (56%) | 41 (50%) | |
| Polyarthritis RF- | 27 (17%) | 5 (6.8%) | 22 (27%) | |
| Polyarthritis RF+ | 11 (7.1%) | 1 (1.4%) | 10 (12%) | |
| Other* | 13 (8.4%) | 7 (9.6%) | 6 (7.3%) | |
| Number of active joints at baseline, median (IQR) | 2.00 (1.00 - 4.00) | 2.00 (1.00 - 3.00) | 2.00 (1.00 - 6.00) | 0.09 |
| Concomitant Methotrexate, n (%) | 70 (53%) | 39 (62%) | 31 (46%) | 0.06 |

[1]n (%); [2]Wilcoxon rank sum test; Pearson's Chi-squared test; Fisher's exact test; IQR: interquartile range; * psoriatic arthritis $n$ =7, systemic arthritis = 4, Undifferentiated = 2

53% for ETN; $p = 0.9$). After PS matching, 41 patients treated with ADA were matched to 41 treated with ETN. In the propensity score-matched cohort, Cox regression did not demonstrate an increased risk of arthritis flare wit ADA compared with ETN neither in the univariate analysis [Hazard Ratio (HR) 0.83; 95% Confidence Intervals (CI) 0.44-1.6; $p = 0.6$] nor after adjustment for concomitant MTX use and time from diagnosis to ADA/ETN initiation (HR 0.79; 95% CI 0.4- 1.56; $p = 0.5$).

**Conclusion**

In this real-world cohort of patients with JIA without uveitis, ETN and ADA showed comparable long-term effectiveness, treatment retention, and flare risk.

**Disclosure of interest**

None declared.

## P002

### Correlation of serum calprotectin with disease activity and laboratory markers in non-systemic juvenile idiopathic arthritis — a monocentric cohort

A. Pinna[1], F. Diomeda[1], F. Castellana[2], G. Lobreglio[3], G. Pellegrino[3], F. Indino[3], C. Malattia[4,5], A. Civino[1,*]
[1]Pediatric Rheumatology and Immunology Unit, 'Vito Fazzi' Hospital, Lecce; [2]Department of Interdisciplinary Medicine (DIM), University of Bari Aldo Moro, Bari; [3]Clinical Pathology Unit, 'Vito Fazzi' Hospital, Lecce; [4]Rheumatology and Autoinflammatory Diseases Unit, IRCCS Istituto Giannina Gaslini; [5]Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genoa, Genoa, Italy
**Correspondence:** A. Civino
*Pediatric Rheumatology 2026*, **24(S1):**P002

**Introduction**

Serum calprotectin (S100A8/A9-sCal) is a promising biomarker for monitoring disease activity and flare prediction in juvenile idiopathic arthritis (JIA). Conventional biomarkers, such as C-reactive protein (CRP) and erythrocyte sedimentation rate (ESR), may remain normal despite joint inflammation.

**Objectives**

To evaluate the ability of sCal to discriminate active from inactive JIA and healthy controls, and to compare its performance with conventional inflammatory biomarkers.

**Methods**

Cross-sectional, single center study including patients fulfilling ILAR criteria for JIA, enrolled consecutively over 6 months. Inactive disease was defined according to Wallace criteria. Healthy controls were included. Assessments included complete blood count, ESR, high-sensitivity CRP (hsCRP), and sCal (Bühlmann sCAL turbo kit). Statistical analysis included Kruskal–Wallis, Dunn's post-hoc tests, logistic regression adjusted for sex, hsCRP, and ESR, and ANOVA for model comparison. Predictive accuracy was assessed using a confusion matrix.

**Results**

The study included 61 subjects: 24 active JIA, 16 inactive JIA, and 21 healthy controls. The JIA cohort includes 16 Oligoarticular, 17 Polyarticular, 5 Enthesitis-related arthritis, and 1 Psoriatic arthritis. Significant differences were observed for sCal, hsCRP, and ESR (Table 1). Post-hoc analysis confirmed that sCal levels in active JIA were significantly higher than in inactive JIA and in controls ($p<0.01$). Logistic regression showed that sCal was associated with active disease (OR 2.8), remaining significant after adjustment for sex, hsCRP, and ESR (OR 2.47, 95% CI: 1.04–5.88). ANOVA analysis confirmed that adding sCal to standard markers significantly improved model deviance ($p=0.01$). The predictive model achieved an overall accuracy of 72.5% (95%CI: 56.1%–85.4%).

**Conclusion**

Integrating sCal into standard biomarker panels may improve disease assessment in JIA. Elevated sCal levels, even with normal ESR and CRP values, may indicate subclinical inflammation and increased flare risk, supporting its role as a useful biomarker for disease monitoring and therapeutic decision-making.

**Disclosure of interest**

None declared.

**Table 1 (Abstract P002)** Laboratory parameters according to disease activity

| | Overall (*n*=61) | Active JIA (*n*=24) | Inactive JIA (*n*=16) | Controls (*n*=21) | *p* |
|---|---|---|---|---|---|
| Age (Y) | 13.00 [8.00,16.25] | 12.00 [7.50,17.50] | 14.50 [10.00,15.50] | 13.00 [8.00,15.00] | 0.84 |
| Male | 25 (41.0) | 9 (37.5) | 7 (43.8) | 9 (42.9) | 0.90 |
| sCal µg/ml | 1.65 [0.96,2.62] | 2.51 [1.69,3.75] | 1.32 [0.79,1.94] | 1.09 [0.90,1.91] | <0.01 |
| hsCRP mg/L | 0.80 [0.30,3.30] | 1.45 [0.65,5.10] | 0.30 [0.20,0.65] | 0.80 [0.50,3.30] | <0.01 |
| WBC$10^3$/µl | 7.75 [6.10,8.88] | 8.11 [6.90,8.98] | 7.82 [6.64, 8.46] | 6.34 [5.64,7.94] | 0.04 |
| Neutrophil $10^3$/µl | 3.52 [2.63,4.80] | 4.24 [3.40,5.68] | 3.67 [2.45, 4.43] | 2.70 [2.45,3.59] | <0.01 |
| ESR mm/h | 4.00 [2.00, 8.00] | 9.00 [3.75, 19.00] | 2.50 [2.00, 4.00] | 2.00 [2.00, 4.00] | <0.01 |

Data are shown as numbers (%), and median [IQR]

## P003

### Serum S100A8/a9 as a potential biomarker of disease activity in juvenile idiopathic arthritis-associated uveitis: a first pilot single-center study from Kazakhstan

A. Tugelbayeva[1,*]
[1]Pediatric Department, Semey Medical University, Semey, Kazakhstan
**Correspondence:** A. Tugelbayeva
*Pediatric Rheumatology 2026*, **24(S1):**P003

**Introduction**

JIA-associated uveitis is a severe complication and a leading cause of vision loss in children. Conventional markers (ESR, CRP) often fail to reflect intraocular inflammation. S100A8/A9 is an emerging biomarker, but data in Asian populations are limited [1,2].

**Objectives**

To evaluate serum S100A8/A9 levels in children with JIA-associated uveitis in a Kazakhstani cohort and to assess its association with ocular inflammation activity.

**Methods**

This pilot cross-sectional study included children diagnosed with JIA according to ILAR criteria. Patients were divided into two groups: JIA with active uveitis (n=15) and JIA without uveitis (n=15). Serum S100A8/A9 levels were measured using ELISA. In addition, tear fluid samples were collected from all participants using standardized Schirmer strips and have been stored at -80 °C for subsequent analysis. Along with standard clinical predictors of uveitis development and progression (age at onset, JIA subtype, ANA status, disease duration, ESR, CRP), ophthalmologic activity was assessed by anterior chamber cell grading according to the Standardization of Uveitis Nomenclature (SUN) classification. Statistical analysis included the Mann-Whitney U test, Spearman correlation, and ROC curve analysis.

**Results**

A total of 30 patients were included (mean age 10.8 ± 3.2 years; 63% female). Median serum S100A8/A9 levels were significantly higher in the active uveitis group compared to JIA patients without uveitis (2.8 [IQR 2.1–3.6] vs. 1.4 [IQR 0.9–2.0] µg/mL, $p = 0.003$). Serum S100A8/A9 showed a moderate positive correlation with anterior chamber cell grade (Spearman $\rho = 0.62$, $p = 0.001$). ROC analysis demonstrated acceptable discriminatory ability for active uveitis (AUC = 0.81). At a cut-off of 2.05 µg/mL, sensitivity was 80% and specificity 73%.

**Conclusion**

In this first pilot study from Central Asia, serum S100A8/A9 levels were elevated in active JIA-uveitis and correlated with inflammation severity. Tear fluid samples were collected for future studies. S100A8/A9 may be a promising non-invasive biomarker. Larger multicenter studies are needed to validate its utility in Asian populations.

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Walscheid K, Heiligenhaus A, Holzinger D, Roth J, Heinz C, Tappeiner C, et al. Elevated S100A8/A9 and S100A12 Serum Levels Reflect Intraocular Inflammation in Juvenile Idiopathic Arthritis-Associated Uveitis: Results from a Pilot Study. Invest Ophthalmol Vis Sci. 2015;56(13):7653–60.
**Trial registration identifying number**. Not applicable — this is an observational study, not a clinical trial.
**Disclosure of interest**
None declared.

## P004

### Predictors of inactive disease after first biologic initiation in juvenile idiopathic arthritis: the role of baseline disease burden

T. Çeltik[1,*], R. Torun[2], T. Aydın[3], Z. Kızıldağ[4], R. İşgüder[5], E. Ünsal[2], B. Bora[2]
[1]Dokuz Eylül University; [2]Dokuz Eylül University Faculty of Medicine, Izmir; [3]Erzurum City Hospital, Erzurum; [4]Trakya University Faculty of Medicine, Edirne; [5]Adana City Hospital, Adana, Türkiye
**Correspondence:** T. Çeltik
*Pediatric Rheumatology 2026*, **24(S1):**P004

**Introduction**
Biologic therapies have improved outcomes in juvenile idiopathic arthritis (JIA); however, inactive disease is not achieved uniformly.
**Objectives**
To identify predictors of inactive disease within 12 months of initiating the first biologic agent and evaluate time to the first inactive-disease episode.
**Methods**
This retrospective cohort included biologic-treated JIA patients aged 0–18 years followed between 2005 and 2021. Inactive disease was defined according to Wallace criteria. Patients already inactive before biologic initiation were excluded from post-biologic analyses. The primary outcome was inactive disease within 12 months of initiating a biologic. Multivariable logistic regression assessed early biologic initiation (<6 months from diagnosis), sex, age at diagnosis, diagnostic delay, overweight/obesity, enthesitis-related arthritis, and an exploratory baseline disease-burden index. This index included routinely available baseline features known to influence disease burden: joint-site count, hip involvement, uveitis, and enthesitis. Kaplan–Meier, Cox regression, and inverse probability of treatment weighting (IPTW) analyses were also performed.
**Results**
Of 225 screened biologic-treated records, 160 patients were eligible. The most common JIA subtypes were RF-negative polyarticular JIA (30.6%) and enthesitis-related arthritis (28.7%). Overall, 66/126 patients (52.4%) achieved inactive disease within 12 months. Key findings are summarized in Table 1. Higher baseline disease burden was independently associated with lower odds of inactive disease and slower attainment of inactive disease, whereas early biologic initiation was not associated with either endpoint.
**Conclusion**
In this biologic-treated JIA cohort, approximately half of patients achieved inactive disease within 12 months. Baseline disease burden, rather than biologic timing alone, was the most consistent determinant of both achieving inactive disease and the pace of its attainment.
**Disclosure of interest**
None declared.

**Table ( Abstract P004)**

| Analysis | Predictor/outcome | Estimate |
|---|---|---|
| Logistic regression | Baseline disease-burden index | aOR 0.74, 95% CI 0.56–0.98; $p$=0.034 |
| Logistic regression | Early biologic initiation | aOR 0.65, 95% CI 0.31–1.36; $p$=0.252 |
| Cox regression | Baseline disease-burden index | HR 0.82, 95% CI 0.70–0.98; $p$=0.024 |
| Cox regression | Early biologic initiation | HR 0.99, 95% CI 0.65–1.50; $p$=0.946 |
| IPTW analysis | Early biologic initiation | OR 0.67, 95% CI 0.33–1.38; $p$=0.28 |

Abbreviations: aOR, adjusted odds ratio; HR, hazard ratio; IPTW, inverse probability of treatment weighting

## P005

### Residual pain in juvenile idiopathic arthritis during transition to adult care: a short-term longitudinal study

T. Çeltik[1,*], R. Torun[2], T. Aydın[3], Z. Kızıldağ[4], R. İşgüder[5], E. Ünsal[2], B. Bora[2]
[1]Rheumatology and Clinical Immunology, University of Rome Campus Biomedico; [2]Rheumatology Unit, "Bambino Gesù" Children Hospital IRCSS, Rome, Italy
**Correspondence:** A. Pilato
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**Introduction**
Increasing evidence suggests that clinical pain may persist in juvenile idiopathic arthritis (JIA) despite limited evidence of ongoing synovial inflammation. During transition to adult care, long-standing disease may contribute to pain mechanisms partially uncoupled from active inflammation. Data specifically addressing residual pain during transitioning JIA remain limited.
**Objectives**
To assess the cross-sectional and short-term longitudinal relationship between joint pain and ultrasound (US)-defined synovial inflammation in patients with JIA during transition.
**Methods**
Young adult non-systemic JIA patients eligible for transition of care underwent a prospective clinical and US evaluation. Clinical assessment included tender/painful-on-motion joints, pain visual analogue scale (VAS), global assessments and disease activity indices. Ultrasound was performed on clinically selected joints. US inflammatory activity was operationally defined as the presence of PD signal within an area of synovial hypertrophy. Analyses were performed at baseline (T0) and follow-up (T1 median interval 4 months, IQR 3-5), at joint and patient level. Paired analyses included joints scanned at both visits. Patients were classified as having no, resolved, new-onset or persistent discordance.
**Results**
Among 209 patients, 154 underwent ultrasound at T0, for 2020 scanned joints. At T0, 140/2020 joints (6.9%) were clinically painful and 67/2020 (3.3%) were PD-positive. Pain VAS correlated with the proportion of painful PD-negative joints at baseline (Spearman's ρ=0.49, $p$<0.001). At T1, 123 patients underwent ultrasound, for 1219 scanned joints. Painful PD-negative joints accounted for 64/1219 joints (5.3%) compared with 94/2020 (4.7%) at T0. In 902 paired patient-joint observations, their prevalence did not significantly change from T0 to T1 (65/902, 7.2% vs. 50/902, 5.5%; McNemar $p$=0.142). However, marked joint-level turnover was observed: 12/65 (18.5%) persisted, 53/65 (81.5%) resolved, and 38 new painful PD-negative joints developed. At patient level, 107 patients had paired ultrasound assessments. Discordance was absent at both visits in 51 patients (47.7%), resolved in 17 (15.9%), newly developed in 18 (16.8%) and persisted in 21 (19.6%). Pain VAS at T1 differed across phenotypes (Kruskal-Wallis $p$<0.001), being higher in persistent (median 5.0 [IQR 4.0-6.0]) and new-onset discordance (4.5 [2.0-7.0]) than in no discordance (1.0 [0.0-1.0]). Baseline discordance was associated with higher T1 pain in unadjusted analyses (median 4.0 vs. 1.0; $p$<0.001; OR for pain VAS >=4: 4.45, $p$<0.001), but not after adjustment for baseline pain.
**Conclusion**
In patients with JIA during transition, joint pain frequently occurred without US-defined synovitis. Over short-term follow-up, pain-inflammation discordance showed stable overall prevalence but marked joint-level turnover. Persistent or newly developed discordance was associated with substantially higher follow-up pain, suggesting the need for systematic pain assessment and tailored pain management strategies in the transitional care setting.
**Disclosure of interest**
None declared.

## P006

### Juvenile idiopathic arthritis: pathway from symptom onset to the first paediatric rheumatologist visit

K. Rebane[1], A. K. Tuomi[1,*], H. Kautiainen[2,3], K. Aalto[1]
[1]Pediatric Rheumatology Clinic, New Children's Hospital, Helsinki University Hospital, Helsinki; [2]Primary Health Care Unit, Kuopio University Hospital, Kuopio; [3]Folkhälsan Research Center, Helsinki, Finland
**Correspondence:** A. K. Tuomi
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**Introduction**
Diagnostic delay of juvenile idiopathic arthritis (JIA) is associated with poor outcome (1), though less is known about the effect of the time lag from the first symptoms of JIA to the first pediatric rheumatologist (PR) appointment.
**Objectives**
We aim to explore the time from first symptoms of arthritis to the referral to a PR, the diagnosis of JIA, and the treatment initiation. In addition, we aim to explore the associations of disease characteristics and outcome issues, especially receiving inactive disease with the time elapsed since seeing a PR.
**Methods**
The study cohort consisted consecutive patients referred to the rheumatology clinic of New Children's Hospital, Helsinki Finland between 1.1. 2021-31.12.2022, having oligo-or polyarthritis and diagnosed with JIA. The follow-up period was 12 months from the first visit. We included data concerning demographics, laboratory parameters, disease characteristics, and medications. Disease activity was evaluated by using JADAS-10 (2).
**Results**
86 patients with newly diagnosed oligoarticular and polyarticular JIA were included. Mean age at onset was 8.2 years (range: 0.5 – 15.1). 58 had oligoarticular (67%), and 28 polyarticular JIA (33%). Referral sources originated from the private or public healthcare, paediatric or orthopaedical clinic, dental or school health care. Patients who had younger age, higher BMI, positive ANA, elevated ESR, or pain mentioned in the referral were seen by the rheumatologist faster. 50% were seen by PR within three months after the first symptoms. Statistically significant difference was revealed between oligoarthritis and polyarthritis in receiving inactive disease at three months from the first visit ($p$=0.049) of which 25 [43% (95% CI: 30 to 57)] had oligoarthritis, and 6 [(21% (8 to 41)] had polyarthritis. The mean JADAS score for patients at the first visit was 9.1 (SD 6,8). 36% of the patients had inactive disease already at three months after the first visit. Entirely, 25% of patients had inactive disease from 3 to 12 months longitudinally.
**Conclusion**
Negative association was found between the time from the symptom onset to the first PR appointment and inactive disease at the one-year time point. Our findings highlight the importance of early initiation of the antirheumatic medication and emphasize the importance of an easy access to a paediatric rheumatologist.

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**Disclosure of interest**
None declared.

P007
**Characteristics of patients with juvenile idiopathic arthritis with revealed active arthritis according to the results of the temporomandibular joint MRI**
A. Artamonov[1], I. Avrusin[1,*], L. Sorokina [1], A. Burka[1], I. Chikova[1], O. Kalashnikova[1], M. Kostik [1]
[1]Hospital Pediatrics, Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation
**Correspondence:** I. Avrusin
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**Introduction**
Juvenile idiopathic arthritis (JIA) is the most common rheumatic disease in pediatric population. It is characterized by chronic inflammation of one or more joints lasting at least 6 weeks with onset at the age of 16 years. According to the criteria of the American College of Rheumatology in 2021, involvement of the hip, wrist, cervical spine (SHP) and temporomandibular joints (TMJ) refers to markers of an unfavorable prognosis.
**Objectives**
The aim of the study was to compare the examination data of patients with JIA with the results of TMJ MRI.
**Methods**
The study included 37 children with JIA who had complaints from the TMJ and underwent an MRI examination of the joint. All patients were examined and treated in the St. Petersburg State Medical University Clinic.
**Results**
Among the 37 examined patients, the most common complaints were pain in the projection of at least one TMJ in the anamnesis (75.7%), sounds in the joint (subjectively) (59.5%). There were also complaints such as jaw stiffness (35.1%), changes in eating habits due to pain in the TMJ (35.1%), fatigue with movements of the lower jaw (32.4%), and pain in extra-articular areas (32.4%). According to TMJ MRI, changes were detected in 22 children. Inflammatory changes (synovitis) were found in 8 (36.3%) of 22 children, while 6 out of 8 children with active synovitis had chondrodegenerative changes. Non-inflammatory changes manifested by TMJ dysfunction were noted in 14 (63.7%) patients. The non-inflammatory changes detected by MRI could reflect both the effects of previously suffered TMJ arthritis and impaired joint growth and remodeling in JIA. For a predictive assessment of the examination results, an analysis of sensitivity and specificity, odds ratio and risk ratio of each indicator in the group with TMJ arthritis according to MRI data was performed. The parameter "sounds in the joint (subjectively)" had a sensitivity of 62.5% and a specificity of 78.6%. OR - 1.67 [0.02; 1.11], and "jaw stiffness" - sensitivity 75.0%, specificity 71.4%. OSH - 7.5 [1.03; 54.1].
**Conclusion**
TMJ remains the most difficult joint to diagnose in course of JIA, including clinical, subjective, and instrumental changes. The lack of standardized methods for assessing function, diagnosis, and imaging limits understanding of the types and frequency of TMJ damage in children with JIA, which can affect not only short-term but also long-term outcomes, such as malocclusion, maxillofacial developmental abnormalities, and TMJ osteoarthritis.
**Disclosure of interest**
None declared.

P008
**Bilateral slipped capital femoral epiphysis in the setting of pediatric systemic sclerosis on tocilizumab therapy : case report with implications for early recognition and treatment**
B. Abu Saleh[1,*]
[1]Pediatric Rheumatology, SSMC, Abu Dhabi, UAE
**Correspondence:** B. Abu Saleh
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**Abstract**
**Introduction**
Slipped capital femoral epiphysis (SCFE) is the most common hip disorder in adolescence, characterized by posterior and inferior displacement of the proximal femoral epiphysis relative to the femoral neck through the growth plate. SCFE typically presents between ages 9 and 16, with a male predominance and increased incidence in Black, Hispanic, and American Indian/Alaska Native children. Bilaterality occurs in up to 80% of cases. The pathogenesis involves mechanical stress and metabolic factors that weaken the proximal femoral physis, with obesity being the most significant risk factor, as highlighted by the American Academy of Pediatrics. Endocrine disorders such as hypothyroidism and growth hormone abnormalities also contribute to physeal vulnerability.
**Objectives**
Systemic sclerosis (SSc), including its juvenile form, is a rare chronic autoimmune connective tissue disease characterized by fibrosis, vasculopathy, and immune dysregulation, affecting the skin and internal organs. Musculoskeletal manifestations are frequent and include arthralgia, arthritis, myopathy, tendon friction rubs, and joint contractures, but SCFE has not been reported as a recognized complication or association in the pediatric SSc population.
**Methods**
This report describes a 12-year-old female with juvenile systemic sclerosis on maintenance immunosuppressive therapy who presented

with bilateral SCFE. The coexistence of these two conditions is previously unreported in the literature. While SCFE is associated with obesity and endocrinopathies, there is no established link to autoimmune connective tissue diseases such as systemic sclerosis. The possibility of disease- or treatment-related factors contributing to physeal instability in this context warrants further investigation. Clinicians should remain vigilant for atypical presentations of SCFE in patients with chronic inflammatory or autoimmune conditions, although current evidence does not support a direct association.

**Disclosure of interest**

None declared.

## P009

### Biologic agent switching in non-systemic juvenile idiopathic arthritis: a tertiary center experience

D. Sarisoy[1,*], F. Aydın[1], P. Ö. Avar Aydın[1], O. Bahçeci[1], E. Erorhan[1], B. Tetik Dinçer[1], Z. B. Özçakar[1]
[1]Pediatric Rheumatology, Ankara University School of Medicine, Ankara, Türkiye
**Correspondence:** D. Sarisoy
*Pediatric Rheumatology 2026*, **24(S1):**P009

**Introduction**

Biologic disease-modifying anti-rheumatic drugs (bDMARDs) are being increasingly used to treat juvenile idiopathic arthritis (JIA). However, managing of "difficult-to-treat" patients who do not respond to first-line bDMARDs remains challenging in daily practice.

**Objectives**

The aim of this study was to determine the frequency and reasons for switching to bDMARDs in non-systemic JIA.

**Methods**

This retrospective study included patients with non-systemic JIA who required bDMARD treatments between June 2011 and June 2025. Clinical and laboratory data, as well as the treatment regimens, were compared between patients who did and did not require bDMARD switching.

**Results**

A total of 84 patients with JIA that required bDMARD treatments were included in the study. Median (IQR) disease duration was 71.5 (66) months and first-line bDMARD was initiated in a median (IQR) 11 (44.25) months. Oligoarthritis was the most common subgroup (41.6%), followed by enthesitis related arthritis (32.1%). First line bDMARD used was a TNF inhibitor for all patients except one. bDMARD switching was necessary for 21 (25%) patients, 81% of the first switches (17/21) were from a TNF inhibitor to another. The most common switch was from etanercet to adalimumab (66.7%, $n$=14), however seven (50%) of these patients were later switched to tocilizumab. bDMARD switching group had higher erytrocyte sedimentation rates at the time of diagnosis ($p$=0.007). bDMARD switch group was further characterised with higher JADAS-27 scores at 6th and 12th months, higher JADI scores at last visit, more articular relapses and lower uveitis rates ($p$=0.03, 0.009, 0.008, 0.04 and 0.022, respectively). A total of 33 switches were made in 21 patients and the most common reason was inadequite response (42.4%, $n$=14), followed by disease flares (33.3%, $n$=11) and adverse affects (24.2%, $n$=8). Nine (10.7%) patients needed multipl bDMARD switchings. First bDMARD used was etanercept for eight patients and anakinra for one patient who required multiple switchings. Three patients that needed multipl switches had concomittant inflammatory diseases (3 familial Mediterranean fever, 1 inflammatory bowel disease).

**Conclusion**

The introduction of bDMARDs has revolitionised the management of JIA. However, patients with a high inflammatory burden and high activity scores may need to switch to other biological treatments. Whilst switching agents is generally effective in the management of these "difficult-to-treat" patients, further data is required to develop clearer guidelines.

**Disclosure of interest**

None declared.

## P010

### Uveitis in patients with non-systemic juvenile idiopathic arthritis: a tertiary care center experience with long term follow up

D. Sarisoy[1,*], F. Aydın[1], P. Ö. Avar Aydın[1], F. N. Yalçındağ[2], Z. B. Özçakar[1]
[1]Pediatric Rheumatology; [2]Ophtalmology, Ankara University School of Medicine, Ankara, Türkiye
**Correspondence:** D. Sarisoy
*Pediatric Rheumatology 2026*, **24(S1):**P010

**Introduction**

Uveitis represents one of the most clinically important extra-articular complications of juvenile idiopathic arthritis (JIA). Due to its frequently asymptomatic and insidious nature, delayed recognition may result in irreversible ocular damage.

**Objectives**

This study aimed to determine the frequency of uveitis and to identify associated risk factors in patients with non-systemic JIA.

**Methods**

A retrospective analysis was conducted on non-systemic JIA patients followed at a tertiary pediatric rheumatology center between 2010 and 2024. Demographic features, clinical findings, and laboratory parameters were extracted from medical records. Patients were categorized into groups with and without uveitis, and comparative analyses were performed to evaluate potential risk factors.

**Results**

144 patients (92 female, 63.9%) were included in the study. Oligoarthritis was the most common subtype (57.6%). Uveitis was detected in 26 patients (18.1%) with a median (IQR) of 67.5 (64.25) months of follow-up. The group with uveitis was characterized by female sex ($p$=0.048), younger age at the time of JIA diagnosis ($p$=0.014), more common ANA positivity ($p$=0.039), and more frequent articular flares ($p$=0.014). No significant difference was found between groups in JIA subtypes or affected joints ($p$>0.05). 12 patients (46.2%) with uveitis experienced complications, including posterior synechiae, glaucoma, cataract, and band keratopathy. Notably, 3 of these patients already had complications at the time of uveitis diagnosis.

**Conclusion**

Uveitis in JIA patients may progress silently and lead to significant morbidity if not detected early. According to our results, particular attention should be given to patients with early disease onset, ANA positivity, female sex, and recurrent joint flares. Structured and regular ophthalmologic screening is essential for early diagnosis and prevention of vision-threatening complications.

**Disclosure of interest**

None declared.

## P011

### Interpreting liver enzyme elevations in biologic-treated juvenile idiopathic arthritis: insights from a pediatric cohort and a clinical algorithm

B. Başer Taşkın[1,*], A. Doğru Kılınç[1], Ö. Akgün[1], N. Aktay Ayaz[1]
[1]Division of Pediatric Rheumatology, Istanbul University Faculty of Medicine, İstanbul, Türkiye
**Correspondence:** B. Başer Taşkın
*Pediatric Rheumatology 2026*, **24(S1):**P011

**Introduction**

Juvenile idiopathic arthritis (JIA) is the most common chronic inflammatory rheumatic disease of childhood. The widespread use of bDMARDs has markedly improved disease control and long-term outcomes. However, elevated liver function tests (LFTs) observed during biologic therapy remain a clinical challenge, as they may be associated with disease activity, infections, or concomitant conventional DMARD use.

**Objectives**

To evaluate the frequency, severity, and clinical course of LFT elevations in children with JIA receiving biologic therapy, to explore associated factors, and to propose a practical monitoring strategy based on real-world data.

**Table 1 (Abstract P011)** Comparison of clinical and laboratory variables between patients with single episodic and recurrent/persistent liver function test elevation

| | Single episodic (*n* = 36) | Recurrent/persistent (*n* = 16) | *p* value* |
|---|---|---|---|
| Age at JIA diagnosis, months | 68.5 (14–198) | 50.5 (9.0–184) | 0.36 |
| Age at biologic initiation, months | 88.0 (30–203) | 77 (15.0–192) | 0.15 |
| Age at LFT elevation, months | 135 (38-212) | 81.5 (16.5-195) | **0.03** |
| Time to LFT elevation after biologic initiation, months | 9.0 (0.5–113) | 3.0 (0.5–25) | 0.08 |
| Follow-up duration, months | 61.5 (6–146) | 91.5 (21–217) | **0.03** |
| Aspartate aminotransferase, U/L | 56.0 (12–400) | 70.0 (26–1406) | **0.035** |
| Alanine aminotransferase, U/L | 55.5 (33–298) | 108.5 (14–2071) | **0.007** |
| Duration of LFT elevation, days | 14.0 (4–30) | 71.5 (10–390) | **<0.001** |
| Duration of biologic therapy, months | 47.5 (9-122) | 39.0 (0.5-152) | 0.59 |

**Methods**
Medical records of children with JIA receiving biologic therapy who developed elevated LFTs during treatment were retrospectively reviewed. Liver function test elevations were classified according to severity and clinical course, and potential etiological factors were analysed.
**Results**
Among 213 biologic-treated JIA patients, LFT elevations were identified in 52 (24.4%). Elevations were predominantly mild (86.5%) and transient, with normalization within one month in 80.8% of cases. Thirty-six patients (69.2%) experienced a single episodic elevation, while 16 (30.8%) had recurrent or persistent abnormalities. Methotrexate was the most frequent concomitant csDMARD. No significant differences were observed between anti-TNF and anti-interleukin therapies regarding LFT elevation patterns. Intercurrent infections, disease activity/systemic flare, obesity, and suspected drug-related causes contributed variably; clinically significant hepatotoxicity was uncommon and permanent biologic discontinuation was rarely required (see Table 1).
**Conclusion**
In biologic-treated children with JIA, LFT elevations are common but usually mild, reversible, and multifactorial. Routine laboratory monitoring combined with structured etiological assessment supports safe continuation of biologic therapy in patients. A stepwise, severity-based clinical algorithm may aid decision-making in routine pediatric rheumatology practice.
**Disclosure of interest**
None declared.

## P012

### Factors associated with the development of articular damage in juvenile idiopathic arthritis

Ç. Yildiz[1,*], B. Küçükali[1], Ö. Özenli Yağcı[1], D. Gezgin Yıldırım[1], S. A. Bakkaloğlu[1]
[1]Pediatric Rheumatology, Gazi University Faculty of Medicine, Ankara, Türkiye
**Correspondence:** Ç. Yildiz
*Pediatric Rheumatology 2026*, **24(S1):**P012

**Introduction**
Juvenile idiopathic arthritis (JIA) is the most common chronic rheumatic disease in childhood and may result in permanent joint damage (1). In non-systemic subtypes, disease course varies from minimal sequelae to progressive structural damage (2). Early identification of patients at risk is essential for risk stratification and individualized treatment.
**Objectives**
This study aimed to investigate factors associated with the development of articular damage and to identify independent predictors in patients with non-systemic JIA.
**Methods**
A total of 216 patients with non-systemic JIA followed at a tertiary center were included in this retrospective study. Demographic, clinical, laboratory, and treatment data were analyzed. Articular damage was assessed using the Juvenile Arthritis Damage Index-Articular (JADI-A), with JADI-A ≥1 indicating damage (2). Factors associated with articular damage were evaluated by univariate and multivariate logistic regression analyses. A p value <0.05 was considered statistically significant.
**Results**
The median follow-up duration of the 216 patients with non-systemic JIA was 60 months (IQR: 21–87.5). JADI-A positivity was detected in 37% (*n*=80) of patients. Patients with articular damage had a younger age at symptom onset ($p$=0.043), longer methotrexate exposure ($p$=0.004), and a higher cumulative number of involved joints during follow-up ($p$<0.001). In univariate analysis, extended oligoarticular course, the PsA/undifferentiated group, and methotrexate treatment duration were associated with articular damage. In multivariate analysis, extended oligoarticular course (OR: 3.33; 95% CI: 1.24–8.95; $p$=0.017) and methotrexate treatment duration (OR: 1.03; 95% CI: 1.01–1.05; $p$=0.002) remained independent predictors of damage. Diagnostic delay and timing of treatment initiation were not associated with articular damage.
**Conclusion**
In patients with non-systemic JIA, articular damage was mainly associated with an extended oligoarticular course and longer methotrexate exposure. No significant relationship was observed with diagnostic delay or timing of treatment initiation. The predictive model showed moderate discriminative performance in the oligoarticular subgroup. Cumulative anti-TNF exposure appeared to correlate with articular damage, likely reflecting higher disease severity, treatment resistance, biologic switching, and prolonged methotrexate use rather than a direct drug effect. Overall, these findings suggest that disease phenotype and cumulative inflammatory burden during follow-up may contribute to structural articular damage.

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**Disclosure of interest**
None declared.

## P013

### Agreement between knee joint position sense and lower extremity dynamic proprioceptive balance test in children/adolescents with juvenile idiopathic arthritis and healthy

Z. Tatar[1], E. Gur Kabul[2,*], M. Bali [3], B. Basakci Calik[3], S. Yuksel[4]
[1]Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Inonu University, Malatya; [2]Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Usak University, Usak; [3]Faculty of Physiotherapy and Rehabilitation, Pamukkale University, Denizli; [4]School of Medicine, Department of Pediatric Rheumatology, Canakkale Onsekiz Mart University, Canakkale, Türkiye
**Correspondence:** E. Gur Kabul
*Pediatric Rheumatology 2026*, **24(S1):**P013

**Introduction**
Assessing proprioception in the knee joint, a crucial joint for function, is very important. Frequently used knee joint position sense tests stand out for their ease of application in clinical settings. However, the extent to which these tests reflect functional abilities is not clear.
**Objectives**
The aim of this study was to investigate the agreement between knee joint position sense and lower extremity dynamic proprioceptive balance test results in children with Juvenile Idiopathic Arthritis (JIA) and healthy children/adolescents.

**Methods**
The study included 35 children/adolescents with JIA (mean age 13.37±2.74 years) and 35 healthy children/adolescents (mean age 12.14±3.53 years). Knee joint position sense (digital goniometer at 15°, 30°, 45°, 60°) was evaluated bilaterally, and lower extremity dynamic proprioceptive balance test was assessed using the Sensamove Sensbalance Maxiboard Software v2 device. Shapiro-Wilk tests were conducted to check the normality of the data. The Bland-Altman plot was built to present the agreement between the tests. The unstandardized beta and the '95% limits of agreement' are presented to provide the difference between the calculations. The concurrent validity of the tests was analyzed with the Pearson correlation coefficient. The significance level was accepted as $p<0.05$ and interpreted.
**Results**
In healthy children/adolescents, a low correlation was observed at 30° ($r=0.370$, $p=0.028$), while no significant correlations were found at 15°, 45°, or 60°. Agreement was observed between the 30° knee joint position sense and lower extremity dynamic proprioceptive balance test (mean difference: 0.91; SD: 1.06; unstandardized beta = 0.792, $p<0.001$). In children/adolescents with JIA, no significant correlations were found between the lower extremity dynamic proprioceptive balance test and knee joint position sense at 15° ($r=-0.224$, $p=0.196$), 30° ($r=-0.019$, $p=0.915$), 45° ($r=0.025$, $p=0.887$), or 60° ($r=0.225$, $p=0.193$). Additionally, there was no agreement between the 30° knee joint position sense and lower extremity dynamic proprioceptive balance test (mean difference:3.11; SD:2.39; unstandardized beta=1.806, $p<0.001$).
**Conclusion**
While agreement was observed between 30° knee joint position sense and lower extremity dynamic proprioception balance in healthy children/adolescents, no knee joint position sense at any angle in children/adolescents with JIA showed agreement with lower extremity dynamic proprioception balance.
**Disclosure of interest**
None declared.

## P014

### Efficacy and safety of hip intra-articular corticosteroid injections in juvenile idiopathic arthritis in a single-center pilot study

F. Di Stasio[1,*], M. Rossano[1], G. B. Beretta[1], S. Mazzitelli[1], D. Montin[1], S. Lanni[1], G. Filocamo[1,2]
[1]Pediatric Immuno-Rheumatology Unit, Fondazione IRCCS Cà Granda Ospedale Maggiore Policlinico; [2]Università degli studi di Milano, Milan, Italy
**Correspondence:** F. Di Stasio
*Pediatric Rheumatology 2026*, **24(S1):**P014

**Introduction**
Intra-articular corticosteroid injection (IACI) is an effective treatment for juvenile idiopathic arthritis (JIA), providing rapid symptom relief and prolonged disease control with a favorable safety profile. However, hip injections are less commonly performed because of technical challenges requiring imaging guidance and anesthesia, as well as safety concerns related to potential complications, including femoral head osteonecrosis, rapidly progressive osteoarthritis, and subchondral insufficiency fractures reported in the literature (1).
**Objectives**
To describe a single-centre experience of hip IACIs in patients with JIA.
**Methods**
We conducted a single-centre observational study including JIA patients who underwent hip IACI between 2018 and 2026. This was a retrospective analysis of prospectively collected data performed according to routine clinical practice. Evaluations at baseline, 3 months, and subsequent follow-up visits included the Italian version of the Juvenile Arthritis Multidimensional Assessment Report (JAMAR) and Physician Global Assessment (PGA, 0-10 VAS). Magnetic resonance imaging (MRI), when available, was reviewed before and after the procedure to assess structural changes and complications.
**Results**
Nine patients underwent 36 ultrasound-guided hip IACIs, with a median follow-up of 2.5 years (IQR 1.3-5.2). The majority were male (5/9). The median age at first injection was 9.1 years (IQR 8.3-12.7), and the median age at disease onset was 7.1 years (IQR 3.2-11.9). JIA subtypes included rheumatoid factor (RF)-negative polyarticular JIA (4/9), RF-positive polyarticular JIA (1/9), enthesitis-related arthritis (1/9), extended oligoarthritis (1/9), and systemic JIA (2/9). Systemic JIA was associated with a higher number of injections per patient: one patient received eight injections in the left hip and five in the right hip, while another underwent six injections in the left hip and seven in the right hip. All patients experienced rapid improvement in pain and stiffness, with the best response observed at the 3-month follow-up. The median Juvenile Arthritis Functionality Scale-lower limbs score decreased from 5 (IQR 3-8.5) before treatment to 1 (IQR 0-1.75) after IACI. Post-procedural MRI was available in five patients. No new structural damage or progression of pre-existing lesions was observed in these patients. One patient underwent total hip arthroplasty at 19 years of age. This patient had radiographic evidence of severe joint deterioration warranting replacement prior to the first injection. In this case, joint injections allowed postponement of surgery until completion of skeletal growth, maintaining acceptable function and pain control.
**Conclusion**
In our cohort, ultrasound-guided hip IACI was a safe and effective procedure in JIA, providing rapid pain relief and improved lower-limb function. No evidence of injection-related structural hip damage was observed during follow-up in most patients.

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**Disclosure of interest**
None declared.

## P015

### The "comparison of STep-up and step-down therapeutic strategies in childhood ARthritiS" (STARS) trial: baseline characteristics of the patients

F. Migliavacca[1,*], F. Ridella[1], M. Burrone[2], M. Mazzoni[2], R. Naddei[3], F. Bovis[4], G. Filocamo[5], F. De Benedetti[6], F. Licciardi[7], A. Civino[8], P. Barone[9], N. Ruperto[10,11], M. Gattorno[2], A. Ravelli[12], A. Consolaro[1,2]
[1]Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genoa; [2]UOC Rheumatology and Autoinflammatory Diseases, IRCCS Giannina Gaslini Institute, Genoa; [3]Department of Translational Medical Sciences, University of Naples Federico II, Naples; [4]Department of Health Sciences, University of Genoa, Genoa; [5]Pediatric Rheumatology, Fondazione IRCCS Cà Granda Ospedale Maggiore Policlinico Milano, Milan; [6]Pediatric Rheumatology, IRCCS Pediatric Hospital Bambino Gesù, Rome; [7]UOC Rheumatology and Autoinflammatory Diseases, University of Torino, Turin; [8]UOSD Rheumatology and Autoinflammatory Diseases, Vito Fazi Hospital, Lecce; [9]Pediatrics, University Hospital of Catania, Catania; [10]UOC Pediatric Rheumatology, Fonazione IRCCS San Gerardo dei Tintori; [11]Department of Medicine and Surgery, University of Milano-Bicocca, Monza; [12]Scientific Directorate, IRCCS Giannina Gaslini Institute, Genoa, Italy
**Correspondence:** F. Migliavacca
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**Introduction**
The "comparison of STep-up and step-down therapeutic strategies in childhood ARthritiS" (STARS) trial aims to investigate whether an early aggressive therapeutic intervention, based on the initial start of synthetic and biologic DMARDs (Step-down strategy), is superior to an approach based on treatment escalation conducted following the treat-to-target principle (Step-up strategy), in children with Juvenile idiopathic arthritis (JIA).
**Objectives**
We present the baseline characteristics of patients in the 2 treatment arms.
**Methods**
JIA patients with recent-onset oligoarthritis or rheumatoid factor negative polyarthritis, ≤ 17 years old at diagnosis and drug-naïve were enrolled between May 2019 and October 2024. Follow-up was completed in November 2025. Disease activity assessments, medication start and stop dates, and severe adverse events/events of special interest were

collected at 3, 6, 9, and 12 months. After the screening visit, patients were randomised into 2 treatment arms: "Step-up" and "Step-down". Patients in the Step-up arm were treated according to a conventional treatment escalation/treat-to-target strategy. Patients in the Step-down arm were treated with an early, combined, aggressive therapy for 6 months; In both arms, treatment was based on disease severity. Children with oligoarthritis without poor prognostic features (per 2011 American College of Rheumatology criteria) were classified as "less severe" while those with RF-negative arthritis or oligoarthritis with poor prognostic features were classified as "more severe". At each step of the study the JADAS10 was calculated for all the patients. The effectiveness of the two therapeutic strategies will be compared by assessing the frequency of clinical remission (CR) at 12 months, defined as the persistence of inactive disease for at least 6 months based on the JADAS10.

**Results**

The main baseline characteristics of the patients are shown in the table below and show the homogeneity between the 2 arms.

| Features | Arm 1 | Arm 2 | *p* | Overall |
|---|---|---|---|---|
| Number of patients | 120 | 120 | | 240 |
| Oligoarthritis, n (%) | 73 (60.8) | 74 (61.7) | | 147 (61.3) |
| Polyarthritis (FR negative), n (%) | 47 (39.1) | 46 (38.3) | | 93 (38.7) |
| More severe group, n (%) | 53 (44.2) | 57 (47.5) | 0.698 | 110 (45.8) |
| Physician global assessment VAS (median [IQR]) | 5.5 [4.0, 7.0] | 5.2 [3.9, 7.5] | 0.915 | 5.5 [4.0, 7.1] |
| N. active joints (median [IQR]) | 2.5 [1.0,5.0] | 3.0 [1.0,6.2] | 0.974 | 3.0 [1.0, 5.0] |
| Erythrocyte Sedimentation Rate (median [IQR]) | 28.0 [10.0, 54.0] | 33.5 [17.0, 49.0] | 0.824 | 30.0 [12.0, 51.0] |
| JADAS10 (median [IQR]) | 13.5 [9.9, 17] | 14.8 [8.5, 20.0] | 0.367 | 14.0 [9.4, 18.1] |

**Conclusion**

With the STARS trial, we aim to provide important evidence supporting the first line treatment choices for children with oligoarticular and polyarticular JIA. If the superiority of an early aggressive therapy was demonstrated, further biological studies on the window of opportunity for JIA patients will be needed.

**Trial registration identifying number**

EU clnical trials Eudrac Number 2018-001931-27 Clinical Trials.gov registry (NCT03728478).

**Disclosure of interest**

None declared.

## P016

### Long term outcome and disease burden of JIA patients managed entirely in the biologic era

F. Ridella[1,*], E. La Camera[2], F. Migliavacca[2], G. Zilli[2], R. Caorsi[3], S. Rosina[3], C. Matucci-Cerinic[3], C. Malattia[3], M. Mazzoni[3], A. Ravelli[4], M. Gattorno[3], A. Consolaro[2,3]

[1]Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI); [2]Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genoa; [3]UOC Rheumatology and Autoinflammatory Diseases; [4]Scientific Directorate, IRCCS Giannina Gaslini Institute, Genoa, Italy

**Correspondence:** F. Ridella

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**Introduction**

Therapeutic options for juvenile idiopathic arthritis (JIA) have markedly expanded over the past 25 years, enabling faster and more frequent achievement of inactive disease and likely reducing the risk of long–term articular and extra–articular damage. However, current evidence on long–term outcomes largely derives from cohorts whose treatment began before the biologic era, with biologic agents introduced only later in the disease course, potentially limiting their impact. Data on patients treated exclusively within the biologic era remain scarce.

**Objectives**

To describe disease status, functional ability, and overall disease burden in an Italian cohort of JIA patients with 20 years of disease duration and disease onset after 2003.

**Methods**

Electronic medical records and internal registries were screened to identify patients with disease onset in 2004 and 2005 and at least 10 visits at the study Unit. Demographic and baseline clinical characteristics were collected. Eligible subjects were invited to participate in either an in–person or remote assessment. During the visit, information about the most recent rheumatology evaluation, transition to adult care, current treatment, occupation, and the impact of JIA on education, employment, and sports participation was collected. Patient–reported outcomes, including physical function and disease activity, were collected using the Juvenile Arthritis Multidimensional Assessment Report (JAMAR).

**Results**

Patients with JIA onset in 2004 and 2005 were screened so far; 45 patients were identified; 7 were excluded due to fewer than 10 follow–up visits. Among the 38 eligible subjects, 5 lacked updated contact information and 3 (5.7%) declined participation. Thirty patients (90% female; median age 24.4 years, IQR 23.2–26.8) were included in the current analysis; 40% had persistent oligoarthritis. Over the disease course, 83% received methotrexate and 67% were treated with at least one biologic agent. At the time of assessment, 80% remained under rheumatologic follow–up and 75% were still receiving JIA–related therapy. Regarding disease impact, 20%, 30%, and 60% reported that JIA had affected their education, employment, and sports participation, respectively. Functional ability was normal in 50% of participants. Median patient–reported JADAS was 0.5 (0–2), and 62% self–reported being in remission.

**Conclusion**

In this cohort of JIA patients managed entirely within the biologic era, most individuals—after 20 years of disease—remain in regular rheumatologic follow–up and continue to receive treatment. Patients reported minimal impact on education and employment, with a greater effect on sports participation. Functional ability was normal or only mildly impaired in most cases. These findings may help clinicians counsel families regarding long–term expectations in the biologic era. Efforts are ongoing to reach all eligible subjects to strengthen the robustness of the analysis.

**Disclosure of interest**

F. Ridella: None declared, E. La Camera: None declared, F. Migliavacca: None declared, G. Zilli: None declared, R. Caorsi: None declared, S. Rosina: None declared, C. Matucci-Cerinic: None declared, C. Malattia: None declared, M. Mazzoni: None declared, A. Ravelli: None declared, M. Gattorno: None declared, A. Consolaro Grant / Research Support with: Pfizer investigator initiated grant, Novartis speaking fees.

## P017

### An investigation of the relationship between functional status and biopsychosocial dimensions in cases with juvenile idiopathic arthritis

A. Barlak[1], O. Tüfekçi[1,*], E. Aliyev[2], M. O. Erkan[2], Y. Bilginer[2], S. Özen[2], E. Ünal[1]

[1]Faculty of Physical Therapy and Rehabilitation; [2]Faculty of Medicine, Department of Pediatrics, Division of Pediatric Rheumatology, Hacettepe University, Ankara, Türkiye

**Correspondence:** O. Tüfekçi

*Pediatric Rheumatology 2026*, **24(S1):**P017

**Introduction**

Juvenile idiopathic arthritis (JIA) is a chronic inflammatory disease of unknown etiology that begins before the age of 16 and is characterized by periods of disease activity and remission. In JIA, patient-reported biopsychosocial characteristics such as pain, fatigue, and disease perception are among the key factors determining the level of functional impairment.

**Objectives**

The study aimed to investigate the relationship between functional status and patient-reported disease activity, joint perception, and fatigue in cases with JIA, and to identify variables that predict functional status.

**Methods**
This study was designed as a descriptive-analytical study. Forty cases with JIA were included in the study. The cases were assessed from a biopsychosocial perspective using the Juvenile Arthritis Biopsychosocial Questionnaire-Patient (JAB-Q-$_P$). The JAB-Q Functionality subscale was used as the dependent variable. The JAB-Q Disease Activity, JAB-Q Joint Perception, and JAB-Q Fatigue subscales were included in the model as independent variables. Descriptive statistics were calculated first during data analysis. After examining the relationships between the variables, multiple linear regression analysis was performed to identify the independent variables predicting functional status. Model assumptions (normality, linearity, multicollinearity) were checked. VIF (Variance Inflation Factor) and tolerance values were examined for multicollinearity. A significance level of $p<0.05$ was applied.
**Results**
As a result of the multiple linear regression analysis, the model was found to be statistically significant ($F(3,34)=11.874$, $p<0.001$). The model was found to explain 51% of the variance in the JAB-Q Functionality subscale. ($R^2 = 0.512$; Adjusted $R^2 = 0.469$). JAB-Q Disease Activity ($\beta = 0.371$, $p = 0.006$) and JAB-Q Joint Perception ($\beta = 0.368$, $p = 0.010$) were identified as independent and significant predictors of functionality. The JAB-Q Fatigue variable, however, did not make a significant contribution to the model. ($\beta=0.259$, $p=0.075$).
**Conclusion**
In this study, it was observed that self-reported joint-related awareness in cases with JIA influenced their functional cognition regarding their joints but was not affected by fatigue. On the other hand, the fact that fatigue did not affect functionality was interpreted to mean that cases maintained their functionality even when they felt tired. In conclusion, considering the cognition related to the affected joints in cases with JIA, it was determined that these cases could remain physically active even if their functionality was negatively affected and they felt tired. Therefore, this study emphasized the importance of supporting the development of functional efficiency—achieved by engaging the muscles surrounding the joint at proper angles of contraction—through cognitive restructuring.
**Disclosure of interest**
None declared.

## P018

### Should pediatric rheumatologists perform screening of thyroid function and autoimmunity in children with juvenile idiopathic arthritis? Results of a systematic review and meta-analysis

I. Liguoro[1], B. D'Agostini[1], F. Franco[1], G. Martini[1,*]
[1]Pediatrics, University Hospital, Udine, Italy
**Correspondence:** G. Martini
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**Introduction**
Recent observational studies outlined the concomitant presence of autoimmune diseases in children with juvenile idiopathic arthritis (JIA), including endocrine autoimmunity. Despite the growing evidence of thyroid involvement in JIA, available data are heterogeneous and fragmented.
**Objectives**
We aimed to systematically review the available evidence on the prevalence of thyroid autoantibody positivity and thyroid dysfunction in patients with JIA.
**Methods**
PubMed/Medline, Cochrane, and Scopus databases were approached to identify studies reporting data on thyroid autoantibody positivity and dysfunction in children with JIA. The review was conducted in compliance with the Preferred Reporting Items for Systematic Reviews and Meta-analyses (PRISMA) guidelines and registered on the PROSPERO system (1).
**Results**
A total of 15 studies were included in the final analysis, encompassing 19,015 children with JIA (13,225 females, 69.6%) with a weighted mean age of 8.0 years (range 1.8-21 years). The pooled prevalence estimates of anti-thyroid antibody positivity and thyroid dysfunction were 0.04 (95% CI: 0.03-0.05) and 0.04 (95% CI: 0.03 - 0.08), respectively. The risk difference for thyroid autoantibody positivity between children with JIA and controls was 0.08 (95% CI: 0.02-0.14). Possible risk factors were analyzed: the presence of family history for thyroid autoimmune disease (OR 3.91, 95%CI 1.86- 8.25), concurrent ANA positivity (OR 1.62, 95%CI 1.13-2.31) and older age (MD 2.10 years, 95% CI 1.32-2.89) were associated with thyroid autoantibody positivity and/or thyroid dysfunction in children with JIA. No significant difference emerged for the specific JIA subtype.
**Conclusion**
Thyroid dysfunction may be detected in children with JIA, with a significant risk difference in comparison to healthy controls. The presence of specific clinical characteristics, such as family history, ANA positivity, and older age at JIA onset, may suggest which patients could benefit from thyroid function and autoantibody screening.

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**Disclosure of interest**
None declared.

## P019

### Does juvenile idiopathic arthritis onset in the COVID era change its phenotype? A monocentric long-term study

G. Mastrangelo[1], F. Vittadello[2], F. Tirelli[1], A. Meneghel[1,*], F. Zulian[1]
[1]Division of Pediatric Rheumatology, Department of Women and Children Health, University of Padua; [2]Department of Research and Statistical Analysis, Explora, Padua, Italy
**Correspondence:** A. Meneghel
*Pediatric Rheumatology 2026*, **24(S1):**P019

**Introduction**
Juvenile idiopathic arthritis (JIA) is a heterogeneous group of chronic arthritis with onset before age 16 and unknown etiology. Clinical observations suggest that JIA cases during the SARS-CoV-2 pandemic may differ from those in the pre-COVID era. SARS-CoV-2 is a known trigger of autoimmune phenomena during infection and in the following weeks.
**Objectives**
We aimed to compare the demographic, laboratory, clinical, treatment characteristics and outcome of JIA patients with disease onset during the SARS-CoV-2 pandemic (2020–2024) versus the pre-pandemic period (2015–2019).
**Methods**
Retrospective analysis of prospectively collected data from children diagnosed with oligoarticular or polyarticular JIA between 2015 and 2024 at the University Hospital of Padua. Patients were stratified into four groups by JIA subtype and era of onset: oligoarticular pandemic-onset (Group 1, $n=76$), oligoarticular pre-pandemic (Group 2, $n=83$), polyarticular pandemic-onset (Group 3, $n=20$), and polyarticular pre-pandemic (Group 4, $n=20$). Demographic, clinical, laboratory, and treatment data were collected. The primary outcome was disease activity at 36 months, assessed by the Wallace criteria. SARS-CoV-2 exposure was defined by a positive RT-PCR on nasopharyngeal swab, reactive IgG antibodies (nucleocapsid and/or spike), or a positive rapid antigen test. Descriptive statistics, chi-squared or Fisher's exact test, and Mann–Whitney U test were used for between-group comparisons; Kaplan-Meier survival analysis compared time to remission between pandemic and pre-pandemic groups.
**Results**
199 children were included. Oligoarticular JIA: pandemic-onset patients (Group 1) were older at diagnosis (7.4 vs. 6.0 years, $p<0.05$), had a significantly lower incidence of uveitis (10.5% vs. 22.9%, $p=0.038$), more frequently received non-steroidal anti-inflammatory drugs (NSAIDs) over conventional synthetic disease-modifying antirheumatic drugs (csDMARDs), and achieved substantially higher remission rates at 36 months (58.3% vs. 33.3%, $p=0.008$), confirmed by Kaplan-Meier analysis (log-rank $p=0.001$) than Group 2. Polyarticular JIA: pandemic-onset patients (Group 3) had a lower proportion of females (55% vs. 90%, $p=0.013$); no significant differences were observed in age at diagnosis, ANA, RF, HLA-B27, uveitis frequency, or disease remission at 36 months (log-rank $p=0.485$) in comparison to Group 4.

**Conclusion**
Oligoarticular JIA patients with onset during the Covid-19 epidemic (Group 1) show a lower incidence of uveitis and a better clinical outcome, with less need for immunosuppressive therapy and greater disease remission at 36 months, than those with pre-pandemic years onset (Group 2). In contrast, polyarticular JIA patients showed no significant differences between the two groups. A longer follow up analysis is needed to confirm these preliminary data.
**Disclosure of interest**
None declared.

P021
**Risk of flare after etanercept versus methotrexate discontinuation in children with juvenile idiopathic arthritis in remission on combined therapy: a retrospective cohort study**
G. Tarantino[1,*], S. Delli Noci[1], M. I. Petrone[1], R. Nicolai[1], A. Aquilani[1], S. Magni-Manzoni[1], F. De Benedetti[1], E. Marasco[1]
[1]Rheumatology Division, Bambino Gesù Children Hospital IRCCS, Rome, Italy
**Correspondence:** G. Tarantino
*Pediatric Rheumatology 2026*, **24(S1):**P021

**Introduction**
TNF inhibitors (TNFi) have revolutionized treatment of juvenile idiopathic arthritis (JIA) with unsatisfactory response to conventional DMARDs such as methotrexate (MTX). Many children on combined therapy (MTX plus TNFi) achieve long-lasting remission, yet no guidelines exist on appropriate drug withdrawal. Practice varies across centers and remains provider-dependent. While withdrawal carries a definite flare risk in adults with rheumatoid arthritis, data in children with JIA are scarce.
**Objectives**
To compare flare rates after etanercept (ETN) or MTX discontinuation in non-systemic JIA patients achieving remission on combined therapy.
**Methods**
JIA patients on MTX and ETN were retrospectively grouped by withdrawn drug. We included patients who received a first course of combined therapy for at least 24 months, who maintained remission for at least 6 months before discontinuation, and were followed for at least 12 months after drug discontinuation. Six patients were included in both groups, having first attempted ETN discontinuation; following a disease flare, combined therapy was restarted for two years before MTX was subsequently discontinued. Time to flare was analyzed by Kaplan-Meier method; groups were compared using the log-rank test. A Cox proportional hazards model estimated the hazard ratio (HR) for flare; the proportional hazards assumption was verified with Schoenfeld residuals ($p$=0.05 threshold).
**Results**
We enrolled 106 patients: 70 stopped ETN, 36 stopped MTX. Groups were comparable for sex, JIA subtype, ANA positivity, and disease duration at ETN and MTX start. Uveitis was more frequent in the ETN-stop group (27% vs. 6%, $p$=0.017). After ETN withdrawal, 54 children (77%) flared within 12 months, versus 11 (31%) after MTX discontinuation. Flare-free survival at 12 months was 27% (95% CI: 18–40%) and 69% (95% CI: 56–86%), respectively. The difference was statistically significant (log-rank $\chi^2$=14.2, $p$=0.0002). Cox analysis confirmed MTX discontinuation carried a significantly lower flare risk versus ETN discontinuation (HR 0.30, 95% CI: 0.16–0.59, $p$=0.0004).
**Conclusion**
Flare risk is significantly higher after ETN discontinuation than after MTX withdrawal in children with non-systemic JIA in remission on combined therapy. When feasible, MTX should be withdrawn before ETN. Large multicenter studies investigating predictors of flare are needed to guide evidence-based withdrawal strategies.

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**Disclosure of interest**
None declared.

P022
**Ultrasound-guided intra-articular corticosteroid injection under intra-articular anesthesia in pediatric rheumatic diseases: clinical response, tolerability, and patient experience**
C. Cirone Papa Giannotti[1], D. Gerent Petry Piotto[1], G. Clemente[1,*], I. F. Manoel[1], M. Mariti Fraga[1], R. Ragazzin[1], M. T. Terreri[1]
[1]Pediatrics, Universidade Federal de São Paulo, Sao Paulo, Brazil
**Correspondence:** G. Clemente
*Pediatric Rheumatology 2026*, **24(S1):**P022

**Introduction**
Intra-articular corticosteroid injection is an established treatment for synovitis in pediatric immune-mediated rheumatic diseases. Ultrasound guidance improves procedural accuracy; however, pain and tolerability remain important concerns, particularly in children.
**Objectives**
To evaluate clinical response, tolerability, safety, and patient-reported experience following ultrasound-guided intra-articular corticosteroid injection under intra-articular anesthesia in pediatric patients.
**Methods**
This prospective study included 57 patients undergoing 83 ultrasound-guided intra-articular injections with triamcinolone hexacetonide under intra-articular anesthesia with 2% lidocaine. Pain and immediate well-being were assessed using a visual analog scale (VAS). Clinical response was defined as absence of arthritis at 60 days. Complications included subcutaneous atrophy, hypopigmentation, infection, and crystal deposition. Mixed models were applied.
**Results**
Median pain VAS was 4.0 (IQR 2.0–5.0), while well-being VAS was 2.0 (IQR 0.0–3.0), demonstrating favorable patient-reported experience. A good clinical response was achieved in 68.7% of injections. Complications were limited to minor local effects, such as cutaneous atrophy (13.3%) and hypopigmentation (12.0%), with no cases of infection or crystal deposition. No significant differences were observed in pain intensity ($p$=0.207), well-being ($p$=0.330), or clinical response ($p$=0.966) across age groups or joint groups. Previous injection experience was not associated with pain, well-being, or clinical response.
**Conclusion**
Ultrasound-guided intra-articular injection performed under intra-articular anesthesia provides not only effective disease control but also a positive patient experience. Protocols combining ultrasound guidance and intra-articular anesthesia may improve patient experience and standardize the procedure, with consistent outcomes regardless of age group or joint involved.
**Disclosure of interest**
None declared.

P023
**Investigation of physical activity attitudes in children and adolescents with juvenile idiopathic arthritis: a comparative study; pilot study**
S. S. Gumus[1,*], S. B. Gun[1], E. G. Kabul[2], B. B. Calık[1], G. Oguz[3], S. Turkucar[3]
[1]Faculty of Physiotherapy and Rehabilitation, Pamukkale University, Denizli; [2]Physiotherapy and Rehabilitation, Usak University, Usak; [3]Department of Pediatric Rheumatology, Pamukkale University Faculty of Medicine, Denizli, Türkiye
**Correspondence:** S. S. Gumus
*Pediatric Rheumatology 2026*, **24(S1):**P023

**Introduction**
Participation in physical activity among children and adolescents with Juvenile Idiopathic Arthritis (JIA) is negatively affected not only by physical limitations such as pain, morning stiffness, and reduced joint range of motion, but also by psychosocial factors such as kinesiophobia, parental anxiety, and reduced self-efficacy in the child (1). They engage in less physical activity compared to healthy children and need to be encouraged to participate in physical activity tailored to their symptoms (2).

**Objectives**
This study was designed to examine the attitudes toward physical activity among children and adolescents with JIA.
**Methods**
This study included 10 children/adolescents diagnosed with JIA (7 girls, 3 boys; mean age 11.8 ± 2.65 years) who met the ILAR diagnostic criteria, and 10 healthy children/adolescents (7 girls, 3 boys; mean age 11.9 ± 1.79 years). After recording the children's and adolescents' demographic and disease-related data, their physical activity attitudes were assessed using the Youth Physical Activity Attitudes Scale (YPAAS), their functional levels using the Childhood Health Assessment Questionnaire (CHAQ-DI), and their quality of life using the Pediatric Quality of Life Questionniare (PedsQL). The Mann-Whitney U test was used for statistical analysis.
**Results**
When the groups were compared, there was a significant difference in the positive subscale of the physical activity attitude scale ($p$=0.010), but no significant difference in the negative subscale ($p$=0.284). In the functional assessment results, significant differences were observed in the reaching ($p$=0.030), walking ($p$=0.030), grasping ($p$=0.020), other ($p$=0.013), and total ($p$=0.002) subscales; however, there were no differences between the groups in the dressing, eating, standing up, and hygiene subscales ($p$>0.05). In terms of quality of life results, both groups were similar ($p$>0.05).
**Conclusion**
Based on the results of this study, we observe that children and adolescents with JIA are unable to motivate themselves regarding their attitudes toward physical activity and are also functionally more limited. Consequently, children and adolescents with JIA need motivation to increase their physical activity levels and should be supported in this regard.

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**Disclosure of interest**
None declared.

## P024

**Rapid universal relapse of systemic juvenile idiopathic arthritis following discontinuation of intravenous tocilizumab in a resource-limited setting**

A. A. Abushhaiwia[1,*]
[1]Pediatric Rheumatology, Tripoli Children's Teaching Hospital, University of Tripoli, Tripoli, Libya
**Correspondence:** A. A. Abushhaiwia
*Pediatric Rheumatology 2026*, **24(S1):**P024

**Introduction**
Systemic Juvenile Idiopathic Arthritis (sJIA) is a severe autoinflammatory disease driven by interleukin-6 (IL-6). The IL-6 receptor inhibitor Tocilizumab is effective in inducing remission and enabling corticosteroid withdrawal. In resource-limited settings such as Libya, treatment discontinuation due to drug unavailability is not uncommon. Data on outcomes following cessation remain limited.
**Objectives**
To describe the clinical, laboratory, and therapeutic outcomes following discontinuation of monthly intravenous Tocilizumab in patients with systemic Juvenile Idiopathic Arthritis (sJIA) in a resource-limited setting.
**Methods**
We retrospectively reviewed four patients with sJIA (3 females, 1 male) who achieved complete clinical remission on monthly intravenous tocilizumab with successful corticosteroid withdrawal, and subsequently discontinued therapy due to non-medical reasons. Clinical, laboratory, and treatment outcome data were analyzed. Relapse was defined as recurrence of systemic and/or articular disease requiring therapeutic escalation.
**Results**
All patients (100%) relapsed within one month of discontinuation. Relapse was characterized by fever, evanescent rash, and polyarthritis, accompanied by marked inflammatory activation (ESR up to 100 mm/hr, elevated CRP100-200 mg\dl, and serum ferritin 450–700 ng/mL). All required hospitalization and treatment escalation with intravenous pulse methylprednisolone followed by high-dose oral corticosteroids and methotrexate. Methotrexate was subsequently intensified via subcutaneous dose escalation, and thalidomide was introduced in refractory cases. Reintroduction of biologic therapy was delayed or unavailable, resulting in persistent disease activity and steroid dependence.
**Conclusion**
Discontinuation of monthly intravenous Tocilizumab was associated with rapid and universal relapse despite prior complete remission and steroid withdrawal. The severity and consistency of relapse underscore biologic-dependent disease control in this cohort and highlight critical inequities in sustained access to biologic therapy in resource-limited settings.

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**Disclosure of interest**
None declared.

## P025

**Paediatric onset Still's disease presenting with myositis and features of macrophage activation syndrome**

A. Rehman[1,*], N. Martin[2]
[1]Pediatrics; [2]RHC, Glasgow, UK
**Correspondence:** A. Rehman
*Pediatric Rheumatology 2026*, **24(S1):**P025

**Introduction**
To our knowledge only 2 cases have previously been reported of childhood onset Still's disease with myositis at presentation. Atypical presentations may delay appropriate treatment.
**Objectives**
To highlight a case of Still's disease in childhood with myositis at presentation.
**Results**
A 9 year old, previously well, Caucasian, non-consanguineous girl presented with a one week history of spiking fevers, headache, blanching rash on trunk, arms and thighs, myalgia and stiffness. She was unwilling to walk and reported pain in her muscles rather than joints. No swollen or restricted joints were found on examination. A septic screen was performed, including LP. No organism was found but she was treated for suspected meningitis with IV antibiotics and dexamethasone. Despite treatment, she deteriorated clinically, unable to mobilise or weight bear with poor oral intake. Inflammatory markers remained elevated with persistent fevers. Ferritin 10,716, WCC 32, AST 75, LDH 509, and CRP 210 were raised. CK was normal and Hb 67 was low. BMA showed no malignancy but macrophages with evidence of haemophagocytosis were identified. MRI thighs showed diffuse, patchy oedema in keeping with myositis. Evanescent rash was felt to be typical of Stills disease and treatment was started with high dose intravenous methylprednisolone, anakinra and a weaning course of prednisolone. She responded quickly with resolution of fever, rash and myalgia as well as normalisation of all blood tests. Steroids were stopped after 8 weeks and 6 months later anakinra was changed from once daily to alternate days with no return of symptoms.
**Discussion**
Myositis is an unusual presenting feature in Stills disease with onset in childhood. To our knowledge only 2 previous cases have been report-

ed. More cases have been reported in adult onset Still's disease but it remains an atypical presentation. Despite recent advances in treatment with IL-1 or IL-6 inhibitors Still's disease continues to have a significant mortality rate and prompt treatment is associated with greater likelihood of successful response. Atypical presenting features may cause unnecessary delay in reaching the correct diagnosis and starting effective treatment.

**Methods**

Retrospective review of patient records.

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**Disclosure of interest**

None declared.

## P026

### Still's disease associated lung disease: experience from a tertiary Italian centre

C. Bracaglia[1,*], M. Pardeo[1], M. Trevisan[1], A. De Matteis[1], I. Caiello[2], L. Menchini[3], M. L. Mennini[3], G. Prencipe[2], F. De Benedetti[1,2]
[1]Division of Rheumatology; [2]Laboratory of Immuno-Rheumatology; [3]Department of Radiology and Bioimaging, IRCCS Ospedale Pediatrico Bambino Gesù, Roma, Italy
**Correspondence:** C. Bracaglia
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**Introduction**

Still's Disease (SD)-associated lung disease (SD-LD) is a severe potentially life-threatening rare condition characterized by peculiar features. The experience on the management of SD-LD patients is limited.

**Objectives**

To describe clinical data and treatment strategies of 9 SD-LD patients followed in a single tertiary paediatric rheumatology centre in Italy.

**Methods**

IL-18 levels were measured by ELISA.

**Results**

Eleven White-Caucasian patients with SD-LD diagnosed between 2017 and 2026 are followed in our division. The median age at SD onset was 1.3 years (range 4 months – 13.4 years) and at LD onset was 2.4 years. None had trisomy 21. Ten out of 11 patients (90%) had history of MAS with recurrent episodes in 7 of them (63%). Eight (73%) patients carried the HLA-DRB1*15 haplotype and 9 (82%) the HLA-DRB1*11. IL-18 levels at LD onset (median 136,559 pg/ml, IQR 68,533-419,89) were significantly elevated ($p$=0.03) compared to levels in 59 SD patients without LD (56,033 pg/ml, IQR 13,828-243,546). Six (54%) out of 11 patients had eosinophils count higher than 1.000 mmc in at least two occasions during disease course. Nine patients were exposed to IL-1 inhibitors before LD diagnosis, 5 to anakinra and 4 to both anakinra and canakinumab. Seven patients had drug reaction to IL-1 or IL-6 inhibitors, 3 to anakinra and 5 to tocilizumab. The 3 patients who reacted to anakinra presented the drug reaction before the onset of LD while the reaction to tocilizumab occurred in 1 patient before the onset of LD and in 3 patients after. After LD diagnosis, 8 patients received intravenous glucocorticoids; IL-1 and IL-6 inhibitors were variably used in addition to other immunomodulatory treatments. All the patients received cyclosporine as T cell immunosuppressant in addition to mycophenolate mofetil in 4 patients and to sirolimus in (1) Targeted therapies were also used in addition to other treatments, ruxolitinib was administrated in 6 patients, emapalumab in 4 patients and arumakimig in 4. The response to treatment was quite heterogeneous. SD-LD improved in 5 patients, 3 on ruxolitinib, 1 on emapalumab and 1 on arumakimig, remained stable in 3 patients and worsened in (2) One patient with the most severe lung involvement, who progressed while on treatment (ruxolitinib, arumakimig and emapalumab), received haematopoietic stem cell transplantation. None of the 11 patients developed LD complications, none required O2 supplementation. Only 1 patient was admitted to ICU for severe infection unrelated to LD. All 11 patients are alive at last follow-up (median follow-up for LD of 2.3 years, range 2 months-8 years) with SD in clinical inactive disease on medication.

**Conclusion**

Patients with SD-LD had highly variable disease course. Treatment strategies are very heterogeneous. Multicentre studies are needed to define management guidelines and to identify the best treatment strategies.

**Disclosure of interest**

C. Bracaglia: None declared, M. Pardeo: None declared, M. Trevisan: None declared, A. De Matteis: None declared, I. Caiello: None declared, L. Menchini: None declared, M. L. Mennini: None declared, G. Prencipe: None declared, F. De Benedetti Grant / Research Support with: Roche, Sobi, Novimmune, Sanofi, Novartis, Elixiron; Regeneron, BMS, Consultant with: Sobi, Novartis, Apollo, Kiniksa.

## P027

### Early biologic therapy, treatment withdrawal, and relapse in systemic juvenile idiopathic arthritis: a single–centre cohort study

F. Jenabi[1,*], A. Leahy[1]
[1]Paediatric Rheumatology, University Hospital Southampton, Southampton, UK
**Correspondence:** F. Jenabi
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**Introduction**

Systemic juvenile idiopathic arthritis (sJIA) is a rare autoinflammatory condition with variable disease course. Biologic therapies have improved outcomes, yet real-world evidence on remission, treatment withdrawal, relapse, and predictors of response remains limited.[1-2]

**Objectives**

To evaluate treatment patterns, time to remission, relapse after treatment withdrawal, and the influence of time to diagnosis, early biologic therapy, methotrexate (MTX) exposure, and withdrawal strategy on clinical outcomes in a single-centre sJIA cohort.

**Methods**

A retrospective cohort study was conducted including all children with confirmed systemic juvenile idiopathic arthritis (sJIA) diagnosed between January 2015 and December 2024 and with available follow–up. Data collected included demographics, inflammatory markers at diagnosis and remission, MTX and biologic exposure, timing of biologic initiation, remission, treatment withdrawal strategy, and relapse. Early biologic therapy was defined as initiation ≤4 months from diagnosis. The primary outcome was time to clinical remission; secondary outcomes included relapse after treatment withdrawal and remission following re–initiation. Exploratory correlations and group comparisons were performed.

**Results**

Twenty patients were included (45% female; median age 9.3 years). Thirteen (65%) received biologic therapy, of whom eight (61.5%) started treatment early. MTX was used in 10/20 (50%). Remission was achieved in 18/20 (90%). Among those achieving remission, 16/18 (88.9%) discontinued all treatment. Relapse occurred in 2/17 (11.8%) after withdrawal, with both patients regaining remission after restarting therapy. Median time to remission was 2.3 months (IQR 1.7–10.1). Early biologic therapy was associated with shorter remission times (median 2.3 vs. 10.2 months; exploratory). Time to diagnosis showed no meaningful association with remission or relapse. MTX–treated patients had slightly lower remission rates (80% vs. 100%) and longer time to remission (median 4.3 vs. 2.0 months), likely because these patients had more severe disease at diagnosis. Median remission duration before treatment cessation was 15 months, with no clear relationship to relapse risk. Relapse occurred across different withdrawal strategies without a consistent pattern.

**Conclusion**

Most children achieved remission rapidly, and relapse after treatment withdrawal was uncommon. Early biologic therapy appeared to be associated with faster remission, whereas time to diagnosis, combined treatment with MTX, remission duration before stopping, and withdrawal strategy did not demonstrate clear influence on relapse. These findings support the early use of biologics and highlight the need for larger studies to guide treatment–withdrawal approaches, identify fac-

tors associated with achieving remission, and explore potential predictors of relapse after treatment withdrawal in sJIA.

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**Disclosure of interest**
None declared.

## P028

### Ferritin induces pro-inflammatory cytokines expression in human monocytes: a potential driver of inflammation in Still's disease


F. Lucioni[1,2,*], G. Rogani[1,3], F. Huijsmans[1,3], A. De Ligt[1], L. Sijbers[1], A. Bodelón De Frutos[1], J. Van Loosdregt[1], B. Vastert[1,3]
[1]Center for Translational Immunology, University Medical Center Utrecht, Utrecht, Netherlands; [2]Pediatric Immuno-Rhematology Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy; [3]Division of Pediatric Rheumatology and Immunology, Wilhelmina Children's Hospital, Utrecht, Netherlands
**Correspondence:** F. Lucioni

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**Introduction**
Serum ferritin levels are elevated in patients with active Still's disease (SD) and correlate with disease activity. Emerging evidence suggests that ferritin may exert immunomodulatory and pro-inflammatory effects, although its exact contribution to disease pathogenesis remains incompletely understood. Recently, macrophage scavenger receptor 1 (MSR1) has been identified as a receptor for extracellular ferritin uptake.
**Objectives**
To investigate whether extracellular ferritin can induce a pro-inflammatory response in human monocytes.
**Methods**
Primary monocytes isolated from healthy donors were stimulated with human ferritin at increasing concentrations for 4 h. Expression of the pro-inflammatory cytokines *IL1B*, *IL6*, *IL8*, and *TNFA* was assessed by quantitative PCR. Moreover, RNA sequencing was performed on monocytes from patients with active SD and inactive SD.
**Results**
Ferritin stimulation induced a clear and dose-dependent increase of *IL1B*, *IL6*, *IL8*, and *TNFA* RNA expression in human monocytes. These findings demonstrate that extracellular ferritin is capable of directly activating monocytes toward a pro-inflammatory phenotype. In parallel, transcriptomic analysis demonstrated increased expression of the ferritin receptor *MSR1* in monocytes from patients with active SD compared with inactive disease.
**Conclusion**
Extracellular ferritin can directly promote pro-inflammatory cytokine expression in human monocytes, supporting a role for ferritin beyond that of a passive biomarker in hyperinflammatory conditions such as SD.
**Disclosure of interest**
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## P029

### Integrated clinical and epigenetic signatures identify response to IL-1B inhibition in pediatric Still's disease


F. Penco[1], G. Cavalca[2], E. Longoni[1], S. Sheffler[1], R. Caorsi[1], S. Palmeri[1,3,*], E. Drago[1,3], A. Bertoni[1], P. Uva[2], G. Fiorito[2], M. Gattorno[1]
[1]UOC Reumatologia e Malattie Auotinfiammatorie; [2]Unità di Bioinformatica Clinica, IRCCS Giannina Gaslini; [3]DINOGMI - Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, Università degli Studi di Genova, Genova, Italy
**Correspondence:** S. Palmeri

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**Introduction**
Still's disease is a heterogeneous autoinflammatory disease driven by innate immune dysregulation, in which IL-1β blockade has improved outcomes. However, many of patients fail to respond, highlighting the need for biomarkers predicting therapeutic response. (1-3) DNA methylation (DNAm)-based biomarkers of biological aging such as epigenetic clocks, recently emerged as promising stratification tools.
**Objectives**
To identify clinical and epigenetic predictors of response to IL-1β-targeted therapy in sJIA patients.
**Methods**
We retrospectively analyzed 84 pediatric Still's disease patients treated with IL-1 inhibitors (Anakinra). A subset of 32 patients underwent integrated clinical and epigenetic profiling at treatment initiation. Clinical response at 12 months was assessed according to clinically inactive disease (CID), based on modified Wallace criteria. Genome-wide DNAm analysis was performed on peripheral blood mononuclear cells (PBMCs). Clinical variables and DNAm-based clocks were evaluated through multivariable regression models adjusted for age and sex.
**Results**
Median disease duration at treatment initiation was 23.5 months (range 2–198). Ten patients started treatment within 6 months from disease onset, 22 after 6 months. Fever was present in all patients at baseline, rash and arthritis occurred in 94% of cases, with a median of 6 active joints (range 0–39). At 12 months, 72% patients achieved complete or partial response to anti IL-1 treatment, with CID in 34%. In univariate analysis, shorter disease duration and lower number of affected joints were associated with higher probability of CID, but the associations were attenuated after correction for age and sex. In contrast, DNAmPhenoAge (4) remained significantly associated with CID even after adjustment for age, sex, and clinical variables, suggesting it captures biological information beyond clinical assessment.
**Conclusion**
Clinical and epigenetic profiling identified DNAmPhenoAge as a potential independent biomarker of response to IL-1β inhibition in sJIA. These findings support biologically distinct response endotypes and biomarker-driven patient stratification in Still's diseases.

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**Disclosure of interest**
None declared.

## P031

### Isn't it ironic? Ferroptosis as a driver of monocyte inflammation and its implications in Still's disease


F. Huijsmans[1,2,*], D. Brookes[2], A. Bodelón[2], G. Rogani[1,2], L. Langevin[3], R. S. M. Yeung[3,4], B. Vastert[1,2], J. V. Loosdregt[2]
[1]Pediatric Rheumatology and Immunology, UMC Utrecht, Wilhelmina Children's Hospital; [2]Center for Translational Immunology, UMC Utrecht, Utrecht, Netherlands; [3]Cell and Systems Biology Research Program, The Hospital for Sick Children Research Institute; [4]Division of Rheumatology, Departments of Pediatrics, Immunology and Institute of Medical Science, The Hospital for Sick Children Research Institute and Toronto University, Toronto, Canada

**Correspondence:** F. Huijsmans
*Pediatric Rheumatology 2026*, **24(S1):**P031

**Introduction**
Still's disease (SD) is an autoinflammatory syndrome marked by profound innate immune dysregulation, yet the mechanisms driving disease pathogenesis remain incompletely understood. Emerging evidence implicates ferroptosis, an immunogenic form of cell death driven by dysregulated iron metabolism and uncontrolled lipid peroxidation, in inflammatory arthritis.
**Objectives**
We aimed to (1) determine whether ferroptosis-associated pathways are activated in patients with SD and (2) investigate how ferroptotic stress influences monocyte activation.
**Methods**
RNA sequencing was performed on monocytes from healthy donors (HD) and patients with SD (onset & remission *n*=7) to assess ferroptosis-associated gene signatures. To investigate the functional consequences of ferroptotic stress, primary human monocytes were treated in vitro with RSL3, a ferroptosis inducer that promotes lipid peroxidation through GPX4 inhibition, with or without the ferroptosis inhibitor Liproxstatin-1. Transcriptomic responses of HD monocytes to RSL3 treatment (*n*=4) were analysed by RNA sequencing. Cytokine production and lipid peroxidation levels of monocytes after RSL3 treatment were quantified by flow cytometry. In parallel, plasma lipid peroxidation levels, measured as malondialdehyde (MDA), were assessed longitudinally in paired samples from SD patients (during active and inactive disease).
**Results**
Monocytes from active SD patients at disease onset demonstrated significant enrichment of the ferroptosis pathway compared with those in remission and HD, as determined by gene set enrichment analysis. This involved increased expression of iron importers, *TFRC* and *ZIP8*, alongside reduced expression of the iron exporter *FPN*, potentially reflecting enhanced iron uptake/accumulation. In addition, these active SD monocytes shared a transcriptional signature with RSL3–treated monocytes, indicating overlap between ferroptosis-induced and SD-associated transcriptional programs. Plasma lipid peroxidation levels (MDA) were significantly elevated during active SD. Functionally, RSL3 induced IL-8 and TNFα production by HD monocytes without causing overt cytotoxicity. This inflammatory response was attenuated by Liproxstatin-1, supporting a ferroptosis-dependent mechanism. Moreover, RSL3 increased lipid peroxidation in monocytes, confirming activation of the ferroptosis pathway.
**Conclusion**
These findings identify ferroptosis as a potential contributor to the pathobiology of active disease in SD. Sublethal ferroptotic stress can drive monocyte activation, supporting a role for ferroptosis beyond terminal cell death. Elucidating the interplay between ferroptosis, inflammatory signalling, and other forms of cell death may uncover novel therapeutic targets for hyperinflammatory syndromes in children.
**Disclosure of interest**
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## P032

### Study of the potential influence of the gut microbiota on paediatric Still's disease, based on animal model

G. Côte[1,*], E. Merlin[1], C. Del'Homme[2], A. Bernalier[2]
[1]Peadiatrics, CHU Estaing; [2]MEDIS, INRAE, CLERMONT FERRAND, France
**Correspondence:** G. Côte
*Pediatric Rheumatology 2026*, **24(S1):**P032

**Introduction**
Systemic juvenile idiopathic arthritis (sJIA), also known as paediatric Still's disease, is an inflammatory disease primarily affecting children aged between one and five years. Both the innate and adaptive immune systems seem to be involved in this disease, but the pathophysiology is not fully understood. Gut microbiota dysbiosis has also been observed in patients with sJIA.
**Objectives**
This study aimed to determine whether gut microbiota can directly modulate systemic juvenile idiopathic arthritis activity.
**Methods**
An in vivo experiment was carried out using the collagen-induced arthritis (CIA) rat model, involving the implantation of human microbiota (from sJIA patients or controls) in germ-free animals.
**Results**
A dysbiosis of the gut microbiota was found in the sJIA patient. Different phenotypes were observed in rats following collagen injections: more severe arthritis and a more pronounced inflammatory response (hepatosplenomegaly, polynucleosis) in rats with control microbiota, and hypereosinophilia and a defective anti-inflammatory response in rats with patient microbiota. Intestinal dysbiosis, notably an overabundance of the genus *Dialister*, as well as an alteration in the production of intestinal metabolites, were observed in rats with patient microbiota.
**Conclusion**
Gut microbiota appears to influence the expression of systemic juvenile idiopathic arthritis (sJIA), affecting the development of arthritis and systemic inflammation, eosinophilia, and the IL-10 anti-inflammatory response.
**Disclosure of interest**
None declared.

## P033

### Early short-course anakinra as first-line therapy combined with glucocorticoids induces sustained drug-free remission in systemic juvenile idiopathic arthritis

K. Chiotopoulou[1], K. Kourtesi[1], S. Prountzos[2], A. Margoni[3], A. Chira[1], L. Fotis[1,*]
[1]Department of Pediatrics, Division of Pediatric Rheumatology, ATTIKON General University Hospital; [2] Department of Radiology, ATTIKON General University Hospital; [3]1st Department of Biochemistry, National and Kapodistrian University of Athens, Athens, Greece
**Correspondence:** L. Fotis
*Pediatric Rheumatology 2026*, **24(S1):**P033

**Introduction**
Systemic juvenile idiopathic arthritis (sJIA) is an autoinflammatory disease in which early interleukin-1 (IL-1) blockade may alter disease trajectory. However, the ability to discontinue treatment after a short therapeutic course remains insufficiently defined.
**Objectives**
To des**c**ribe the clinical phenotype and outcomes of patients with sJIA achieving sustained remission after first-line short-course anakinra combined with glucocorticoids.
**Methods**
We retrospectively analyzed 30 patients with sJIA followed in a single tertiary pediatric rheumatology center. Clinical, laboratory, and treatment data were collected. Patients receiving anakinra as first-line therapy were categorized according to outcome.
**Results**
Anakinra was used as first-line therapy in 18/30 patients (60%). Five patients (28%) achieved sustained drug-free remission following discontinuation of short-course anakinra. All five received concomitant glucocorticoids during the first month, followed by tapering. In this subgroup, median age at diagnosis was 2.05 years (1.22–10.66), median anakinra duration was 4.27 months (3.91–5.85), and median follow-up was 3.95 years (0.39–6.41). At presentation, all patients had fever (100%), while rash and arthritis were present in 4/5 patients (80%). Arthralgia and pharyngitis were present in 3/5 (60%), and lymphadenopathy and abdominal pain in 2/5 (40%). Among patients with failure to maintain remission (*n*=6), treatment failure occurred early, with a median time to failure of 2.45 months (1.68–5.06). In an exploratory comparison, patients achieving sustained remission were younger at diagnosis **(**2.05 [1.22–10.66] vs. 8.36 [2.79–13.14] years**)** and demonstrated higher inflammatory markers, including WBC **(**22.24 [7.33–27.63] vs. 15.70 [8.97–27.40] $\times 10^3/\mu L$**)**, CRP **(**99.2 [45.7–161.9] vs. 68.5 [22.4–137] mg/L**)**, ESR **(**90 [66–112] vs. 55.5 [2–156] mm/h**)**, and ferritin **(**1058.9 [236–1955] vs. 264 [59.8–4457] µg/L**)**. Clinical features, including fever, rash, and arthritis, were similarly frequent between groups and did not clearly distinguish responders from non-responders.

**Conclusion**
A subset of patients with sJIA treated with early short-course anakinra combined with glucocorticoids achieved sustained drug-free remission, while non-responders exhibited early treatment failure. These findings suggest rapid divergence into distinct disease trajectories and support the existence of an early IL-1–responsive phenotype driven primarily by inflammatory burden rather than clinical presentation.

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**Disclosure of interest**
None declared.

## P034

**Inflammatory bowel disease in Still's disease**

M. Trevisan[1,2,*], M. Pardeo[1], A. De Matteis[1], I. Caiello[3], G. Prencipe[3], F. De Benedetti[1,3], C. Bracaglia[1]
[1]Division of Rheumatology, IRCCS Ospedale Pediatrico Bambino Gesù; [2]Department of Systems Medicine, University of Rome "Tor Vergata"; [3]Laboratory of Immuno-Rheumatology, IRCCS Ospedale Pediatrico Bambino Gesù, Rome, Italy
**Correspondence:** M. Trevisan
*Pediatric Rheumatology 2026*, **24(S1):**P034

**Introduction**
Still's disease (SD) is a rare systemic inflammatory condition usually characterized by daily fever, rash, and arthritis. Gastrointestinal (GI) involvement and inflammatory bowel disease (IBD) are uncommon and poorly described, although they occur.
**Objectives**
To describe clinical, laboratory, and endoscopic features of patients with SD-associated IBD in a single tertiary pediatric rheumatology center.
**Methods**
We retrospectively analyzed patients with SD who developed GI involvement confirmed by endoscopy and histological examination. Clinical and laboratory data were collected at SD onset, at IBD diagnosis, and 12 months later (T12).
**Results**
Six patients were enrolled (2 females). Median age at SD onset was 7.5 years (IQR 0.4-14.8), and median time from SD diagnosis to GI onset was 2.3 years (1.1-5.6). Only one patient showed GI symptoms at disease onset. All patients carried HLA-DRB1*11, and 50% also carried HLA-DRB1*15. Five out of six patients had a refractory disease with a history of macrophage activation syndrome (MAS), and two patients developed lung disease (LD). At SD onset, no bowel thickening or elevated fecal calprotectin was observed. At IBD diagnosis, only two patients showed concomitant SD flare, including one with MAS. GI manifestations included hepatomegaly (6/6), without significant liver enzyme elevation, diarrhea (4/6), and weight loss (3/6). Laboratory findings showed increased inflammatory markers (median CRP 6.3 mg/dL, IQR 1.2-8.6), hypergammaglobulinemia (median IgG 14 g/L, IQR 11,5-18,4), and elevated fecal calprotectin (median 371 mg/kg, IQR 238-1,557). IL-18 and CXCL9 levels were markedly elevated, without significant differences compared with SD onset. All patients showed evidence of bowel thickening on ultrasound. The endoscopy revealed colitis in all patients and ileitis in 3. Histology mainly showed non-specific mucosal inflammation, with an ulcerative colitis-like pattern in 50% and a Crohn-like pattern in one case. At IBD diagnosis, 2 patients were on anakinra, one on canakinumab, and one on arumakimig. 5/6 patients continued interleukin-1 inhibitors, while 3 added Janus kinase inhibitors and 5 received topical mesalazine. Two patients underwent short courses of intravenous vedolizumab with limited benefit. At T12, 5/6 patients achieved a complete mucosal remission with normal calprotectin values. One patient had persistent asymptomatic pancolitis despite anakinra and upadacitinib therapy.
**Conclusion**
GI involvement in SD is a rare inflammatory complication, unrelated to disease activity, and characterized by non-specific histological findings. This complication should be considered in patients with unexplained elevation of the inflammatory markers or weight loss. Most patients achieve clinical and mucosal remission with combined immunomodulatory therapy.

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**Disclosure of interest**
M. Trevisan: None declared, M. Pardeo Consultant with: Sobi, Novartis, A. De Matteis: None declared, I. Caiello: None declared, G. Prencipe: None declared, F. De Benedetti Consultant with: Abbvie, SOBI, Novimmune, Novartis, Roche, Pfizer, C. Bracaglia Consultant with: Sobi, Novartis.

## P036

**Difficult-to-treat Still's disease with refractory arthritis requiring dual biologic therapy: serum calprotectin as a biomarker under IL-6 blockade**

R. Craveiro Costa[1], E. Bandeira[2], I. Madureira[1,*], C. Henriques[1], M. Conde[1]
[1]Unidade de Reumatologia Pediátrica; [2]Serviço de Radiologia, Hospital de Dona Estefânia, Unidade Local de Saúde de São José, Lisboa, Portugal
**Correspondence:** I. Madureira
*Pediatric Rheumatology 2026*, **24(S1):**P036

**Introduction**
Persistent chronic refractory arthritis despite multiple targeted therapies in pediatric-onset Still's disease remains a major challenge. Monitoring disease activity under IL-6 blockade is difficult because conventional inflammatory biomarkers may be suppressed.
**Objectives**
To describe the case of a child with pediatric-onset Still's disease and persistent destructive arthritis despite dual biologic therapy (bDMARDs) and methotrexate (MTX), highlighting the role of serum calprotectin in monitoring recurrent articular inflammation under tocilizumab (TCZ).
**Methods**
Retrospective review of clinical, laboratory and imaging data from a child with early-onset Still's disease complicated by macrophage activation syndrome (MAS) and evolving refractory polyarthritis.
**Results**
An 11-month-old girl presented with prolonged fever, evanescent rash, polyarthritis with cervical involvement and MAS. Extensive infectious, malignant and genetic investigation, including an autoinflammatory panel, was unrevealing. MAS responded to methylprednisolone pulses and anakinra, but polyarthritis developed during steroid taper despite escalation of anakinra up to 8 mg/kg/day. TCZ, started at 16 months and combined with subcutaneous MTX, controlled arthritis and enabled steroid withdrawal for ~1 year. She later relapsed with polyarthritis and was switched to canakinumab, which failed despite escalation to 6 mg/kg every 4 weeks, with recurrence of systemic symptoms. Reintroduction of TCZ with methylprednisolone pulses controlled systemic inflammation, but recurrent polyarthritis prevented steroid withdrawal. Further strategies included JAK inhibition added to TCZ and MTX and repeated synovectomies with transient benefit. Due to persistent arthritis despite multiple targeted therapies, dual biologic therapy with etanercept added to TCZ and MTX was initiated, resulting in partial improvement and lower serum calprotectin levels, although residual active arthritis persisted, requiring low-dose prednisolone. Under TCZ, C-reactive protein, erythrocyte sedimentation rate and serum amyloid A remained normal despite active arthritis, whereas serum calprotectin better reflected disease activity. Despite prolonged immunosuppression and structural damage, including bilateral humeral head geodes, the patient maintained good functional status.
**Conclusions**
This case highlights the therapeutic and monitoring challenges of difficult-to-treat chronic arthritis in pediatric-onset Still's disease. Active arthritis may persist despite targeted therapies and apparent biochem-

ical control under IL-6 blockade. Serum calprotectin may better reflect articular inflammation than conventional inflammatory biomarkers in this setting. Dual bDMARD therapy may represent a rescue strategy in selected severe refractory cases, underscoring the need for novel therapeutic approaches.

**Disclosure of interest**
None declared.

## P037

### Still's disease in a pediatric heart transplant recipient: diagnostic and therapeutic challenges

S. Yücel[1,*], N. Gürbüz[1], G. Kaymak[1], E. Gülyiğit[1], T. Çetin Arıkan[1], A. H. Kamiloğlu[1], G. Avcı Demirciler[1], G. Kayan Kaşıkçı[2], Z. Ülger Tutar[3], G. Aksu[1]
[1]Department of Pediatric Rheumatology, Ege University Faculty of Medicine, Izmir; [2]Department of Pediatric Cardiology, Kayseri City Hospital, Kayseri; [3]Department of Pediatric Cardiology, Ege University Faculty of Medicine, Izmir, Türkiye
**Correspondence:** S. Yücel
*Pediatric Rheumatology 2026*, **24(S1):**P037

**Introduction**
Still's disease is a systemic autoinflammatory disease characterized by prominent systemic features due to dysregulation of innate immunity.

**Objectives**
We present a pediatric patient with Still's disease in whom the diagnosis was delayed because of severe cardiac disease and post-transplant immunosuppression.

**Methods**
Clinical course, laboratory findings, treatment response, and follow-up were reviewed retrospectively.

**Results**
We report the case of a 14-year-old girl who was diagnosed with heart failure due to Carvajal syndrome. In October 2022, she was admitted due to persistent fever, arthralgia, pericardial effusion and elevated inflammatory markers. This clinical table was first attributed to thyroid storm due to amiodarone-induced thyrotoxicosis and severe osteoporosis. She had orthotopic heart transplantation for end-stage heart failure in March 2023. She continued to receive tacrolimus and everolimus after the transplant. During steroid tapering after the surgery, clinical symptoms persisted. A thorough evaluation by our team discovered prolonged fever, evanescent rash in association with fever spikes, pericardial effusion, arthralgia, hepatosplenomegaly, and elevated inflammatory markers. The patient fulfilled clinical criteria for Still's disease. In October 2024, Anakinra was started. Distinct clinical and laboratory responses occurred within days of starting treatment, with complete pain resolution. Dose tapering was followed by a disease flare marked by bilateral ankle arthritis and morning stiffness, and hyperferritinemia. In June 2025, anakinra was stopped, and canakinumab was started. Since that time, the patient has remained clinically asymptomatic, with normalization of inflammatory markers and no infectious or transplant-related complications, and is currently being followed closely by both the cardiology and rheumatology teams. (see Table 1)

**Table 1 (Abstract P037)** Summary of clinical course and treatment timeline

| Date / Period | Key Clinical Events | Laboratory Findings | Treatment / Intervention | Outcome |
|---|---|---|---|---|
| Mar 2023 | Orthotopic heart transplantation | - | Tacrolimus + everolimus maintenance | Stable graft |
| Oct 2024 | Reevaluation → Still's disease diagnosed | CRP 39 mg/L, ESR 36 mm/h, ferritin 541 ng/mL | Anakinra | Rapid clinical improvement |
| Jun 2025 | Persistent arthritis, hyperferritinemia (1500 ng/mL) | - | Switched to canakinumab (150 mg q3w) | Sustained remission |

**Conclusion**
This case shows the diagnostic difficulty of Still's disease in a pediatric cardiac transplant recipient. Persistent systemic inflammation despite plausible alternative explanations should lead to repeated rheumatologic assessment. In this patient, IL-1 blockade was associated with clinical remission and was not followed by infectious or transplant-related complications.

**Consent to publish**
Written informed consent for publication was obtained from the patient's legal guardian.

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**Disclosure of interest**
None declared.

## P038

### Bispecific ANTI–IL-1B/IL-18 antibody for the treatment of refractory macrophage activation syndrome in a child with systemic juvenile idiopathic arthritis

A. Villarejo Perez[1,*], R. Gonzalez Preciado[1], E. Enriquez Merayo[1], N. Salmon Rodriguez[2], L. I. Gonzalez Granado[3], J. De Inocencio[1]
[1]Pediatric Rheumatology Unit; [2]General Pediatrics Unit; [3]Immunodeficiencies Unit, University Hospital 12 De Octubre, Madrid, Spain
**Correspondence:** A. Villarejo Perez
*Pediatric Rheumatology 2026*, **24(S1):**P038

**Introduction**
Macrophage activation syndrome (MAS) is a life-threatening complication of juvenile onset Still disease in which interleukin-18 (IL-18) plays a pathogenic role. MAS825 is a bispecific monoclonal antibody targeting both interleukin-1β (IL-1β) and IL-18. We report a patient with Still disease and refractory MAS successfully treated with MAS825.

**Objectives**
To describe the successful use of MAS825, a bispecific anti-IL-1β/IL-18 monoclonal antibody, in a child with systemic juvenile idiopathic arthritis complicated by refractory macrophage activation syndrome.

**Methods**
A 9-year-old boy presented with a 10-day history of fever associated with leukocytosis (26,000/mm$^3$), neutrophilia (94%), elevated inflammatory markers (CRP 33 mg/dL), hyperferritinemia (25,300 ng/mL), thrombocytopenia (88,000/mm$^3$), and coagulopathy (INR 1.6). He was transferred to our Pediatric Intensive Care Unit (PICU) due to acute myopericarditis. On admission, echocardiography revealed a left ventricular ejection fraction of 35% with pericardial effusion, and intravenous immunoglobulins were administered. The patient remained febrile with progressive clinical deterioration, developing two days later a maculopapular rash involving the trunk and extremities, elbow monoarthritis, and polyserositis (pericardial, bilateral pleural effusion, and ascites). Laboratory work-up showed worsening hyperferritinemia (37,600 ng/mL), hypertriglyceridemia (362 mg/dL), elevated transaminases (AST 120 U/L), and hypofibrinogenemia (157 mg/dL). A diagnosis of Still disease complicated by MAS was established, and treatment with intravenous methylprednisolone pulses and intravenous anakinra was initiated, achieving initial clinical improvement. After four days, methylprednisolone was tapered to 1 mg/kg/day; however, the patient developed warm shock requiring PICU transfer. Clinical stabilization was achieved only after additional methylprednisolone pulses (30 mg/kg) and escalation of intravenous anakinra to every 6 h. Tofacitinib was subsequently added. During the following two weeks, several attempts to taper intravenous anakinra resulted in recurrent warm shock episodes, requiring repeated methylprednisolone pulses and re-escalation of intravenous anakinra to every 6 h. Tacrolimus was also introduced without clinical benefit. Cytokine profiling demonstrated markedly elevated IL-18 levels (>100,000 pg/mL) and CXCL9 levels (4,968 µg/mL). Compassionate-use approval for MAS825 was therefore requested. MAS825 (10 mg/kg intravenously every 2 weeks) was initiated after discontinuation of all previous therapies, leading to rapid

clinical improvement after the first dose and normalization of laboratory parameters after the second dose. At 12-month follow-up, the patient remains asymptomatic, and MAS825 dosing has been successfully tapered to every 3 weeks.

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**Disclosure of interest**
None declared.

## P039

**Pediatric-onset Still's disease presenting with bilateral dacryoadenitis: an unusual autoinflammatory presentation**

Z. Torunoglu[*,1], E. Aslan [1], N. Akay [1], M. Yildiz[1], O. Kasapcopur[1]
[1]Division of Pediatric Rheumatology, Department of Pediatrics, Istanbul Unıversity-Cerrahpasa, Cerrahpasa Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** Z. Torunoglu
*Pediatric Rheumatology 2026*, **24(S1):**P039

**Introduction**
Still's disease (SD) is a rare systemic autoinflammatory disorder that includes pediatric-onset Still's disease (POSD) and adult-onset Still's disease forms within the same disease spectrum. It is characterized by fever, evanescent rash, lymphadenopathy, serositis, and hepatosplenomegaly. Because no specific diagnostic test exists, the diagnosis requires exclusion of infectious, malignant, and autoimmune conditions. Although orbital inflammation may occur in immune-mediated diseases, dacryoadenitis is a rare manifestation of Still's disease. In children with lacrimal gland enlargement, alternative diagnoses such as Graves' disease, idiopathic orbital inflammation, sarcoidosis, vasculitis, IgG4-related disease, and infection should be considered.
**Objectives**
We report a case of POSD in a patient with bilateral dacryoadenitis who exhibited resistance to corticosteroid therapy, with the aim of highlighting the importance of early recognition of atypical manifestations of SD to prevent diagnostic delay and optimize management.
**Methods**
A 19-year-old woman with PoSD, diagnosed at age 16, presented with a 2-week history of bilateral painful swelling and erythema of the lateral upper eyelids, accompanied by arthritis of the left wrist and bilateral proximal interphalangeal joints. Despite oral methylprednisolone (2 mg/kg/day), she subsequently developed fever, severe sore throat, and a salmon-colored rash. Laboratory evaluation revealed elevated inflammatory markers and ferritin, while autoimmune serologies were unremarkable. Orbital MRI demonstrated bilateral lacrimal gland enlargement consistent with dacryoadenitis. These findings were attributed to a Still's disease flare. Following discontinuation of Etanercept and initiation of Anakinra (100 mg/day), systemic symptoms resolved within 24 h, and orbital swelling markedly improved within 1 month. The patient remained in clinical remission during corticosteroid tapering. This case highlights dacryoadenitis as a rare and potentially misleading manifestation of PoSD. Because orbital findings may mimic infectious or other inflammatory disorders, recognition of this atypical presentation is essential to avoid diagnostic delay and unnecessary treatment. The prompt response to interleukin-1 blockade further supports the autoinflammatory nature of the disease.

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**Disclosure of interest**
None declared.

## P040

**Enthesitis related arthritis: a tertiary center experience with long-term follow-up**

D. Sarisoy[*,1], F. Aydın[1], P. Ö. Avar Aydın[1], O. Bahçeci[1], E. Erorhan[1], B. Tetik Dinçer[1], Z. B. Özçakar[1]
[1]Pediatric Rheumatology, Ankara University School of Medicine, Ankara, Türkiye
**Correspondence:** D. Sarisoy
*Pediatric Rheumatology 2026*, **24(S1):**P040

**Introduction**
Enthesitis-related arthritis (ERA) is a distinct category of juvenile idiopathic arthritis characterized by a chronic disease course and a potential to develop permenant damage.
**Objectives**
This study aimed to describe long-term clinical outcomes and treatment patterns of ERA patients followed in a tertiary care center.
**Methods**
This retrospective study included patients diagnosed with ERA between January 2008 and January 2025. Clinical and laboratory data and treatment regimens were analyzed and compared between treatment courses before and after 2020.
**Results**
A total of 36 patients were included, with a male predominance (66.7%) and a median age at diagnosis of 10 years. Over a median follow-up of 64 (70.5) months, 19 patients (52.8%) achieved remission off medication criteria at least once. Nineteen patients (52.8%) experienced disease relapses after achieving remission on or off medication. A total of 67 treatment courses were administered to 36 patients. Five courses consisted of non-steroidal anti-inflammatory drugs (NSAIDs) and/or intra-articular corticosteroid injections (IACIs), while 20 consisted of conventional disease-modifying anti-rheumatic drugs (cDMARDs) alone. Forty-two courses included biologic disease-modifying anti-rheumatic drugs (bDMARDs), either as monotherapy or in combination with cDMARDs. Thirty-five treatment courses ended up with a new flare after achieving remission. Eight (22.8%) flares needed a step-up in treatment. Before 2020, bDMARDs were used in 13 courses (39.4%) in contrast to 29 courses (85.3%) seen after 2020 ($p<0.05$). The time taken to achieve remission on medication were similar over time for both time periods. However, treatment courses that ended with a new flare were more common before 2020 ($p=0.001$).
**Conclusion**
Long-term real-life data in patients diagnosed with ERA demonstrates that although remission can be achieved in a substantial proportion, disease flares remain common. Although about one-third of the flares responded to NSAIDs and cDMARDs, most flares required bDMARDs for sustained remission. These findings highlight the need for continious monitoring and optimized treatment strategies to improve long-term outcomes in ERA.
**Disclosure of interest**
None declared.

## P042

**Clinical and magnetic resonance imaging features of spinal involvement in pediatric rheumatologic diseases: a single-center experience**

O. Necipoglu Banak[1,*], A. E. Yildiz[2], O. Basaran[1], Y. Bayindir[1], V. Cam[1], D. Unal[1], H. Ercan Emreol[1], M. O. Erkan[1], H. D. Dara Kar[1], S. Ozkan[1], E. Sag[1], Y. Bilginer[1], S. Ozen[1]
[1]Pediatric Rheumatology; [2]Radiology, Hacettepe University, Ankara, Türkiye
**Correspondence:** O. Necipoglu Banak
*Pediatric Rheumatology 2026*, **24(S1):**PR042

**Introduction**
Spinal involvement in pediatric rheumatologic diseases is rare but clinically significant because overlapping inflammatory, structural, and extra-spinal lesions can complicate both diagnosis and follow-up.
**Objectives**
To describe spinal magnetic resonance imaging (MRI) findings and compare disease-related lesion patterns and their associations with complications.

**Methods**
A retrospective study was conducted of children with rheumatologic diseases who had spinal lesions. Demographic information, disease category, lesion distribution, active inflammatory lesions, chronic structural lesions, complications, extra-spinal musculoskeletal findings, and treatment data were extracted from clinical records and radiologic assessments.
**Results**
A total of 110 patients were included. Disease groups were chronic recurrent multifocal osteomyelitis (CRMO) ($n$=40), enthesitis-related arthritis (ERA) ($n$=19), oligoarticular juvenile idiopathic arthritis (JIA) ($n$=15), ERA/CRMO overlap ($n$=15), polyarticular JIA ($n$=8), synovitis-acne-pustulosis-hyperostosis-osteitis (SAPHO) syndrome ($n$=7), and psoriatic arthritis (PsA) ($n$=6). The interval from primary diagnosis to spinal MRI was longest in polyarticular JIA (median 5.2 years) and near zero in CRMO and SAPHO ($p$=0.025). Thoracic and lumbar regions were most frequently involved. Multifocal disease was common, with contiguous lesions in 30 patients (27.3%) and non-contiguous lesions in 21 (19.1%). Three regional signatures emerged: cervical involvement characterized JIA subtypes (oligoarticular JIA 87%, polyarticular JIA 88%; $p$<0.001), thoracic involvement predominated in osteitis-spectrum diseases (CRMO 78%, SAPHO 100%; $p$<0.001), and lumbar involvement was the hallmark of ERA (89%; $p$=0.003). ERA/CRMO overlap showed pan-spinal involvement across regions (40–73%). Active vertebral body and non-vertebral lesions were detected in 88 (80.0%) and 48 patients (43.6%), respectively. ERA was characterized by facet joint inflammation (68%), CRMO by endplate osteitis (45%), and oligoarticular JIA by atlantoaxial involvement (62%). Disc degeneration was frequent in polyarticular JIA (62%) and PsA (67%). Complications occurred in 73 patients (66.4%), mainly discopathy ($n$=52), fracture ($n$=31), and height loss ($n$=29); all fractures were thoracic. Major associations were malalignment with neural compression, disc degeneration with discopathy, endplate osteitis with height loss, chronic lesions with neural compression, and peripheral osteitis with height loss.
**Conclusion**
In this cohort, spinal involvement in pediatric rheumatic diseases showed heterogeneous MRI patterns. Standardized radiologic classification may improve recognition, follow-up, and treatment assessment in children with suspected inflammatory spinal disease. Spinal involvement in pediatric rheumatologic diseases showed disease-related MRI patterns rather than a uniform distribution.
**Disclosure of interest**
None declared.

## P043

### Comparative cytokine profiles in enthesitis-related arthritis and adult spondyloarthritis

N. Z. Özaslan[1,*], M. Kalçık Unan[2], H. Uzuner[3], G. Dilek[2], N. Şahin[1], A. Karadenizli[3], N. Gökçen[2], K. Nas [2], H. E. Sonmez[1]
[1]Department of Pediatric Rheumatology, Kocaeli University, Kocaeli; [2]Department of Physical Medicine and Rehabilitation, Division of Rheumatology and Immunology, Sakarya University, Sakarya; [3]Department of Medical Microbiology, Molecular Research and Antibody Laboratory, Faculty of Medicine, Kocaeli University, Kocaeli, Türkiye
**Correspondence:** N. Z. Özaslan
*Pediatric Rheumatology 2026*, **24(S1):**P043

**Introduction**
Although enthesitis-related arthritis (ERA) and axial and peripheral spondyloarthritis (SpA) lie on the spondyloarthritides spectrum, they differ in age at onset, patterns of involvement, and putative immunopathology.
**Objectives**
This study aimed to compare circulating cytokine/chemokine profiles in treatment-naïve ERA and adult SpA to identify shared and divergent biological signatures.
**Methods**
In a combined cross-sectional/longitudinal design conducted at a tertiary center (January 2024–January 2025), we evaluated ERA ($n$=30) and adult SpA patients ($n$=30) alongside age-/sex-matched healthy pediatric ($n$=20) and adult ($n$=20) controls. Clinical features, disease activity, and routine laboratory parameters were recorded. Serum TNF-α, IL-17A, IL-22, GM-CSF, CXCL4/PF4, CXCL10, CXCL16, and NRG4 were quantified by ELISA.

**Table 1 (Abstract P043)** Comparison of cytokine profiles between patients and healthy controls

| | Enthesis related arthritis | Adult spondyloarthritis | Healthy pediatric controls | Healthy adult controls | *P* value |
|---|---|---|---|---|---|
| CXCL16, pg/mL | 287 (133-367) | 305 (41-433) | 167 (90-285) | 258 (32-354) | <0.001 |
| **GMCSF, pg/mL** | 38 (15-359) | 65 (27-196) | 26 (16-144) | 28 (15-90) | 0.003 |
| **NRG-4, ng/mL** | 37.34 (10.83-94.06) | 50.23 (4.85-99.22) | 23.29 (8.53-89.71) | 25.19 (15.14-95.05) | <0.001 |
| **PF4, ng/mL** | 38.96 (4.94-73.72) | 36.55 (2.92-62.71) | 11.89 (3.52-44.13) | 2.95 (2.07-17.10) | 0.001 |
| **IL-17A, pg/mL** | 1.35 (0.81-3.01) | 1.21 (0.11-3.12) | 1.12 (0.11-2.13) | 1.35 (0.13-2.82) | 0.101 |
| **TNF-α, pg/mL** | 30.40 (17.42-334.63) | 59.54 (22.18-143.96) | 21.46 (18.65-106.27) | 50.3 (33.29-348.04) | 0.159 |
| **IL-22, pg/mL** | 1.26 (0.62-4.12) | 1.22 (0.72-4.88) | 1.28 (0.18-1.88) | 1.22 (0.14-1.62) | 0.243 |
| **CXCL10, pg/mL** | 24.97 (17.19-272.84) | 25.08 (15.95-51.50) | 29.57 (20.30-66.36) | 22.04 (15.8-186.60) | 0.222 |

CXCL16; C-X-C motif chemokine ligand 16, GMCSF; Granulocyte-Macrophage Colony-Stimulating Factor, NRG-4; Neuregulin-4, PF4; Platelet Factor 4, IL-17A; Interleukin-17A, TNF-α; Tumor Necrosis Factor alpha, IL-22; Interleukin-22, CXCL10; C-X-C motif chemokine ligand 10

**Results**
ERA showed more peripheral arthritis (83.3%) and enthesitis (90%), whereas adult SpA showed more sacroiliitis/axial involvement (93.3%) and longer morning stiffness. Versus controls, both patient groups had elevated CXCL16 ($p$<0.001) and PF4 ($p$=0.001). GM-CSF and NRG4 were increased in both diseases and were higher in adult SpA than ERA (GM-CSF: 65 vs. 38 pg/mL, $p$=0.003; NRG4: 50.23 vs. 37.34 ng/mL, $p$<0.001). TNF-α, IL-17A, IL-22, and CXCL10 showed no significant group differences (see Table 1).
**Conclusion**
These findings provide a rationale for biomarker-informed phenotyping and future stratified treatment approaches; however, single-center sampling and ELISA-only measurements warrant multicenter validation.
**Disclosure of interest**
None declared.

## P045

### Unrecognised chronic nonbacterial osteomyelitis in Paraguay: a retrospective application of the 2025 EULAR/ACR classification criteria in culture-negative paediatric osteomyelitis

A. Laterza[1,*], N. Cabrera[1], J. Segovia[2], J. Mura[1], H. Lezcano[2]
[1]Universidad Catolica 'Nuestra Señora de la Asunción'; [2]Instituto de Prevision Social, Asunción, Paraguay
**Correspondence:** A. Laterza
*Pediatric Rheumatology 2026*, **24(S1):**P045

**Introduction**
Chronic nonbacterial osteomyelitis (CNO) is a rare autoinflammatory bone disease with an estimated incidence of 0.4–2.3/100,000 children/year. It is frequently misclassified as infectious osteomyelitis, leading to unnecessary antibiotic courses and significant diagnostic delay. Few data from Latin America exist, and clinical awareness in the region remains very low.

**Objectives**
To retrospectively identify paediatric patients with probable CNO within a 2-year institutional database of culture-negative osteomyelitis in Paraguay, applying the newly published EULAR/ACR 2025 weighted classification criteria.
**Methods**
We performed a retrospective review of electronic medical records at *Instituto de Previsión Social (IPS)*, Asunción, the largest public tertiary referral centre in Paraguay, searching for all patients under 18 years with a diagnosis of osteomyelitis, negative bone or blood cultures, and available imaging (conventional radiograph and/or MRI) between January 2024 and December 2025. Previously healthy children with no haematological or oncological disease and no sustained clinical response to antibiotic therapy were included. EULAR/ACR 2025 classification criteria (threshold ≥55 points) were applied to each case. Whole-body MRI is unavailable at our centre; imaging pattern was therefore scored conservatively as unifocal (+3 pts). Given the retrospective design, ESR, CRP, haemoglobin and fever were not systematically available; as no patient had documented severe systemic inflammation, normal values were assumed for these domains (+32 pts), consistent with the typically mild or normal inflammatory profile reported in CNO.
**Results**
Of the 28 cases of osteomyelitis during the study period, 16 patients were identified (median age 10.5 years, range 1–16; 56% male) as having negative cultures (blood and secretion cultures). All fulfilled entry criteria. None met exclusion criteria. Five patients achieved a total score ≥55 and were classified as probable CNO. Two additional patients were borderline (score ~49), lacking biopsy data that could have moved their score above the threshold. One infant aged under 1 year scored 0 in the age domain, which prompted evaluation for monogenic autoinflammatory bone diseases including deficiency of IL-1 receptor antagonist (DIRA) and Majeed syndrome — conditions that share clinical overlap with CNO but require distinct management. The median time from the onset of symptoms to the point of suspected diagnosis exceeded 12 months across the entire cohort, which is consistent with international reports on the delay in the diagnosis of CNO.
**Conclusion**
This is the first systematic screening for CNO in Paraguay, involving the collaborative work of radiologists, paediatricians, bacteriologists and rheumatologists. Applying EULAR/ACR 2025 criteria to a retrospective cohort of culture-negative, antibiotic-unresponsive paediatric osteomyelitis revealed probable CNO in nearly one-third of cases. The scoring tool was feasible without whole-body MRI, though its unavailability likely led to underscoring of lesion pattern. These findings underscore the urgent need to raise awareness of CNO and related autoinflammatory bone diseases — including DIRA in infants — among paediatric clinicians in Latin America, and support the establishment of a prospective national registry.

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**Disclosure of interest**
None declared.

## P046

### Unrecognised chronic nonbacterial osteomyelitis in Paraguay: applying the 2025 EULAR/ACR classification criteria

A. M. Laterza[1,2,*], J. Seovia[3], J. Mura[1], H. Lezcano[4], M. Laconich[5], N. Cabrera[6]
[1]Universidad Catolica Nuestra Señora de la Asunción; [2]Pediatrics; [3]Radiology; [4]Pathology; [5]Microbiology; [6]Rheumatology Pediatric, IPS, Asunción, Paraguay
**Correspondence:** A. M. Laterza
*Pediatric Rheumatology 2026*, **24(S1):**P046

**Introduction**
Chronic nonbacterial osteomyelitis (CNO) is a rare autoinflammatory bone disease frequently misclassified as infectious osteomyelitis, leading to unnecessary antibiotic courses and significant diagnostic delay. Few data from Latin America exist, and clinical awareness in the region remains very low.
**Objectives**
To retrospectively identify paediatric patients with probable CNO within a 2-year institutional database of culture-negative osteomyelitis at a Paraguayan tertiary centre, applying the newly published EULAR/ACR 2025 weighted classification criteria.
**Methods**
Retrospective review of electronic medical records at *Instituto de Previsión Social* (IPS), Asunción using the ICD-10 code M86 (osteomyelitis) as the primary search term, selecting all patients under 18 years with negative bone and blood cultures and available imaging between January 2024 and December 2025. Previously healthy children without haematological or oncological disease and with no sustained clinical response to antibiotic therapy were included. EULAR/ACR 2025 classification criteria were applied to each case using available clinical, imaging, and laboratory data. Whole-body MRI is unavailable at our centre; imaging pattern was scored conservatively as unifocal. C-reactive protein was not systematically recorded and was assumed normal given the retrospective design.
**Results**
Out of 28 patients over the study period, 16 had negative cultures (57%) and were included (median age 11 years, range 1–16; 56% male). The remaining 12 had confirmed bacterial isolates and were excluded. Among the included patients, 3 (19%) achieved a score ≥55 and were classified as probable CNO, with scores of 78, 62 and 61 - all characterised by MRI abnormalities, age ≥3 years, absence of fever, and normal or mildly elevated VSG. Eleven patients (69%) were borderline, most within 2–8 points of the threshold; the most common limiting factors were absence of biopsy, unavailability of VSG, and presence of fever. One patient with a VSG of 81 mm/h scored 53 (borderline) but would have classified as CNO had inflammatory markers been lower — a finding that warrants clinical reassessment. One infant aged under 1 year scored 27 points and did not meet the age criterion, prompting to suspect for monogenic autoinflammatory bone diseases.
**Conclusion**
Three of 16 included children fulfilled EULAR/ACR 2025 criteria, and 11 were borderline - suggesting a substantial unrecognised burden in this setting. The scoring tool was applicable without whole-body MRI. These findings underscore the need to raise understanding of CNO and related autoinflammatory bone diseases, and support the implementation of structured multidisciplinary diagnostic protocols at paediatric centres in Paraguay.

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3. Hofmann SR, et al. Curr Osteoporos Rep. 2017;15:542–554.

**Disclosure of interest**
None declared.

## P047

### Diffuse cutaneous sclerosis as part of the phenotype of a type I interferonopathy

A. Capilla Miranda[1,*], A. Villarejo Perez[2], L. Fernandez Silveira[1], M. Camacho Lovillo[1]
[1]Pediatric Infectious diseases, Rheumatology and Immunology, Hospital Universitario Virgen del Rocio, Seville; [2]Pediatric Rheumatology, Hospital 12 de Octubre, Madrid, Spain
**Correspondence:** A. Capilla Miranda
*Pediatric Rheumatology 2026*, **24(S1):**P047

**Introduction**
Type I interferonopathies comprise a heterogeneous group of autoinflammatory disorders characterized by dysregulation of the interferon pathway, leading to systemic inflammation with variable manifestations of autoimmunity and/or immunodeficiency. Mutations in the *ATAD3A* gene, which encodes a mitochondrial membrane protein, have recently been associated with activation of the cGAS–STING pathway and overexpression of type I interferon-related genes. The coexistence of neurological and cardiac manifestations in a patient presenting with

diffuse cutaneous sclerosis should raise suspicion of an underlying autoinflammatory or interferon-mediated disorder rather than a primary autoimmune disease alone. Early genetic testing and assessment of type I interferon signatures are essential for accurate diagnosis and may help guide targeted therapeutic strategies in these patients.

**Objectives**

To describe the case of a pediatric patient with diffuse cutaneous sclerosis associated with an *ATAD3A* mutation, highlighting the importance of considering type I interferonopathies in the differential diagnosis of atypical autoimmune phenotypes.

**Methods**

We report the case of a 5-year-old boy referred to Pediatric Rheumatology because of progressive skin tightening and joint contractures. His past medical history was notable for hospital admission at 2.5 months of age due to status epilepticus, followed by the development of hypotonia and global developmental delay. He had also been diagnosed with hypertrophic cardiomyopathy. Physical examination revealed marked skin induration involving both upper and lower limbs, leading to severe restriction of joint mobility. Extensive diagnostic workup included immunological studies (lymphocyte subsets and immunoglobulin levels), autoantibody screening, pulmonary function testing, and skin biopsy, all of which were unremarkable. Electroneurography demonstrated severe motor and sensory axonal polyneuropathy affecting all four limbs. Genetic analysis identified a pathogenic mutation in *ATAD3A*. The patient was treated with intravenous corticosteroid pulses followed by oral tapering, methotrexate, and subsequent addition of tocilizumab. This therapeutic approach resulted in significant improvement in skin involvement and joint mobility, although substantial functional disability persists.

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**Disclosure of interest**

None declared.

## P048

### Clinical practices in the management of biologic DMARDs for CAPS, MKD, and TRAPS an international cross-sectional survey from the JIR CliPS project

A. L. Cunha[1,*], R. Caorsi[2], I. Elhani[3,4], Y. Butbul Avie[5], M. Delplanque[6], S. La Bella[7], F. Haslak[8], A. Mamutova[9], T. Hinze[10], R. Bourguiba[11], M. K. Cuceoglu[12], K. Pateras[13,14], H. Wittkowski[15], V. Hentgen[16] on behalf of JIR-CliPS Working Group on Autoinflammatory Diseases

[1]Department of Community Medicine, Information and Health Decision Sciences, University of Porto, Porto, Portugal; [2]Rheumatology and Autoinflammatory Diseases, IRCCS Istituto Giannina Gaslini, Genova, Italy; [3]General Pediatrics, Hospital Centre Versailles, Le Chesnay; [4]Internal Medicine, AP-HP, Tenon Hospital, Paris, France; [5]Pediatric Rheumatology, Ruth Rappaport Children's Hospital, Haifa, Israel; [6]Internal Medicine, Sorbonne Université, AP- HP, Centre de référence des maladies autoinflammatoires et des amyloses (CEREMAIA), Paris, France; [7]Department of Medicine and Surgery, University of Milano-Bicocca, Milano, Italy; [8]Department of Pediatric Rheumatology, Istanbul Medeniyet University Faculty of Medicine, Üsküdar, Türkiye; [9]Participant of a Short-Term Scientific Mission at COST Action CA21168, Unaffiliated Researcher, Berlin; [10]Clinic for Paediatric and Adolescent Rheumatology, St. Josef-Stift Hospital, Northwest German Center for Rheumatology, Sendenhorst, Germany; [11]Internal Medicine, Hopital des Forces de sécurité intérieure, La Marsa, Tunisia; [12]16 Pediatric Rheumatology, Konya City Hospital, Konya, Türkiye; [13]Department of Statistics, Athens University of Economics and Business, Athens; [14]Department of Public and One Health, University of Thessaly, Karditsa, Greece; [15]Department of Paediatric Rheumatology and Immunology, University Children's Hospital Muenster, Muenster, Germany; [16]Department of pediatrics, French Reference Center for Autoinflammatory Diseases and Amyloidosis, Versaille, France

**Correspondence:** A. L. Cunha

*Pediatric Rheumatology 2026*, **24(S1):**P048

**Introduction**

Cryopyrin-associated periodic syndromes (CAPS), mevalonate kinase disease (MKD), and tumor necrosis factor (TNF) receptor-associated periodic syndrome (TRAPS) are rare monogenic autoinflammatory diseases for which biological DMARDs (bDMARDs), particularly interleukin (IL)-1 inhibitors, represent the primary treatment. Real-world practices concerning bDMARD use vary considerably across healthcare systems.

**Objectives**

To describe international real-life clinical practices for bDMARD management in CAPS, MKD, and TRAPS.

**Methods**

This cross-sectional study utilized a standardized online questionnaire administered between April and December 2025, within the JIR CliPS project (COST Action CA21168). Analyses included descriptive statistics, Chi-square or Fisher's exact tests.

**Results**

Overall, 61 CAPS, 49 MKD, and 51 TRAPS treating physicians answered the questionnaire from 18 countries. Most were pediatric rheumatologists at university hospitals with ≥10 years of experience. Prior to bDMARDs, corticosteroids during flares (CAPS 60.6%, MKD 87.8%, TRAPS 82.3%) and colchicine (CAPS 42.6%, MKD 51.0%, TRAPS 49.0%) were the most commonly reported treatments. Continuous prophylactic bDMARD treatment with IL-1 inhibitors (anakinra and canakinumab) were preferred by the majority in all three diseases, however, 24% of TRAPS physicians preferred TNF inhibitors as first-line agents. IL-1 treatment was reported as available, reimbursed or not, by 82.0% CAPS, 81.6% MKD, and 80.4% TRAPS respondents. IL-1 inhibitor availability was consistently associated with first-line biologic selection across all three diseases, with restricted access associated with increased use of alternative agents ($p<0.001$ for CAPS; $p=0.007$ for MKD and TRAPS). Discontinuation of bDMARDs was rarely considered for CAPS patients with central nervous system (CNS) involvement (9.8%), rising to 36.1% for CAPS without CNS involvement, and to 44.9% and 41.2% for MKD and TRAPS. (see Table 1)

**Conclusion**

This survey highlights consistent real-world patterns in bDMARD use across physicians treating CAPS, MKD, and TRAPS, with a preference for continuous prophylactic therapy and IL-1 inhibitors. IL-1 inhibitor availability was associated with first-line agent selection, underscoring the influence of healthcare-system factors on treatment decisions.

**Disclosure of interest**

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**Table 1 (Abstract P048)** Main results of the JIR CliPS survey on bDMARD use in CAPS, MKD, and TRAPS

| **Demographics, *n* (%)** | **CAPS (*N*=61)** | **MKD (*N*=49)** | **TRAPS (*N*=51)** |
|---|---|---|---|
| Paediatric rheumatologist | 47 (77.0) | 39 (79.6) | 38 (74.5) |
| ≥10 years caring for monogenic autoinflammatory diseases | 29 (47.5) | 26 (53.1) | 24 (47.1) |
| **Treatment strategies, n (%)** | **CAPS (*N*=60)** | **MKD (*N*=49)** | **TRAPS (*N*=50)** |
| Prophylactic maintenance (Continuous)On-demand therapy (during flares only) | 52 (85.2)8 (13.1) | 39 (79.6)10 (20.4) | 41 (80.4)9 (17.6) |
| First-line bDMARD: anti-IL-1 anti-TNF anti-IL6 | 55 (91.7) 2 (3.3) 3 (5.0) | 43 (87.8)6 (12.2)0 (0.0) | 38 (76)12 (24) 0 (0.0) |
| Considered stopping bDMARDs in controlled disease | CNS/SNHL: 6 (9.8); without CNS/SNHL: 22 (36.1) | 22 (44.9) | 21 (41.2) |

CNS/SNHL: CNS involvement/sensorineural hearing loss

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## P050

### Chronic recurrent multifocal osteomyelitis in a resource-limited setting: diagnostic challenges in a three-patient case series

A. A. Abushhaiwia[1,*], A. M. Nuwayji[2]
[1]Pediatric Rheumatology, Tripoli Children's Teaching Hospital, Faculty of Medicine university of Tripoli; [2]Pediatric Rheumatology, Tripoli Children's Teaching Hospital, Tripoli, Libya
**Correspondence:** A. A. Abushhaiwia
*Pediatric Rheumatology 2026*, **24(S1):**P050

**Introduction**
Chronic Recurrent Multifocal Osteomyelitis (CRMO) is a rare autoinflammatory bone disorder characterized by sterile, recurrent, and multifocal osteitis. Whole-body MRI is the gold standard for detecting subclinical lesions, but its availability is limited in resource-constrained settings, leading to diagnostic delays and mismanagement.
**Objectives**
To evaluate the clinical presentation, diagnostic process, and treatment outcomes of pediatric patients with Chronic Recurrent Multifocal Osteomyelitis, with a focus on the impact of limited access to Whole-body MRI on diagnostic delay.
**Methods**
We conducted a retrospective case series of three pediatric patients presenting with recurrent bone pain and swelling suggestive of Chronic Recurrent Multifocal Osteomyelitis. Clinical data, laboratory findings, imaging studies, and histopathological results were reviewed. Diagnostic evaluation included inflammatory markers, microbiological testing, and targeted imaging using Magnetic Resonance Imaging. Infectious and malignant etiologies were excluded through appropriate investigations, including biopsy when indicated. Due to limited access to Whole-body MRI in Tripoli, some patients underwent imaging abroad. Treatment approaches and clinical outcomes were analyzed.
**Results**
All patients were male (ages 3–18 years) with symptom duration ranging from 18 months to 4 years. Two patients had elevated inflammatory markers. MRI demonstrated multifocal inflammatory bone lesions. One patient was initially misdiagnosed and treated for Bone Tuberculosis without improvement. Due to the lack of advanced imaging in Tripoli, patients required international referral for further evaluation, contributing to delayed diagnosis. All patients were ultimately diagnosed with CRMO based on clinical, radiological, and histopathological findings. Treatment with nonsteroidal anti-inflammatory drugs, with or without Methotrexate, resulted in favorable clinical outcomes. (see Table 1)

**Table 1 (Abstract P050)** Summary of patients with Chronic Recurrent Multifocal Osteomyelitis

| Variable | Case 1 | Case 2 | Case 3 |
|---|---|---|---|
| Age / Sex | 18 / M | 3 / M | 6 / M |
| Symptom Duration | 4 y | 18 m | 3 y |
| Clinical Features | Recurrent limb pain | Wrist & ankle swelling | Limping, ankle pain |
| Sites Involved | Tibia, ulna | Tibia, humerus, femur | Ankles, femur |
| Key Findings | MRI + biopsy (chronic inflammation) | Multifocal MRI lesions | Misdiagnosed as Bone Tuberculosis |
| Treatment & Outcome | NSAIDs + MTX → improved | NSAIDs → improved | NSAIDs + MTX → improved |

**Conclusion**
CRMO diagnosis in resource-limited settings is challenging due to restricted access to advanced imaging modalities. Increased clinical awareness and adapted diagnostic strategies are essential to avoid misdiagnosis and unnecessary treatments.
**Disclosure of interest**
None declared.

## P051

### Amyloidosis in patients with monogenic IL-1 associated autoinflammatory diseases (FMF, CAPS, TRAPS): experience of one Russian center

E. Fedorov[1], S. Salugina[1], A. Torgashina[1], M. Beketova[1,*]
[1]V.A. Nasonova Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** M. Beketova
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**Introduction**
AA-amyloidosis is the most severe life-threatening complication of monogenic autoinflammatory diseases (mAIDs). The frequency of amyloidosis may depend on the region of residence.
**Objectives**
To evaluate the incidence of AA-amyloidosis in mAIDs, clinical and genetic characteristics in patients (pts) with mAIDs and amyloidosis in Russia.
**Methods**
The study included 47 pts with mAIDs, including FMF 19(40%), CAPS – 18(38%), TRAPS 10(21%) pts. There are 23 male and 24 female pts. The age of the pts ranged from 7 to 51 years; Me 16[12;20] years. FMF pts were diagnosed according to Tel Hashomer Criteria, CAPS and TRAPS – Eurofever/PRINTO criteria. All pts underwent a standard rheumatological examination, Sanger sequencing of the MEFV, NLRP3 and TNFRSF1A genes, and a biopsy with congo-red staining (43 duodenum, 3 kidney, and 1 subcutaneous fat tissue).
**Results**
Amyloidosis was detected in 13 (28%) of 47 pts. Among them there were 8 female and 5 male pts. The age of pts with amyloidosis was from 7 to 51 years, Me 17[13;32] years, without amyloidosis Me 16[12;17] years old. The duration of the disease with amyloidosis is Me 16[12…32] years, without amyloidosis Me 11[8;16] years. Amyloidosis was detected in 3(16%) out of 19 pts with FMF, in 6(33%) out of 18 pts with CAPS, and in 3(30%) out of 10 pts with TRAPS. In the group with amyloidosis, 2 (15%) received colchicine and 4 (31%) - Canakinumab. In the group without amyloidosis, colchicine was given to 14 (41%), Canakinumab - 7 (21%), Adalimumab - 1 (3%), other DMARDS - 3(9%). Among pts with FMF there were the following genotypes: homozygosity p.M694V, compound heterozygosity p.M680I/E148Q, heterozygote p.M694V. All TRAPS pts, who developed amyloidosis, had the p.T79M (p.T50M) genetic variant.
**Conclusion**
Among pts with monogenic IL1-associated mAIDs in Russia, pts with CAPS have the highest risk of developing amyloidosis, while patients with FMF have the lowest risk, possibly as a result of better awareness of FMF among doctors, timely diagnosis and appointment of colchicine therapy. One of the risk factors for developing amyloidosis is a delay in prescribing therapy. Among TRAPS pts, another risk factor is the type of mutation.
**Disclosure of interest**
None declared.

## P052

### Using the diagnostic score for periodic fever on cohort with familial mediterranean fever in real clinical practice

M. Beketova[*,1], S. Salugina[1], E. Fedorov[1], I. Nikishina[1], A. Torgashina[1], E. Zakharova[2], A. Lila[1]
[1]V.A. Nasonova Research Institute of Rheumatology; [2]Research Center of Medical Genetics, Moscow, Russian Federation
**Correspondence:** M. Beketova
*Pediatric Rheumatology 2026*, **24(S1):**P052

**Introduction**
Given the clinical diversity of monogenic autoinflammatory diseases, tools such as diagnostic score for periodic fever should help identify

and refer for the genetic testing. This score was established and validated on a pediatric population, with a mean age at disease onset of 4.3 years. But how effective can it be in real clinical practice?

**Objectives**

to evaluate the effectiveness of the diagnostic score for periodic fever in group with a previously established diagnosis of familial mediterranean fever (FMF).

**Methods**

Patients (pts) with FMF, who were observed in V.A. Nasonova Research Institute of Rheumatology, retrospectively assessed using the diagnostic score for periodic fever (the cut-off – 15% probability to be positive). Median age 11 [5.5–16.5] yrs, 89% children; M:F=1:1,7. Inclusion criteria: established diagnosis of FMF according criteria Eurofever/Printo. Exclusion criteria: absence of periodic fever, presence of additional variants in genes responsible for the development of other mAIDs. The final analysis included 83 pts. All pts underwent genetic analysis.

**Results**

Among 83 pts, a high risk to be positive at the genetic testing (group 1) observed in 54 cases (65%; Me 81.6[63-93.45] % probability to be positive) and low risk (group 2) – in 29 pts (35%; Me 0.7[0-3.3] %). The variant M694V (with other genetic variants or not) was dominant in the both (70% and 69%). There was statistical difference in frequency of occurrence M694V in heterozygous (more often in group 2, $p$=0.014), but not in homozygous or compound heterozygous. Comparing aspects' diagnostic score of groups with high- and low-risk, group 1 was characterized by an earlier onset (Me 1.6[0.925;3] vs. 8[4;10] yrs, $p$<0,001). The age of onset in group 1 did not exceed 5 yrs. Abdominal pain (100% vs. 79%, $p$<0,001) and aggravated family history (74% vs. 41%, $p$=0.004) were also more common in group 1. In contrast, group 2 was associated with a higher incidence of stomatitis (34% vs. 7%, $p$=0.002). There was no statistical difference in other clinical symptoms and laboratory markers.

**Conclusion**

Using the diagnostic score for periodic fever was an effective tool for referral for genetic testing in 65% pts of our cohort. However, focusing only on the diagnostic score to prescribe a genetic testing, 35% pts might be undiagnosed. Supposed, the decisive role of the positive result is the earlier age of onset. Probably, it's necessary to modify the diagnostic score to increase the efficiency of diagnosing FMF (especially in pts with a later onset).

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**Disclosure of interest**

None declared.

## P053

### AA-amyloidosis in a patient with TNFRSF1A-associated autoinflammatory syndrome and carriage of the R92Q(R121Q) variant

E. Fedorov[1], S. Salugina[1], M. Beketova[1,*]
[1]V.A. Nasonova Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** M. Beketova
*Pediatric Rheumatology 2026*, **24(S1):**P053

**Introduction**

TRAPS is an autoinflammatory syndrome associated with a mutation of the TNFRSF1A gene, a frequent and severe complication of which is AA-amyloidosis. The p.R92Q(R121Q) variant is considered to be low-penetrant, which in some carriers may produce an autoinflammatory phenotype, lighter than typical TRAPS, which almost never leads to amyloidosis. However, isolated cases of amyloidosis in patients with this variant have been described.

**Objectives**

To describe a case of AA-amyloidosis in a patient carrying variant p.R92Q(R121Q) of the TNFRSF1A gene.

**Methods**

A girl with recurrent febrile fever (ethnically Russian) was examined at the pediatric department of the V.A. Nasonova Research Institute of Rheumatology. The TNFRSF1A gene was sequenced according to Sanger with the study of exons 2,3,4. A biopsy of the duodenum was performed with congo-red staining.

**Results**

She has been ill since Nov 2017, at the age of 9. The disease manifested itself in prolonged (up to 2 months) episodes of fever up to 40.5 °C, accompanied by an unstable macular rash at fever altitude and an increase of CRP 152 mg/l, ESR 92 mm/h, fibrinogen 5.46 g/ l, leukocytosis 41.8 × 109/l with a neutrophil fraction of 90%. In December 2017, prednisolone (25 mg/day) was prescribed orally with effect and recurrence with dose reduction. She was first admitted to the Institute of Rheumatology in Feb 2017. Echocardiography shows minimal pericardial effusion. A study of the TNFRSF1A gene revealed a variant of p.R92Q(R121Q). The patient's mother had a similar variant without phenotypic manifestations. Since Feb 2018 Canakinumab has been prescribed (150 mg once every 8 weeks) with the disappearance of symptoms and normalization of acute-phase reactants. Prednisolone has been discontinued. There were no fever relapses, CRP, ESR are persistently normal, and in 2022 the interval between Canakinumab injections was increased to 12 weeks. In March 2026, 9 years after the onset of the disease, a duodenal biopsy was performed, revealing amyloid deposits. Protein in daily urine was 0.19 g/l. There were no symptoms, acute phase reactants, blood creatinine and urea were normal.

**Conclusion**

Patients with an autoinflammatory phenotype, carriers of variant p.R92Q(R121Q) of the TNFRSF1A gene, may develop amyloidosis, even despite successful treatment with an IL1- inhibitor. The doctor should remain alert to the risk of amyloidosis in such patients.

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**Disclosure of interest**

None declared.

## P055

### Liver, spleen, kidney, and parotid gland stiffness by 2D shear wave elastography in paediatric patients with familial mediterranean fever during attack and remission periods

B. Acun Sayılı[1,*], I. Yalçınkaya[2], Ç. Yıldız[1], I. Akdulum[2], D. Gezgin Yıldırım[1], Ö. Boyunağa[2], S. A. Bakkaloğlu[1]
[1]Department of Pediatric Rheumatology; [2]Department of Radiology, Gazi University Faculty of Medicine, Ankara, Türkiye
**Correspondence:** B. Acun Sayılı
*Pediatric Rheumatology 2026*, **24(S1):**P055

**Introduction**

Familial Mediterranean fever (FMF) is a monogenic autoinflammatory disease caused by MEFV gene mutations that activate the pyrin inflammasome and drive IL-1β-mediated inflammation. Despite colchicine-based treatment, subclinical inflammation persists during remission, potentially causing tissue stiffness changes undetectable by conventional imaging. Two-dimensional shear wave elastography (2D-SWE), a non-invasive technique quantifying tissue stiffness, may bridge this diagnostic gap.

**Objectives**

The aim of this study is to evaluate tissue elasticity properties of the liver, spleen, kidneys, and parotid glands using 2D-SWE in paediatric patients with FMF during both attack and remission periods, and to compare these findings with healthy controls.

**Methods**

In this prospective study, 113 pediatric FMF patients (78 in remission and 35 in inflammatory attack) and 50 healthy controls were included. Liver, spleen, kidney, and parotid gland stiffness were measured using

2D-SWE by a blinded radiologist. Disease activity was assessed with AIDAI and Pras scores.

**Results**

The 2D-SWE values were significantly higher in FMF patients than in controls, particularly in the liver and spleen during both attack and remission periods ($p$<0.05, all). Spleen 2D-SWE demonstrated the highest diagnostic performance for identifying inflammatory attack (AUC: 0.851; 95% CI: 0.761–0.942; $p$<0.001), with a cut-off of 7.5 kPa (85% sensitivity, 55–60% specificity). Liver and parotid 2D-SWE showed moderate discriminative ability, while renal 2D-SWE had limited clinical relevance. Parotid 2D-SWE correlated with disease duration. No statistically significant differences were found in 2D-SWE values of the liver, spleen, kidneys, and parotid glands according to *MEFV* mutation.

**Conclusion**

Persistently elevated liver and spleen stiffness, even during remission, may suggest ongoing subclinical inflammation in FMF. Among evaluated organs, spleen 2D-SWE appears to be the most informative marker of disease severity. 2D-SWE may offer a noninvasive approach for detecting organ involvement, though its clinical utility remains to be validated in larger studies.

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**Disclosure of interest**

None declared.

## P056

### Hematopoietic stem cell transplantation for pediatric adenosine deaminase 2 deficiency: first case from UAE with complete ADA2 enzyme normalization

B. Abu Saleh[1,*]
[1]Pediatric Rheumatology, SSMC, Abu Dhabi, UAE
**Correspondence:** B. Abu Saleh
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**Introduction**

Deficiency of adenosine deaminase 2 (DADA2) is a rare autosomal recessive autoinflammatory disorder caused by biallelic mutations in the ADA2 gene, presenting with a spectrum of vasculopathy, immunodeficiency, and bone marrow failure. The disease is highly heterogeneous, with variable severity even among individuals with identical mutations. First-line therapy for vasculitic and inflammatory manifestations is tumor necrosis factor inhibitor (TNFi) treatment, but hematologic features such as bone marrow failure and refractory cytopenias are largely unresponsive to TNFi and glucocorticoids. Allogeneic hematopoietic stem cell transplantation (HSCT) is recognized as a definitive curative option for patients with severe hematological involvement, immunodeficiency, or refractory disease, as established by international consensus and cohort studies.[1][2][3][4][5].

**Objectives**

This report describes a 12-year-old male from Gaza with genetically confirmed ADA2 deficiency, complicated by severe neutropenia, recurrent fevers, vasculitic and musculoskeletal manifestations, and invasive fungal infection. The patient underwent allogeneic HSCT from an HLA-matched sibling donor in Abu Dhabi, United Arab Emirates. The peri-transplant course was notable for prolonged hospitalization, persistent cytopenias, and infectious complications, managed with immunosuppressive and antimicrobial therapies. Post-transplant, the patient demonstrated marked clinical improvement, including resolution of inflammatory symptoms, restoration of independent ambulation, and normalization of ADA2 enzyme activity. At eight months post-HSCT, there was no evidence of recurrent vasculitic or infectious events, and musculoskeletal recovery was ongoing with physiotherapy.

**Methods**

This case represents the first documented instance of successful HSCT for DADA2 in a patient from Gaza, reported from Abu Dhabi, with complete biochemical correction and reversal of clinical manifestations. The outcome aligns with published data showing normalization of ADA2 enzyme activity and resolution of hematological, immunological, and vascular phenotypes following HSCT. These findings reinforce the role of HSCT as a curative therapy for DADA2 in patients with severe disease, and highlight the importance of early recognition and multidisciplinary management in optimizing outcomes.

**Disclosure of interest**

None declared.

## P057

### Bridging pediatric and adult autoinflammatory bone disease: a systematic literature review of CNO and SAPHO across age groups

C. Matucci-Cerinic[1,*], M. Neri[2], S. M. Orsi[1], R. De Luca[3], J. Schoones[4], G. Viglizzo[5], R. Caorsi[1], A. Alunno[6], S. Bellando-Randone[3], M. Gattorno[1]
[1]Rheumatology and Autoinflammatory Diseases, IRCCS Istituto Giannina Gaslini, Genova; [2]IRCCS Azienda Ospedaliero-Universitaria Bologna, Bologna; [3]Division of Rheumatology, Scleroderma Unit, Department of Experimental and Clinical Medicine, AOU Careggi, University of Florence, Firenze, Italy; [4]Directorate of Research Policy (formerly Walaeus Library), Leiden University Medical Center, Leiden, Netherlands; [5]Dermatology, IRCCS Istituto Giannina Gaslini, Genova; [6]Internal Medicine and Nephrology Division, Department of Life, Health and Environmental Sciences, University of L'Aquila, L'Aquila, Italy
**Correspondence:** C. Matucci-Cerinic
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**Introduction**

Inflammatory bone diseases are a heterogeneous group of disorders characterized by sterile bone inflammation. Among these, Chronic non-bacterial osteomyelitis (CNO) and synovitis–acne–pustulosis–hyperostosis–osteitis (SAPHO) syndrome are characterized by sterile, multifocal bone lesions and frequent association with skin manifestations (acne, psoriasis and palmo-plantar pustulosis (PPP)) and inflammatory bowel diseases (IBD).

**Objectives**

To systematically review and critically appraise the available literature on SAPHO and CNO in children and adults, in order to highlight clinical and radiological differences and to explore age- and sex-related phenotypic clusters.

**Methods**

A systematic literature search was conducted across PubMed, Embase, Web of Science, Cochrane Library, Emcare, Academic Search Premier, and Google Scholar from inception to May 8 2025, following PRISMA 2020 guidelines. Clinical trials, observational studies and case series including at least 3 cases of CNO/SAPHO patients of any age. Data were extracted independently by two reviewers and synthesized descriptively. Frequencies of clinical and serological features of adults and children were compared with the Chi square test. When reported in the manuscripts, single-patient data were extracted to perform binary logistic regression analysis and are reported as odds ratio (95% confidence intervals).

**Results**

A total of 4,779 papers were retrieved. After title/abstract screening and duplicate removal, 293 articles underwent full-text review, of which 155 met the inclusion criteria, including 1,801 pediatric and 2,612 adult patients. Most children were reported as having CNO, whereas in adults SAPHO was the predominant diagnosis. Both in the pediatric and the adult cohort, there was a female predominance (F: M 2:1 children, 3:1 adults). Skin manifestations were significantly more frequent in adults (72vs21%, $p$<0.00001), with PPP being the most common manifestation ($p$<0.00001). Both in children and adults, acne resulted more frequent in males ($p$<0.001), while PPP in females ($p$<0.005). Axial involvement (anterior chest wall, spine and sacroiliac joints) was significantly more frequent in the adult population ($p$<0.00001), whereas peripheral (long bones, hands, feet) and pelvic involvement were more frequent in children ($p$<0.00001). Individual patients' data were available for 500 patients, and were used to perform a logistic regression analysis that demonstrated distinct age-related disease patterns. Adult patients were predominantly characterized by axial skeletal involvement (ante-

rior chest wall, sternum, ribs, spine), whereas pediatric patients more frequently showed peripheral involvement (long bones, hands/feet).

**Conclusion**

This is the first systematic literature review to comprehensively compare inflammatory bone osteitis across ages, integrating data on both CNO and SAPHO syndrome in both children and adults. Although these conditions share core features of sterile, multifocal bone inflammation, distinct age- and sex-related patterns emerge. Pediatric disease is characterized by predominant peripheral skeletal involvement, while the adult phenotype shows a clear predominance of axial involvement and skin manifestations, particularly PPP. Overall, these findings support the hypothesis of a continuous clinical spectrum of autoinflammatory bone disease, with phenotypic expression influenced by age and sex, rather than strictly separate disease entities.

**Disclosure of interest**

None declared.

## P059

### Childhood-onset interstitial lung disease as the sole manifestation of STING-associated vasculopathy of infancy (SAVI): an opportunity for precision therapy

P. Kaur[1,*], S. Chandola[2], A. Kurup[1], K. Jat[1], M. Jana[2], N. Bagri[1]
[1]Pediatrics; [2]Radiodiagnosis & Interventional Radiology, All India Institute of Medical Sciences, New Delhi, New Delhi, India
**Correspondence:** P. Kaur
*Pediatric Rheumatology 2026*, **24(S1):**P059

**Introduction**

A 13-year-old boy presented with gradually progressive breathlessness over five years. Onset was at 8 years of age, with an episode of pneumonia requiring admission for intravenous antibiotics. Thereafter, he had a persistent dry cough, intermittent fever, poor appetite, weight loss, and an episode of hemoptysis, which resolved spontaneously. He was presumptively diagnosed with pulmonary tuberculosis (TB) and started on anti-tubercular therapy empirically (ATT) for six 6 months. His symptoms worsened despite ATT, with distress and hypoxemia requiring low-flow home oxygen. Examination was significant for tachypnea, mild distress, hypoxia on room air (saturation 80%), clubbing, and hepatosplenomegaly.

**Objectives** Chest X-ray revealed hyperinflated lung fields, with emphysematous lung changes and pulmonary arterial hypertension (PAH) on computed tomography chest. Pulmonary function tests showed a mixed pattern with severely reduced single-breath diffusing capacity. A bronchoscopy done previously was unremarkable, but for scarring at the entry of the right upper lobe bronchus. Laboratory investigations revealed anemia (Hb g/dL), absolute lymphopenia for age, elevated serum C-reactive protein (9.3 mg/L), and a positive anti-nuclear antibody (immunofluorescence 1:100, 2+). Considering the diagnosis of childhood-onset interstitial lung disease (ChILD), he was started on metered-dose inhalers (MDIs) of Foracort, oral corticosteroids, and diuretics for PAH. While investigating the aetiology of ChILD, whole exome sequencing (WES) yielded a pathogenic variant in the STING1 gene (c.842G> A), suggestive of STING-Associated Vasculopathy with Onset in Infancy (SAVI). Interestingly, there were no associated cutaneous or systemic symptoms. However, the interferon signatures were markedly elevated with respect to the control. Hence immunomodulation was commenced with intravenous methylprednisolone pulse (30 mg/kg/dose for 3 days) followed by weekly steroid tapering (5 mg per week) over 6 weeks, and tofacitinib (0.4 mg/kg/day). Meanwhile, Anfirolumab was arranged through compassionate access and initiated at 5.5 mg/kg/dose monthly. Tofacitinib was withheld thereafter. The child has received five doses of Anifrolumab, with attending improvement in respiratory parameters, stabilisation of pulmonary function tests, and subjective improvement as per patient global assessment.

**Methods**

Interstitial lung disease (ILD) is a known presentation of autoinflammatory syndromes, being a leading cause of morbidity and mortality. They invariably progress to irreversible, progressive fibrosis culminating in respiratory failure, unless timely treated with therapy targeting downstream cytokine pathways. Autoinflammatory syndromes such as SAVI should be considered as a differential diagnosis during the evaluation of adolescent or childhood-onset interstitial lung disease, even in the absence of family history and concomitant cutaneous or systemic vasculopathic features. Targeted therapies such as JAKi and interferon receptor blockers may salvage the remnant pulmonary function and improve quality of life, as evident in the index case.

**Disclosure of interest**

None declared.

## P060

### An unusual presentation of a rare autoinflammatory condition: SAPHO syndrome

A. Tanatar[1,*]
[1]Pediatric Rheumatology, Marmara University Pendik Training and Research Hospital, Istanbul, Türkiye
**Correspondence:** A. Tanatar
*Pediatric Rheumatology 2026*, **24(S1):**P060

**Introduction**

SAPHO (synovitis, acne, pustulosis, hyperostosis, and osteitis) syndrome is a rare chronic inflammatory entity characterized by bone, joint, and skin involvement. The most prominent feature of SAPHO syndrome is sterile, inflammatory osteitis.

**Objectives**

To report a pediatric patient with SAPHO syndrome mimicking malignancy.

**Methods**

A 15-year-old male presented to the emergency room several times over 15 days with severe back pain and was unable to walk. The patient had night fevers for 2 months and had lost 10 kg. The patient's medical history includes a tonsillo-adenoidectomy at age 8. There is no known family history of rheumatological diseases. He has been using isotretinoin for 2 months for acne vulgaris and is a smoker. His weight was 57 kg (30th percentile), and his height was 173 cm (60th percentile). Physical examination revealed acne fulminans-like papulopustular lesions on the face, trunk, and back. There was severe pain and tenderness, with a limited range of motion in both sacroiliac joints. Pain and swelling were present in the medial third of the right clavicle. Laboratory findings included leukocyte count 10,510/μL, hemoglobin 13.9 g/dL, platelet count 364,000/μL, ESR 22 mm/hr (0–20 mm/hr), and CRP 24 mg/L (0-5 mg/L). Biochemistry parameters were normal. Infectious screenings were negative. ANA, RF, CCP, and HLA-B27 tests were negative. Abdominal ultrasound showed a slightly increased longitudinal length of the liver (approximately 165 mm) and a spleen length of approximately 124 mm, which was at the upper limit of normal. A PA chest X-ray and lumbar vertebra MRI were normal. Sacroiliac MRI showed a significant signal increase on fat-suppressed sequences involving opposite articular surfaces of both sacroiliac joints (acute sacroiliitis). Bone marrow aspiration and biopsy performed to rule out malignancy were normal. The patient was started on ibuprofen for SAPHO. At the 15-day follow-up, a dramatic response to ibuprofen was observed. Sacroiliac pain had significantly decreased but persisted. Clavicle pain had resolved, but the patient complained of pain with pressure in the anterior chest wall, sternum, and ribs. Laboratory investigations revealed a leukocyte count of 8800/μL, hemoglobin of 14 g/dL, platelet count of 275,000/μL, ESR of 4 mm/hr (0–20 mm/hr), and CRP of 13.6 mg/L (0-5 mg/L). Low-dose steroid and subcutaneous methotrexate were started. Complete remission with methotrexate was achieved in the 3rd month. Acute-phase reactants became negative. At the last follow-up, the patient had been in complete remission for a year.

**Conclusion**

In a patient presenting with severe back pain and B symptoms, it is necessary to rule out infections and malignancies first, but early diagnosis and treatment are possible with a thorough history and careful physical examination.

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**Disclosure of interest**

None declared.

P061

**Anakinra therapy in colchicine-resistant familial Mediterranean fever: a real-world pediatric cohort**

B. Sözeri[1,*], N. Kara Çanlıoğlu[1], S. Atamyıldız Uçar[1], E. Tunce[1], H. Öztürk Aydın[1]
[1]Department of Pediatric Rheumatology, Ümraniye Training and Research Hospital, İstanbul, Türkiye
**Correspondence:** B. Sözeri
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**Introduction**
Interleukin-1 blockade has an important role in the management of colchicine-resistant familial Mediterranean fever (FMF). Anakinra is widely used because of its rapid onset of action and short half-life; however, treatment persistence may be affected by recurrent flares, injection-related intolerance, and allergic reactions. Real-world pediatric data regarding long-term anakinra use remain limited.
**Objectives**
To evaluate treatment characteristics, efficacy, flare patterns, allergic reactions, and biologic switching patterns in pediatric FMF patients receiving anakinra therapy.
**Methods**
This retrospective cohort study included 70 pediatric FMF patients treated with anakinra for colchicine-resistant disease. Demographic characteristics, treatment duration, flare features, reasons for treatment discontinuation or biologic switch, and PRAS disease severity scores were evaluated. Clinical response was assessed in patients receiving anakinra for at least 3 months by comparing baseline and third-month PRAS scores.
**Results**
Among 70 patients receiving anakinra, 47 (67.1%) were switched to canakinumab, 15 (21.4%) continued anakinra therapy, and 8 (11.4%) discontinued treatment without biologic switch. Anakinra was used as first-line biologic therapy in 65 patients, whereas 5 patients received secondary anakinra following canakinumab discontinuation; 2 of these patients subsequently required re-initiation of canakinumab. The most common reason for switching from anakinra to canakinumab was recurrent flares (66.0%), followed by allergic reactions (34.0%). One additional patient discontinued anakinra because of allergy without subsequent biologic transition. Overall allergic reaction frequency among all anakinra-treated patients was 22%. Median full-dose anakinra treatment duration was 0.98 (0.49–1.67) months, while median total treatment duration was 4.60 (1.00–13.11) months. Switching occurred earlier in patients with allergic reactions compared to those switched because of recurrent flares [0.73 (0.49–1.0) vs. 7.52 (4.14–13.11) months]. Baseline median PRAS score was 8 (7–10). Among 39 patients treated with anakinra for at least 3 months, PRAS scores decreased from 8 (7–10) to 6 (6–8) at month 3 ($p<0.001$). Flares occurred in 39 patients (55.7%), with a median time to first flare of 3 (2–5) months. Median dosing interval during flares was 5 (1–8) days. Among 23 patients treated exclusively with anakinra, 15 remained flare-free and 9 continued treatment at last follow-up.
**Conclusion**
Anakinra was associated with improvement in disease activity scores in pediatric colchicine-resistant FMF patients. Switching to canakinumab was common and mainly related to recurrent flares and allergic reactions. Anti-drug antibody development may contribute to secondary loss of response during anakinra therapy and warrants further investigation.
**Disclosure of interest**
None declared.

P062

**Intrafamilial concordance in familial mediterranean fever: a sibling pair analysis using kappa and intraclass correlation**

B. Başer Taşkın[1,*], A. Doğru Kılınç[1], B. Danışman Avcı[1], A. Taşkın[2], G. Kavrul Kayaalp[1], F. G. Demirkan[3], A. Tanatar[4], M. Çakan[5], Ö. Akgün[1], N. Aktay Ayaz[1]
[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine; [2]Pediatric Neurology, Istanbul Medipol University; [3]Pediatric Rheumatology, Kanuni Sultan Suleyman Training and Research Hospital; [4]Pediatric Rheumatology, Marmara University Pendik Training and Research Hospital; [5]Pediatric Rheumatology, Zeynep Kamil Women and Children's Diseases Training and Research Hospital, İstanbul, Türkiye

**Table 1 (Abstract P062)** Concordance analysis between siblings for clinical and treatment characteristics

| | Kappa (κ) | 95% CI | SE | *p*-value |
|---|---|---|---|---|
| Fever | 0.261 | 0.075–0.447 | 0.095 | 0.002 |
| Abdominal pain | 0.176 | −0.022–0.374 | 0.101 | **0.038** |
| Chest pain | 0.333 | 0.162–0.504 | 0.087 | **<0.001** |
| Arthritis | 0.229 | 0.057–0.401 | 0.088 | **0.006** |
| Arthralgia | 0.320 | 0.163–0.477 | 0.080 | **<0.001** |
| Exercise-induced leg pain | 0.497 | 0.338–0.656 | 0.081 | **<0.001** |
| Myalgia | 0.363 | 0.189–0.537 | 0.089 | **<0.001** |
| Response to colchicine | 0.223 | 0.045–0.401 | 0.091 | **0.002** |
| Colchicine resistance | 0.308 | 0.083–0.533 | 0.115 | **<0.001** |

**Correspondence:** B. Başer Taşkın
*Pediatric Rheumatology 2026*, **24(S1):**P062

**Introduction**
Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease and is characterized by recurrent inflammatory attacks. Despite its monogenic nature, considerable variability in clinical manifestations has been observed, even among individuals carrying identical MEFV mutations.
**Objectives**
This study aimed to evaluate intrafamilial phenotypic concordance among siblings with FMF and to investigate similarities in clinical features, disease severity, laboratory parameters, and treatment responses.
**Methods**
This retrospective cohort study included 274 patients forming 137 affected sibling pairs followed at tertiary pediatric rheumatology centers. Clinical manifestations, laboratory findings, disease severity scores, and treatment characteristics were recorded. Concordance for categorical variables was evaluated using Cohen's kappa (κ) coefficient, and intraclass correlation coefficients (ICC) were calculated for continuous variables.
**Results**
The median age at diagnosis was 75 months (range 3–214), with a median diagnostic delay of 12 months (range 0–160). The median annual attack frequency decreased from 12 (range 0–60) before treatment to 1 (range 0–48) after colchicine therapy. The most common clinical manifestations were abdominal pain (83.6%), fever (77.3%), and arthralgia (57.7%). Concordance between siblings was generally low for most clinical features (κ = 0.176–0.320), whereas higher agreement was observed for pleuritis (κ = 0.742). Disease severity scores showed moderate-to-strong concordance, particularly ISSF scores (ICC = 0.761 at baseline). Concordance for attack duration was moderate (ICC = 0.475), while no significant concordance was observed for attack frequency. Laboratory parameters demonstrated low-to-moderate concordance (ICC = 0.311–0.506). Complete response to colchicine was achieved in 80.8% of patients, and 12.1% required biologic therapy (see Table 1).
**Conclusion**
Siblings with FMF demonstrate considerable phenotypic variability despite shared genetic background and environmental exposures, suggesting that factors beyond the MEFV genotype contribute to the clinical expression of the disease.
**Disclosure of interest**
None declared.

P063

**Exploring the possible effects of MEFV gene mutations on multisystem inflammatory syndrome in children**

G. Oğuz[1,*], S. Turkucar[1], M. Korkut Oguz[2], S. Yuksel[1], M. Yilmaz[2], D. Gurses[2], K. Karaer[3]
[1]Pediatric Rheumatology; [2]Pediatric Cardiology; [3]Medical Genetics, Pamukkale University, Denizli, Türkiye
**Correspondence:** G. Oğuz
*Pediatric Rheumatology 2026*, **24(S1):**P063

**Introduction**
Multisystem Inflammatory Syndrome in Children (MIS-C) is a post-infectious hyperinflammatory response to SARS-CoV-2 characterized by

fever, cytokine storm, and multisystem organ involvement. MEFV gene mutations, the genetic basis of Familial Mediterranean Fever (FMF), are known to amplify inflammatory responses in conditions such as Kawasaki Disease and systemic JIA. Whether MEFV variants similarly modify the clinical course of MIS-C remains unknown.

**Objectives**

This study aims to evaluate the relationship between the frequency and severity of MIS-C and MEFV gene variants.

**Methods**

This retrospective study included 45 pediatric MIS-C patients followed at Pamukkale University Faculty of Medicine between 2020 and 2022. Demographic data, laboratory parameters, organ system involvements, and MEFV gene variant analyses were retrieved from medical records. Disease severity was classified as mild, moderate, or severe based on respiratory/hemodynamic support requirements and organ dysfunction scores.

**Results**

The median age at diagnosis was 97 months; 64.4% were male. Disease severity was mild in 14 (31.1%), moderate in 14 (31.1%), and severe in 17 (37.8%) patients. The most common organ involvements were gastrointestinal (82.2%), hematological (82.2%), and cardiac (75.6%). MEFV gene variants were identified in 11 patients (24.4%), all in heterozygous form. The most frequent variant was E148Q (13.3%, *n*=6), followed by R202Q (2.2%) and Exon 10 mutations including M694V, M680I, V726A, and R761H (8.8% total). Critically, none of the 14 mild cases carried an MEFV variant, whereas all 11 mutation-positive patients presented with moderate (*n*=6) or severe (*n*=5) disease ($p$=0.013; RR=1.7, 95% CI: 1.21–2.21). Renal involvement was exclusively observed in MEFV mutation carriers (27.3% vs. 0%, $p$=0.012). No significant differences were found between groups in CRP, IL-6, ferritin, or other inflammatory markers ($p$>0.05).

**Conclusion**

MEFV gene variants were present in approximately one-quarter of MIS-C patients, a prevalence exceeding known carrier rates in the general Turkish population. All mutation-positive patients developed moderate-to-severe disease, and renal involvement clustered exclusively in this group. Despite similar inflammatory marker profiles across groups, MEFV carrier status appears to drive a qualitative shift in tissue-level inflammation — consistent with a two-hit model in which genetic predisposition primes the inflammasome and SARS-CoV-2 acts as the triggering second hit. In FMF-prevalent regions, early MEFV screening in MIS-C patients may support risk stratification and personalized management.

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**Disclosure of interest**

None declared.

## P064

### Blau syndrome: a single-centre experience from India beyond the classic triad

N. K. Bagri[1,2,*], N. Shaikh[3], P. Kaur[1], N. Gupta[1], M. R. Chowdhury[1], D. Bhadu[1], V. Gupta[4], R. Chawla[5], M. Jana[6]

[1]Pediatrics, All India Institute of Medical Sciences, New Delhi; [2]Pediatrics, All India Institute of Medical Sciences; [3]Shaikh, All India Institute of Medical Sciences, New Delhi; [4]Dermatology, All India Institute of Medical Sciences; [5]Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi; [6]Radiodiagnosis and Interventional Radiology, All India Institute of Medical Sciences, New Delhi, India

**Correspondence:** N. K. Bagri

*Pediatric Rheumatology 2026*, **24(S1):**P064

**Introduction**

Blau syndrome is a rare, monogenic granulomatous autoinflammatory syndrome resulting from a gain-of-function mutation in the gene encoding nucleotide-binding and oligomerisation domain-containing protein 2 (NOD 2). The classic triad of Blau syndrome consists of arthritis, uveitis, and dermatitis. Due to variable presentation, these patients present to various specialists before definitive diagnosis.

**Objectives**

To review the clinical course, radiological characteristics, and outcomes of patients with Blau syndrome from a single tertiary care hospital.

**Methods**

Data of patients with Blau syndrome enrolled in the pediatric rheumatology clinic (2015-2026) were reviewed and collected in a pre-designed proforma and analyzed with descriptive statistics.

**Results**

Blau syndrome was genetically confirmed in all eighteen patients (15 children and 3 adults). The median (IQR) age of children at diagnosis was 9 (4-17) years. The median (IQR) age of symptom onset was 24 (6-30) months, ranging from 3 to 336 months. The classic triad of uveitis, arthritis and dermatitis was present in 12 (66.6%) during 28.6 patient years of follow-up. Arthritis was the most common initial symptom (*n*=8/18, 44.4%). Uveitis was present in *n*=17 (94.4%). *N*=8 (44.4%) children had systemic features- fever, lymphadenopathy, and hepatosplenomegaly. Dysmorphism (*n*=2) was apparent in *n*=7(38.9%). Articular ultrasound showed characteristic synovial proliferation, with a globular contour similar to a 'sac of marbles' in those with active arthritis. Adalimumab was administered to n=11(61.1%) children. N=14 (77.7%) were in clinical remission at the last follow-up. Visual impairment was the most common debilitating morbidity noted.

**Conclusion**

Blau syndrome should be considered as a differential diagnosis in the presence of early-onset arthritis with characteristic ultrasonographic findings, and uveitis. Early diagnosis and initiation of therapy are essential to curtail the morbidities, particularly ocular.

**Disclosure of interest**

None declared.

## P065

### Clinical comparison of homozygous and compound heterozygous MEFV mutations in childhood FMF: a single-center experience

Y. Yaman[1], O. Altug Gucenmez[1,*], B. E. Kosali[1]

[1]Department of Pediatric Rheumatology, University of Health Sciences, Dr. Behçet Uz Pediatric Diseases and Surgery Training and Research Hospital, Izmir, Türkiye

**Correspondence:** O. Altug Gucenmez

*Pediatric Rheumatology 2026*, **24(S1):**P065

**Introduction**

Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease and is characterized by recurrent self-limited inflammatory attacks (1). It is caused by mutations in the MEFV gene, which encodes pyrin. Although some MEFV variants are associated with more severe disease, genotype–phenotype correlations and treatment responses remain heterogeneous in pediatric FMF (2).

**Objectives**

This study aimed to compare pediatric FMF patients with homozygous and compound heterozygous *MEFV* mutations in terms of demographic characteristics, age at disease onset, age at diagnosis, attack frequency, attack duration, clinical manifestations, colchicine response, and need for anakinra treatment.

**Methods**

This single-center retrospective study included 134 pediatric patients with FMF followed in a pediatric rheumatology clinic. Patients were grouped according to MEFV mutation status as homozygous or compound heterozygous. Demographic, clinical, attack-related, and treatment data were obtained from medical records. Age at symptom onset and diagnosis, annual attack frequency, attack duration, clinical manifestations, colchicine response, and anakinra requirement were compared between groups. Mann–Whitney U, chi-square, and Fisher's exact tests were used as appropriate.

**Results**

A total of 134 pediatric FMF patients were included, of whom 99 (73.9%) had homozygous and 35 (26.1%) had compound heterozygous MEFV mutations. No statistically significant differences were observed between the two groups in age at disease onset, age at diagnosis, annual attack frequency, or attack duration. Clinical manifestations were similarly distributed between groups. Although colchicine response was numerically higher in the compound heterozygous group and

anakinra requirement was numerically higher among homozygous patients, neither difference reached statistical significance.

**Conclusion**

In this pediatric FMF cohort, homozygous and compound heterozygous MEFV mutation patterns were not associated with significant differences in clinical phenotype or treatment response. Although colchicine response tended to be higher in compound heterozygous patients and anakinra requirement tended to be higher in homozygous patients, neither difference was statistically significant. These findings suggest that genotype alone may be insufficient to predict disease course or therapeutic needs in pediatric FMF.

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**Disclosure of interest**

None declared.

## P066

**Is infantile colic more common in children with familial Mediterranean fever?**

O. Altug Gucenmez[1,*], B. E. Kosali[1]
[1]Department of Pediatric Rheumatology, University of Health Sciences, Dr. Behçet Uz Pediatric Diseases and Surgery Training and Research Hospital, Izmir, Türkiye
**Correspondence:** O. Altug Gucenmez
*Pediatric Rheumatology 2026*, **24(S1):**P066

**Introduction**

Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease, characterized by recurrent self-limited episodes of fever, serositis, abdominal pain, and systemic inflammation (1). Although FMF usually becomes clinically apparent during childhood, nonspecific gastrointestinal symptoms may be observed earlier in life (2). However, whether infantile colic may be more frequent in children who later develop autoinflammatory diseases such as FMF remains unclear.

**Objectives**

This study aimed to evaluate whether children diagnosed with FMF differ from healthy controls in terms of a previous history of infantile colic. We also aimed to compare demographic, perinatal, feeding-related, and maternal characteristics between the two groups in order to identify possible factors associated with infantile colic history.

**Methods**

This study included a total of 200 children, consisting of 100 patients with FMF and 100 healthy controls. Demographic and perinatal data, including age, sex, birth weight, birth order, and the primary caregiver during the first six months of life, were recorded for all participants. Early feeding characteristics, including breastfeeding and complementary feeding during the first six months, were collected as dichotomous variables. A history of infantile colic diagnosis was assessed as present or absent based on parental report and/or medical history. Maternal characteristics, including maternal age, education level, and employment status, were also documented. The frequency of infantile colic and related variables were compared between the FMF and control groups.

**Results**

A total of 100 children with FMF and 100 healthy controls were evaluated. Statistically significant differences were observed between the groups in terms of age, maternal age, maternal education level, birth order, and complementary feeding during the first six months of life ($p$<0,05). Despite these differences in demographic and early-life characteristics, there was no statistically significant difference between children with FMF and healthy controls regarding a history of infantile colic diagnosis. These findings suggest that infantile colic history was not more frequent among children with FMF compared with their healthy peers.

**Conclusion**

In this study, infantile colic was not associated with a later diagnosis of FMF. Although several demographic, maternal, and early feeding-related variables differed between the FMF and control groups, the frequency of infantile colic diagnosis was similar. These findings suggest that infantile colic may not represent an early clinical indicator of FMF. Further prospective studies with larger cohorts are needed to clarify whether early-life gastrointestinal symptoms have any relationship with autoinflammatory diseases.

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**Disclosure of interest**

None declared.

## P067

**Clinical characteristics, genetic findings, and treatment outcomes in children with PFAPA syndrome: a single-center experience**

C. Ozbakir[1], O. Altug Gucenmez[1,*], B. E. Kosali[1]
[1]Department of Pediatric Rheumatology, University of Health Sciences, Dr. Behçet Uz Pediatric Diseases and Surgery Training and Research Hospital, Izmir, Türkiye
**Correspondence:** O. Altug Gucenmez
*Pediatric Rheumatology 2026*, **24(S1):**P067

**Introduction**

Periodic fever, aphthous stomatitis, pharyngitis, and cervical adenitis (PFAPA) syndrome is a common autoinflammatory fever syndrome of early childhood, characterized by recurrent self-limited febrile episodes with aphthous stomatitis, pharyngitis, and/or cervical lymphadenitis, and well-being between attacks (1). Corticosteroids effectively terminate attacks but may shorten attack intervals in some patients, while the roles of colchicine and tonsillectomy in long-term control remain clinically relevant (2).

**Objectives**

This study aimed to evaluate the demographic and clinical characteristics, attack patterns, laboratory findings, MEFV mutation status, and treatment responses to corticosteroids, colchicine, and tonsillectomy in children followed with a diagnosis of PFAPA syndrome.

**Methods**

We retrospectively reviewed the medical records of patients followed with PFAPA syndrome between January 2021 and January 2025. Demographic characteristics, age at diagnosis, attack duration, interval between attacks, clinical manifestations, laboratory parameters, genetic analysis results, and treatment modalities were recorded. Responses to corticosteroid treatment during attacks, colchicine prophylaxis, and tonsillectomy were evaluated. Treatment response was assessed according to reduction in attack frequency, control of febrile episodes, and postoperative clinical remission.

**Results**

A total of 22 patients were included in the study. The mean age was 77.6 months, and 50% of the patients were male. The median attack duration was 4 days (IQR: 3–6), and the median interval between attacks was 30 days (IQR: 25–41.3). MEFV mutations were detected in 36.4% of patients ($n$=8). All patients showed clinical improvement with corticosteroid therapy during febrile attacks; however, increased attack frequency following corticosteroid use was observed in 45.5% of cases. Colchicine was used in seven patients, and a decrease in attack frequency was observed in 57.1% (4/7). Tonsillectomy was performed in 63.6% of patients ($n$=14). No postoperative complications were reported. Complete clinical recovery was achieved in all patients who underwent tonsillectomy during follow-up.

**Conclusion**
This single-center experience demonstrates that corticosteroids are effective for acute control of PFAPA attacks, although increased attack frequency may occur in a substantial proportion of patients. Colchicine may reduce attack frequency in selected cases, particularly when prophylactic treatment is considered. Tonsillectomy was associated with complete clinical recovery and no postoperative complications in this cohort, supporting its role as an effective treatment option for selected children with PFAPA syndrome. Larger prospective studies are needed to better define individualized treatment strategies and long-term outcomes in pediatric PFAPA.

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**Disclosure of interest**
None declared.

## P068

**Need for biologic treatment re-initiation after discontinuation in pediatric colchicine-resistant familial Mediterranean fever**

Y. Uğur Es[1,*], E. Özçelik[1], S. Erdem Torun [1], D. Öztürk[1], S. N. Yoğun[1], M. Işıklar Ekici[1], E. N. Şener[1], E. Çelikel[1], Z. Ekici Tekin[1,2], C. Karagöl[1], S. Ertem [1], E. Esen Teker[1], B. Çelikel Acar[1]
[1]Pediatric Rheumatology, Ankara Bilkent City Hospital; [2]Pediatric Rheumatology, Ankara Yıldırım Beyazıt University, Ankara, Türkiye
**Correspondence:** Y. Uğur Es
*Pediatric Rheumatology 2026*, **24(S1):**P068

**Introduction**
Data regarding the discontinuation and, when necessary, re-initiation of biologic therapy in patients with colchicine-resistant familial Mediterranean fever (FMF) are limited.
**Objectives**
This study aimed to evaluate the outcomes of biologic therapy discontinuation in pediatric FMF patients with colchicine-resistant disease and to identify the factors associated with the re-initiation of biologic therapy.
**Methods**
Medical records of FMF patients followed at a tertiary pediatric rheumatology center between January 2013 and December 2024 were retrospectively reviewed. Patientswith colchicine-resistant FMF who received biologic therapy were included. Demographic, clinical, laboratory, and treatment-related characteristics were compared between patients with and without biologic discontinuation and between those requiring and not requiring biologic treatment re-initiation after discontinuation. Kaplan–Meier analysis was performed to evaluate biologic treatment re-initiation–free survival.
**Results**
Of 1290 patients analyzed, 57 received biologic therapy. Biologic treatment was successfully discontinued in 20 patients (35.1%). Biologic treatment re-initiation was required in 11 patients (55%) after discontinuation, with a median re-initiation–free survival of 18 months. The symptom-to-diagnosis interval was significantly longer and family history of colchicine-resistant FMF was more common among patients who achieved discontinuation. No significant clinical or laboratory predictors of biologic treatment re-initiation were identified. Among patients requiring re-initiation, 90.9% carried homozygous M694V mutations and most had moderate-to-severe disease activity prior to biologic therapy.
**Conclusion**
Biologic therapy could be successfully discontinued in approximately one-third of pediatric patients with colchicine-resistant FMF. Nevertheless, more than half of the patients required reinitiation of biologic treatment following discontinuation. Homozygous M694V mutations may be associated with a lower likelihood of sustained remission after cessation of biologic therapy.

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**Disclosure of interest**
None declared.

## P070

**Non-autoinflammatory mimickers of familial Mediterranean fever: a case series**

E. Aslan[1,*], S. Sahin[1], E. Aslan[1], Z. Torunoglu[1], H. Babazade[2], M. Aydin[3], N. Akay[1], U. Gul[1], E. Kilic Konte[1], M. Yildiz[1], T. Zubarioglu[2], E. Ozek Yucel[3], E. Kiykim[2], K. Barut[1], C. Aktuglu-Zeybek[2], O. Kasapcopur[1]
[1]Pediatric Rheumatology; [2]Pediatric Nutrition and Metabolism; [3]Pediatric Allergy and Immunology, Istanbul University- Cerrahpasa, Istanbul, Türkiye
**Correspondence:** E. Aslan
*Pediatric Rheumatology 2026*, **24(S1):**P070

**Introduction**
Familial Mediterranean Fever (FMF) is the most common monogenic autoinflammatory disease characterized by recurrent fever and serositis. While under-recognition remains a concern in low-prevalence regions, overdiagnosis poses a significant challenge in endemic areas, where several non-autoinflammatory conditions may closely mimic FMF.
**Objectives**
To highlight clinical clues for differential diagnosis in patients initially suspected of FMF.
**Methods**
**Case 1**
A 17-year-old girl had a four-year history of recurrent abdominal pain attacks lasting 5-10 days with repeatedly normal acute phase reactants and with no pathogenic *MEFV* variant. Previously labeled as SURF, she was on colchicine. An attack with hyponatremia and hypomagnesemia prompted porphyria workup. Genetic analysis identified a pathogenic HMBS variant, confirming acute intermittent porphyria.
**Case 2**
A 9-year-old girl had recurrent fever and abdominal pain with markedly elevated acute phase reactants during attacks and normal intervals. No pathogenic MEFV variant was detected. She was labeled as SURF and colchicine response was partial. Lipemic serum during an attack revealed severe hypertriglyceridemia with elevated amylase and lipase, consistent with hypertriglyceridemia induced recurrent acute pancreatitis.
**Case 3**
A 14-year-old girl had episodes of abdominal pain and arthritis since age six. A heterozygous R202Q MEFV variant was noted, and she was referred with suspected FMF. During a subsequent episode she developed periorbital and lip edema accompanied by an urticarial rash. Autoinflammatory exome panel identified a pathogenic F12 variant, confirming hereditary angioedema type 3. C1 esterase inhibitor therapy was effective.
**Case 4**
An 8-year-old boy had a three-year history of monthly ankle pain, erythema, and swelling resembling erysipelas-like erythema, lasting 5–7 days, triggered by exercise. Palpable purpura led to an initial consideration of IgA vasculitis alongside FMF. Colchicine response was partial, MEFV analysis revealed no pathogenic variant. Two years later, recurrent vasculitic but atypical lesions prompted further evaluation. Hepatosplenomegaly and hypertriglyceridemia led to the diagnosis of familial chylomicronemia syndrome. The erysipelas-like erythema and atypical lesions were ultimately attributed to vascular stasis secondary to severe hypertriglyceridemia.
**Conclusion**
In endemic regions, FMF is often the primary consideration in children with periodic fever or recurrent abdominal pain. However, more

prolonged attacks with normal acute phase reactants, colchicine resistance, negative *MEFV* gene analysis, headache, and urinary incontinence during attacks (1) should prompt diagnostic reconsideration.

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**Disclosure of interest**
None declared.

## P071

**Clinical spectrum of pediatric patients with heterozygous MEFV gene variants**

E. Erorhan[1,*], F. Aydın[1], P. Ö. Avar Aydın[1], O. Bahçeci[1], D. Sarısoy[1], B. Tetik Dinçer[1], Ö. Taş Aslan[1], Z. B. Özçakar[1]
[1]Department of Pediatrics, Division of Pediatric Rheumatology, Ankara University School of Medicine, Ankara, Türkiye
**Correspondence:** E. Erorhan
*Pediatric Rheumatology 2026*, **24(S1):**P071

**Introduction**
The *MEditerranean FeVer* (*MEFV*) gene plays an important role in the regulation of the innate immune system. Familial Mediterranean fever (FMF) is the prototype disease associated with *MEFV* gene mutations. In recent years, heterozygous *MEFV* variants have also been linked to a broader range of inflammatory diseases beyond FMF.
**Objectives**
To investigate the diagnostic spectrum, demographic characteristics, and clinical findings of pediatric patients carrying heterozygous *MEFV* variants.
**Methods**
Pediatric patients with heterozygous *MEFV* variants who were followed at a single rheumatology referral center between January 2012 and January 2025 were included in this study. Demographic data, diagnoses, and clinical manifestations were reviewed.
**Results**
A total of 270 patients were included, with a median age of 7 years. Diagnoses were FMF in 67%, IgA vasculitis in 7%, PFAPA syndrome in 6%, inflammatory bowel disease in 3%, juvenile idiopathic arthritis in 3%, chronic nonbacterial osteomyelitis in 2%, Behçet disease in 1%, and other vasculitides in 1% of the study population. 14% of the patients were asymptomatic carriers. During follow-up, 22 of 74 patients (30%) who were initially diagnosed with other inflammatory disorders subsequently developed findings compatible with FMF. Colchicine treatment was prescribed not only for classical FMF attacks but also in selected patients with other inflammatory diseases, with favorable clinical outcomes.
**Conclusion**
Heterozygous *MEFV* variants may be associated with a broad spectrum of inflammatory diseases in addition to FMF. Follow-up is important, as some patients may later fulfill criteria for FMF. *MEFV* gene testing should be considered in patients with severe or atypical inflammatory diseases, especially in regions where FMF is common. Colchicine may offer clinical benefit in selected cases.
**Disclosure of interest**
None declared.

## P072

**H syndrome with skin involvement, arthrogryposis-like joint deformities, arthralgia and fasciitis, exophthalmos, skeletal abnormalities and short stature: case report and therapeutical management**

E. A. Conti[1,*], M. Jouret[2], A. Belot[1,2]
[1]CIRI INSERM; [2]Hopital Femme Mere Enfant, Lyon, France
**Correspondence:** E. A. Conti
*Pediatric Rheumatology 2026*, **24(S1):**P072

**Introduction**
H syndrome is a rare autosomal recessive histiocytosis caused by biallelic *SLC29A3* variants, typically presenting with cutaneous induration, hypertrichosis, skeletal deformities and variable systemic inflammation. Therapeutic strategies targeting cytokine and MAPK pathways are increasingly reported, but evidence remains limited.
**Objectives**
A 6-year-old girl born to consanguineous Tunisian parents presented with early gait disturbance, camptodactyly, hallux valgus, short stature with normal GH axis, and long-standing indurated hypertrichotic plaques. MRI (March 2024) revealed extensive perifascial and intermuscular inflammatory infiltration of the lower limbs, retroperitoneum and paravertebral regions. Skin and muscle biopsies showed deep dermal and hypodermal histiocytic infiltrates with panniculitis. Genetic testing identified homozygous *SLC29A3* P324L variants. CRP peaked at 37.5 mg/L without fever. The baseline interferon signature was negative, became weakly positive (score 5) shortly before treatment, and normalized after therapy.
**Methods**
Tocilizumab was initiated at 12 mg/kg IV every 4 weeks and intensified to every 2 weeks due to persistent inflammation, with subsequent spacing and later tapering, resulting in clinical improvement, catch-up growth and complete radiologic resolution of infiltrates after 6 months from the beginning of the treatment. In March 2026, a clinical flare with lower-limb pain and limp corresponded to persistent retroperitoneal, retromediastinal and lateral-thigh infiltrates on MRI. Tocilizumab was tapered, and cobimetinib (MEK inhibitor) was introduced based on histologic evidence of MAPK pathway activation.

**Learning points**

- High-dose IL-6 blockade (tocilizumab) can induce clinical and radiologic remission and reduce systemic and tissue inflammation, but may not fully control histiocytic proliferation when MAPK activation persists.
- MEK/MAPK inhibitors (e.g., cobimetinib) may be effective when pERK is elevated or when IL-6 blockade alone is insufficient, supporting the concept that targeting the MAPK pathway can reduce deep-tissue histiocytosis.
- Sequential targeting of IL-6 and MAPK pathways is a rational therapeutic strategy in selected patients and warrants prospective evaluation.
- MRI is the modality of choice for monitoring disease activity in paediatric patients and is particularly useful for detecting deep-tissue lesions.

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**Disclosure of interest**
None declared.

## P073

**Expanding the genetic spectrum of NEMO-NDAS: a pediatric case driven by a novel IKBKG splice-site variant arising from pseudogene conversion**

F. Anselmi[1,*], P. Dusser[1], I. Kone- Paut[1]
[1]Bicetre University Hospital, Paris, France
**Correspondence:** F. Anselmi
*Pediatric Rheumatology 2026*, **24(S1):**P073

**Introduction**
NEMO-NDAS is a rare autoinflammatory disorder caused by pathogenic variants in the *IKBKG* gene.Its clinical and molecular spectrum remains incompletely defined.
**Objectives**
To report a pediatric case of early-onset autoinflammatory disease and to characterize a novel pathogenic *IKBKG* splice-site variant, expanding the genetic spectrum of NEMO-NDAS.

**Methods**
We analyzed the clinical course, laboratory, histopathology, imaging, and genetic features of an 8-year-old girl presenting with early-onset recurrent inflammatory episodes. A comprehensive diagnostic work-up, including extensive infectious, immunological, and inflammatory assessments, was undertaken.Genetic investigations included an NGS panel for autoinflammatory diseases, trio-based whole genome sequencing (WGS), in silico splice prediction, and RNA transcript analysis.
**Results**
Disease onset occurred at 20 months of age with recurrent febrile episodes lasting 24–72 h, occurring approximately twice monthly. Episodes were associated with painful subcutaneous lesions predominantly involving the lower limbs and buttocks, occasionally extending to the trunk and upper limbs, and intermittent arthralgia. Skin biopsy demonstrated lobular and septal panniculitis with a dense neutrophilic infiltrate.Articular imaging was unremarkable.During flares, laboratory evaluation revealed marked systemic inflammation with elevated CRP, ESR, and serum amyloid A, sometimes associated with mild anemia. Cytokine profiling showed increased IL-6 and IL-18 levels, with negative interferon signature. Between episodes, low-grade biological inflammation peristed. Autoimmune investigations were negative, and no evidence of immunodeficiency or infectious etiology, including mycobacterial disease, was identified. Multiple therapeutic approaches (colchicine, NSAIDs, corticosteroids, low-dose daily anakinra) resulted in only partial or transient responses. Anakinra administered during flares at 6 mg/kg/day led to rapid clinical improvement, including resolution of fever and reduction of cutaneous lesions. However, inflammatory markers remained slightly elevated between episodes. Initial NGS panel testing was negative.Trio WGS identified a heterozygous 18-nucleotide deletion affecting the canonical splice acceptor site of intron 4 in *IKBKG* (c.519-19_519-2del), inherited from the father. Variant interpretation was challenging due to >99% sequence homology with the *IKBKGP1* pseudogene. Functional analyses resolved this ambiguity: in silico predictions indicated disruption of the native splice acceptor site, and RNA transcript analysis confirmed exon 5 skipping, resulting in an in-frame deletion (p.Ala174_Lys224del).This established a class 5 pathogenic variant consistent with NEMO-NDAS.The underlying mechanism is most consistent with interallelic gene conversion from the *IKBKGP1* pseudogene to *IKBKG*.
**Conclusion**
This case expands the genetic spectrum of NEMO-NDAS by identifying a novel splice-site *IKBKG* variant arising from pseudogene conversion.It highlights the importance of integrating genomic and transcriptomic analyses in complex loci.Although IL-1 blockade controls clinical flares, persistent subclinical inflammation suggests incomplete disease control. n light of the molecular diagnosis, escalation toward targeted therapies, including anti-TNF agents, is currently under evaluation.
**Disclosure of interest**
None declared.

## P074

### Five-year diagnostic spectrum of autoinflammatory diseases in a tertiary pediatric rheumatology centre: FMF and beyond

H. Öztürk Aydin[1,*], E. Tunce[1], S. Atamyıldız Uçar[1], N. Kara Çanlıoğlu[1], M. Eser[2], B. Sözeri[1]
[1]Pediatric Rheumatology; [2]Medical Genetics, Umraniye Training and Research Hospital, Istanbul, Türkiye
**Correspondence:** H. Öztürk Aydin
*Pediatric Rheumatology 2026*, **24(S1):**P074

**Introduction**
Autoinflammatory diseases (AIDs) are a heterogeneous group of disorders driven by innate immunity dysregulation. FMF is the most prevalent monogenic AID in endemic regions including Türkiye, yet the frequency and distribution of newly diagnosed AIDs in tertiary paediatric rheumatology centres remain incompletely characterised.
**Objectives**
To determine the frequency and diagnostic distribution of newly diagnosed AIDs with a particular focus on FMF versus non-FMF entities among patients evaluated for suspected AIDs over a 5-year period in a tertiary pediatric rheumatology center.
**Methods**
This retrospective study included all patients evaluated for suspected AIDs at our tertiary paediatric rheumatology clinic between 2021 and 2026, identified via ICD-10 codes E85.0–E85.9 and R50.9. Newly diagnosed AID cases and diagnosis distribution were recorded annually. Descriptive statistical analyses were performed.
**Results**
A total of 11,437 patients were evaluated for diagnosed and suspected AID over 5 years, of whom 698 received a new AID diagnosis. FMF was the most frequent diagnosis ($n$=383, 54.9%), followed by PFAPA ($n$=203, 29.1%) and syndrome of undifferentiated recurrent fever (SURF; $n$=53, 7.6%). Among non-FMF monogenic AIDs, the most common were FCAS1 ($n$=17), HIDS ($n$=13), DADA2 ($n$=10), FCAS2 ($n$=7), TRAPS ($n$=6), Blau syndrome ($n$=5), and MWS/CAPS ($n$=1). Annual newly diagnosed FMF cases were 71 (3.6%), 116 (4.9%), 126 (4.0%), 40 (1.7%), and 30 (1.9%) of all suspected evaluations from the first to the fifth year, respectively. In the FMF cohort, the median age at symptom onset was 5.1 years (IQR 3.2–8.5) and the median diagnostic delay was 1.9 years (IQR 0.8–4.2). The most frequent MEFV variant was M694V (42%), followed by M680I, E148Q, and V726A.
**Conclusion**
FMF accounted for more than half of all newly diagnosed AIDs, confirming its central role in endemic settings. A broad spectrum of non-FMF AIDs was also identified, spanning polygenic conditions (PFAPA, SURF) and seven distinct monogenic entities, underscoring the diagnostic complexity in tertiary centres. A median diagnostic delay of nearly 2 years in FMF highlights the need for heightened awareness and structured diagnostic pathways. These findings provide a real-world framework for AID distribution in a high-prevalence region and may inform genetic testing strategies and clinical training in paediatric rheumatology.

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**Disclosure of interest**
None declared.

## P075

### Beyond urticaria: persistent eczematous fingertip lesions leading to the diagnosis of cryopyrin-associated periodic syndrome

H. Adiguzel Dundar[1,*]
[1]Department of Pediatric Rheumatology, Behcet Uz Pediatric Diseases And Surgery Training And Research Hospital, Izmir, Türkiye
**Correspondence:** H. Adiguzel Dundar
*Pediatric Rheumatology 2026*, **24(S1):**P075

**Introduction**
Cryopyrin-Associated Periodic Syndrome is classically characterized by recurrent fever, urticaria-like rash, and systemic inflammation. Atypical dermatologic manifestations may delay diagnosis.
**Objectives**
To present an atypical CAPS case with chronic eczematous fingertip lesions without classical urticaria-like rash or prominent recurrent fever attacks.
**Methods**
Clinical, laboratory, genetic, and treatment data of a child diagnosed with CAPS were retrospectively evaluated. A 4-year-old girl was referred because of persistent inflammatory marker elevation and chronic treatment-resistant skin lesions suspected to be associated with an underlying rheumatic disease. Cutaneous symptoms had started at 2 years of age, and she had been followed in dermatology clinics for approximately two years before referral to pediatric rheumatology. Retrospective history revealed intermittent episodes of fever and abdominal pain beginning at approximately 1.5 years of age; however, these episodes resolved spontaneously within 1–2 days and were not

initially recognized as inflammatory attacks. The patient also had a history of bullous lesions involving the fingers and wrists. At presentation, cutaneous involvement was limited to the distal aspects of all fingers, characterized by fragile eczematous and psoriasiform erythematous lesions with desquamation and focal hemorrhagic areas. Pruritus was not prominent. Unlike the classical CAPS phenotype, urticaria-like rash was absent and recurrent fever was not a major complaint at presentation. Laboratory investigations demonstrated persistent elevation of acute phase reactants, including C-reactive protein and serum amyloid A, even outside clearly defined attacks. Familial Mediterranean fever was initially considered; however, MEFV mutation analysis was negative. Empirical colchicine treatment was initiated while an autoinflammatory gene panel was pending, without improvement in either skin lesions or inflammatory markers. Genetic analysis identified a heterozygous NLRP3 p.Gln705Lys variant. Audiologic evaluation was normal. Following initiation of anakinra therapy, both inflammatory markers and cutaneous lesions improved dramatically. CAPS should be considered in children with persistent treatment-resistant inflammatory skin lesions and unexplained elevation of acute phase reactants, even in the absence of classical urticaria-like rash or prominent recurrent fever attacks.

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**Disclosure of interest**
None declared.

## P076

### Clinical features and outcomes of pediatric protracted febrile myalgia syndrome: a multicenter retrospective cohort study

H. Ilgaz Tuzen[1,*], E. Tunce[2], E. Kılıç Könte[3], E. Özçelik[4], F. Aydın[5], A. Şen[6], A. Soylu[7], B. Acun Sayılı[8], M. Çakan[9], L. Koru[10], T. Çeltik[11], P. Garipçin[12], P. Prencuva Akyürek[13], G. Sümer[14], H. D. Dara Kar[15], S. Ayduran[16], T. Kasap[17], S. Şener[18], F. G. Demirkan[19], S. Yüksel[17], S. Türkuçar[16], E. Sağ[15], S. S. Kilic[20], N. Aktay Ayaz[13], A. Paç Kısaarslan[12], B. Bora[11], F. Haşlak[10], S. Bakkaloğlu Ezgü[8], E. Bağlan[6], Z. B. Özçakar[5], B. Çelikel Acar[4], Ö. Kasapçopur[3], B. Sözeri[2], B. Kasap Demir[1,21]
[1]Pediatric Rheumatology, İzmir City Hospital, İzmir; [2]Pediatric Rheumatology, Ümraniye Training and Research Hospital; [3]Pediatric Rheumatology, Cerrahpasa Faculty of Medicine, İstanbul University- Cerrahpasa, İstanbul; [4]Pediatric Rheumatology, University of Health Sciences, Ankara Bilkent City Hospital; [5]Pediatric Rheumatology, Ankara University School of Medicine; [6]Pediatric Rheumatology, Ankara Etlik City Hospital, Ankara; [7]Pediatric Nephrology, Dokuz Eylül University Faculty of Medicine, İzmir; [8]Pediatric Rheumatology, Gazi University Faculty of Medicine, Ankara; [9]Pediatric Rheumatology, Zeynep Kamil Women and Children's Diseases Training and Research Hospital; [10]Pediatric Rheumatology, Istanbul Medeniyet University, İstanbul; [11]Pediatric Rheumatology, Dokuz Eylül University Faculty of Medicine, İzmir; [12]Pediatric Rheumatology, Erciyes University Faculty of Medicine, Kayseri; [13]Pediatric Rheumatology, Istanbul Faculty of Medicine, Istanbul University, İstanbul; [14]Pediatric Rheumatology, Bursa Uludag University, Bursa; [15]Pediatric Rheumatology, Hacettepe University Faculty of Medicine, Ankara; [16]Pediatric Rheumatology, Pamukkale University, Denizli; [17]Pediatric Rheumatology, Çanakkale Onsekiz Mart University, Çanakkale; [18]Pediatric Rheumatology, Adana City Training and Research Hospital, Adana; [19]Pediatric Rheumatology, University of Health Sciences Kanuni Sultan Suleyman Research and Training Hospital, İstanbul; [20]Pediatric Immunology and Rheumatology, Bursa Uludag University, Bursa; [21]Pediatric Rheumatology and Nephrology, İzmir Katip Çelebi University, İzmir, Türkiye
**Correspondence:** H. Ilgaz Tuzen
*Pediatric Rheumatology 2026*, **24(S1):**P076

**Introduction**
Familial Mediterranean fever (FMF), the most common monogenic autoinflammatory disease, may present with various myalgia patterns. Protracted febrile myalgia syndrome (PFMS) is a rare and severe manifestation with limited pediatric data.

**Objectives**
This multicenter study aimed to define the clinical, laboratory, genetic, imaging, and treatment characteristics of pediatric PFMS and to identify factors associated with disease course and outcomes.
**Methods**
This retrospective multicenter study included 103 pediatric patients diagnosed with PFMS at 19 pediatric rheumatology centers in Türkiye. Data were collected using a standardized case report form. Demographic, clinical, laboratory, genetic, imaging, treatment, and outcome data were analyzed. Subgroup comparisons and exploratory logistic regression analyses were performed for selected clinical outcomes.
**Results**
A total of 103 patients (57 females, 46 males) were included, with a median age of 16.4 years and a median follow-up of 5.3 years. All patients presented with severe myalgia accompanied by elevated acute-phase reactants and normal creatine phosphokinase levels. Fever was present in 77.7% of patients, while 22.3% had afebrile attacks. PFMS was the first manifestation of FMF in 26.2% of cases. The M694V variant was absent in 9.7% of patients, and asymmetric myalgia was observed in 14.6%, highlighting clinical heterogeneity. Corticosteroids were administered in 80.6% of patients, with rapid clinical response in 96.4%. Anti-IL-1 therapy was required in 9.7% of patients and resulted in complete clinical response in all cases. Relapse occurred in 9.7% of patients during follow-up. Exploratory analyses showed that relapse was associated with pulse methylprednisolone use and longer hospitalization, whereas early steroid response was associated with reduced need for anti-IL-1 therapy.
**Conclusion**
PFMS in childhood represents a heterogeneous and diagnostically challenging FMF phenotype with a broader clinical spectrum than previously recognized. Afebrile presentations, occurrence as the first manifestation of FMF, and the need for biologic therapy in selected patients highlight the variability of the disease. While corticosteroids remain the mainstay of treatment, anti-IL-1 therapy is an effective option in refractory or recurrent cases.
**Disclosure of interest**
None declared.

## P077

### Use of biologic therapy in patients with monogenic autoinflammatory diseases: data from a federal centre

S. Salugina[1], A. Sukhanina[2], E. Fedorov[2], I. Nikishina[2,*], A. Torgashina[3]
[1]Pediatric Department, V. A. Nasonova Research Institute of Rheumatology, Moscow; [2]Pediatric Department; [3]Laboratory for Rare Rheumatic Diseases and Sjögren's Syndrome, V. A. Nasonova Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** I. Nikishina
*Pediatric Rheumatology 2026*, **24(S1):**P077

**Introduction**
In the pre-biologic era, therapeutic options for patients (pts) with monogenic autoinflammatory diseases (mAIDs) were mainly limited to colchicine for familial Mediterranean fever (FMF), while NSAIDs and glucocorticoids (GCs) were used as symptomatic treatment in other mAIDs. Biologic therapy (BT) has expanded treatment options, enabling control of inflammatory activity and prevention of complications, including amyloidosis and fatal outcomes.
**Objectives**
To determine the frequency of BT prescription and evaluate its effectiveness and tolerability in pts with mAIDs.
**Methods**
The study included 104 pts with mAIDs followed at the V.A. Nasonova Research Institute of Rheumatology who received BT between 2013 and 2026: 50 with CAPS (42 MWS, 8 CINCA), 20 with TRAPS, and 34 with FMF. Females and males were equally represented. Median age was 19.8 [8; 28] years (range 1–63); 68 pts were children and 36 adults. Median age at disease onset was 5.8 [0.2; 7] years, and median disease duration was 16.1 [5; 21] years. Pts received IL-1, TNF, IL-6 inhibitors, and other targeted therapies.
**Results**
Before BT, 43.3% of pts had received colchicine, mainly pts with FMF, all of whom had used colchicine; about half had received GCs, more often pts with

CAPS and TRAPS. BT was prescribed to 41.8% of pts with mAIDs followed at the centre. IL-1 inhibitors were the predominant agents, used in 96.2% of pts: all pts with CAPS, 88.2% with FMF, and 80% with TRAPS. Canakinumab was used in 76 pts (73.1%) and anakinra in 24 (23.1%). Anakinra was prescribed more often in CAPS, particularly CINCA (34%), less often in FMF (17.6%), and in one pt with TRAPS (5%). TNF inhibitors were used in 14.4% of pts, mainly in FMF (29.4%), and IL-6 inhibitors in 6.7%, more often in TRAPS (15%). Other targeted therapies included secukinumab, omalizumab, and tofacitinib ($n$=1 each). Complete or partial response to BT was achieved in 83.6% of pts. Among pts receiving IL-1 inhibitors, response was observed in 76% overall and in 76%, 73.4%, and 81.3% of pts with CAPS, FMF, and TRAPS, respectively. Response to TNF inhibitors was observed in 46.6% overall, including 60% of pts with FMF and 33.3% with TRAPS. Response to IL-6 inhibitors was achieved in 57.1%; these agents were used in individual pts. Other targeted therapies were ineffective. Tolerability was satisfactory, and no BT discontinuation was required.

**Conclusion**

BT was used in a substantial proportion of pts with mAIDs followed at a federal rheumatology centre. IL-1 inhibitors were the predominant agents and showed the most favourable effectiveness profile, particularly in CAPS, FMF, and TRAPS. Complete or partial response was achieved in most pts, and treatment was generally well tolerated. TNF and IL-6 inhibitors may be considered alternative options in selected pts, mainly with FMF and TRAPS.

**Disclosure of interest**

None declared.

## P078

### Familial case series of monogenic autoinflammatory diseases complicated by AA amyloidosis

A. Sukhanina[1], S. Salugina[2], E. Fedorov[1], I. Nikishina[1,*]
[1]Pediatric Department, V. A. Nasonova Research Institute of Rheumatology, Moscow; [2]Pediatric Department, V. A. Nasonova Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** I. Nikishina
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**Introduction**

AA amyloidosis is a severe complication of monogenic autoinflammatory diseases (mAIDs) resulting from persistent uncontrolled inflammation. The highest risk is observed in untreated familial Mediterranean fever up to 60%, but remains substantial in CAPS 20–25% and TRAPS 10–25%. Amyloidosis significantly worsens prognosis due to vital organ involvement, particularly the kidneys, leading to chronic kidney disease and increased mortality risk.

**Objectives**

Description of familial cases of mAIDs with the development of AA amyloidosis.

**Methods: Family observation № 1**

Patient A., female, 14 years old, had disease onset at the age of 2 years with recurrent episodes of fever up to 39–40 °C lasting 14–30 days and recurring every 1–1.5 months. From the age of 9 years, arthralgia, skin rash, eyelid edema, conjunctivitis, and abdominal pain developed, and elevated ESR/CRP developed. A heterozygous mutation in the TNFRSF1A gene (p.Thr79Met) was identified, confirming TRAPS. Treatment with the IL-1 inhibitor canakinumab was initiated. At the age of 14 years, AA amyloidosis was confirmed by duodenal mucosal biopsy. Patient B., male, 13 years old, a third-degree cousin of patient A., had recurrent abdominal pain since early childhood and fever episodes from the age of 11 years, with elevated ESR/CRP. The same TNFRSF1A mutation was identified, confirming TRAPS. Duodenal mucosal biopsy revealed AA amyloidosis, and treatment with canakinumab was initiated. The family history included multiple cases of chronic kidney disease and AA amyloidosis across several generations, including one fatal outcome.

**Family observation № 2**

Patient C., female, 12 years old, presented from early childhood with recurrent arthritis, skin rash, and ocular hyperemia, followed by recurrent aphthous stomatitis from the age of 9 years. A heterozygous mutation in the NLRP3 gene (p.Ala441Thr) was identified, confirming CAPS (Muckle–Wells syndrome). AA amyloidosis was confirmed by duodenal mucosal biopsy. Her sister, mother, and grandmother had the same diagnosis; renal AA amyloidosis was confirmed in the grandmother at the age of 61 years. Several other relatives had similar clinical manifestations and died at a young age. All affected family members are receiving treatment with the IL-1 inhibitor canakinumab.

**Results**

In the first family, five cases of AA amyloidosis were identified, including one fatal outcome. In the second family, three biopsy-confirmed cases of AA amyloidosis were detected, including one fatal outcome; amyloidosis was suspected in two additional relatives.

**Conclusion**

Monogenic autoinflammatory diseases are associated with a high risk of AA amyloidosis, even in childhood. Careful assessment of family history is essential. Notably, in the presented patients, AA amyloidosis was morphologically confirmed in the absence of proteinuria, suggesting possible subclinical disease and highlighting the importance of early diagnosis, genetic testing, family screening, surveillance for amyloidosis, and timely targeted therapy.

**Disclosure of interest**

None declared.

## P080

### Paediatric recurrent pericarditis: clinical phenotypes, autoinflammatory features and response to IL-1 blockade in a tertiary single-centre case series

J. Torguet Carbonell[1,*] on behalf of Pediatric Rheumatology Unit, Estefanía Moreno[1], Mireia López[1]
[1]Paediatric Rheumatology Unit, Rheumatology Department, Vall d'Hebron University Hospital, Barcelona, Spain
**Correspondence:** J. Torguet Carbonell
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**Introduction**

Paediatric recurrent pericarditis (PRP) is an uncommon inflammatory condition including idiopathic, post-cardiac injury and autoinflammatory phenotypes. Increasing evidence suggests a central role of innate immunity and IL-1-mediated inflammation, particularly in steroid-dependent or refractory disease.

**Objectives**

To describe clinical characteristics, etiological workup, treatment strategies and outcomes in a paediatric recurrent pericarditis cohort, focusing on IL-1 blockade.

**Methods**

Retrospective single-centre study including 7 paediatric patients followed between 2018 and 2026. Clinical presentation, inflammatory markers, autoimmune and genetic studies, treatment exposure, duration, relapses and outcomes were analysed.

**Results**

Seven patients (5 females) with disease onset between 6 and 13 years were included (median age 11 years) (Table 1). Five presented an idiopathic/autoinflammatory phenotype, including one familial case and one associated with humoral immunodeficiency. Two developed post-cardiac injury syndrome after congenital heart surgery. One evolved to chronic constrictive pericarditis requiring pericardiectomy. All patients experienced recurrent inflammatory flares with elevated acute phase reactants. Autoimmune studies were negative in all cases, and autoinflammatory genetic panels were non-contributory in tested patients. All received NSAIDs and/or colchicine. Five required systemic corticosteroids because of recurrent or steroid-dependent disease. Five patients received anakinra for refractory disease, with marked clinical and analytical improvement and significant steroid-sparing effect. Two achieved sustained remission after withdrawal, while one relapsed during tapering.

**Conclusion**

PRP represents a heterogeneous inflammatory spectrum ranging from post-cardiac injury syndromes to probable autoinflammatory disease. Steroid dependence and relapses during tapering were frequent. IL-1 blockade was effective in most patients, supporting the role of innate immune dysregulation in PRP. Early recognition of refractory phenotypes may help prevent complications such as constrictive pericarditis.

**Disclosure of interest**

None declared.

**Table 1 (Abstract P080)** Clinical features and outcomes

| Sex/Age | Phenotype | Treatment (no anti-IL1) | Anti-IL1, treatment duration | Outcome |
|---|---|---|---|---|
| M, 13y 5 m | Idiopathic / autoinflammatory | Corticosteroids + colchicine | 1.5 mg/Kg/day, ongoing (6 m) | Mild relapse during tapering, clear response to Anakinra |
| F, 10y 1 m | Idiopathic / autoinflammatory | NSAIDs + corticosteroids | 1 mg/Kg/day, ongoing (6 m) | Sustained remission, steroid-dependent disease, excellent response to IL-1 blockade |
| M, 13y | Idiopathic / autoinflammatory | NSAIDs + colchicine | No | Familial PRP associated with repaired congenital heart disease; good response to colchicine |
| M, 11y | Idiopathic / autoinflammatory | Corticosteroids | 2 mg/Kg/day, 20 m | Constrictive pericarditis, pericardiectomy |
| F, 6y | Post-surgical (ASD repair) | Corticosteroids + colchicine | 2 mg/Kg/day, 17 m | Steroid-dependent, sustained remission on anakinra |
| F, 11y | Idiopathic / autoinflammatory +immunodeficiency | Colchicine | 1 mg/Kg/day, 3y | Good response |
| F, 6y 10 m | Post-surgical + Kabuki syndrome | NSAIDs + colchicine | No | Favorable evolution |

P081

## Hemophagocytic lymphohistiocytosis secondary to Epstein-Barr virus infection in a patient with Aicardi-Goutières syndrome - a case report

K. Kapten[1,*], J. Roszkiewicz[1], E. Smolewska[1]
[1]Pediatric Cardiology and Rheumatology, Medical University of Lodz, Lodz, Poland
**Correspondence:** K. Kapten
*Pediatric Rheumatology 2026*, **24(S1):**P081

### Introduction
Secondary hemophagocytic lymphohistiocytosis (HLH) is a life-threatening cytokine-mediated inflammatory syndrome, most often triggered by infection. Its development in a patient with Aicardi–Goutières syndrome (AGS), an early-onset progressive, monogenic inflammatory encephalopathy resulting in the overproduction of type I interferon, is a rarity not yet described in literature.
### Objectives
This case report aimed to present diagnostic challenges of the possible clinical overlap between interferonopathies and HLH, as well as difficult therapeutic considerations that extend beyond standard protocols. A. is a four-year-old girl diagnosed with AGS type 5 with a genetically confirmed heterozygotic mutation in the gene SAMHD1 with a severe neurological presentation of the disease, including microcephaly, developmental delay, and spasticity. As targeted therapies may improve some of the disease manifestations (1), A. has been receiving baricitinib, an oral selective Janus kinase inhibitor, since January 2025. During that time, severe infections were observed, including pneumonia, which led to hospitalization and deterioration in the patient's clinical state. After excluding malignancy in the bone marrow biopsy, A. was transported to the Department of Rheumatology in a bad clinical state. In addition to neurological impairment, the patient exhibited significant livedo reticularis and frostbite-like skin lesions. Laboratory results showed anemia, thrombocytopenia, coagulopathy with hypofibrinogenemia, and hypoalbuminemia, as well as elevated ferritin, lactate dehydrogenase, and sCD25 levels, recognized hallmarks of HLH. Serological testing revealed antibodies for EBV, and EBV DNA was detected by polymerase chain reaction. The patient needed multiple platelet transfusions, as well as cryoprecipitate, fibrinogen, and albumin supplementation. Initiation of high-dose intravenous methylprednisolone yielded no effect. On a multidisciplinary team consultation, the rituximab treatment protocol was commenced, followed by the administration of intravenous immunoglobulins (IVIG) and tapering doses of steroids. A significant improvement in the patient's clinical condition was observed, accompanied by a gradual normalization of laboratory parameters.
### Conclusion
A secondary HLH in patients with interferonopathies should be suspected if the typical laboratory findings are present, especially following infections like EBV. Targeted immunomodulatory therapy, such as rituximab, and adjunctive IVIG can be a successful treatment option.
### Methods
The authors present a rare case of a four-year-old girl, A., with AGS who developed HLH secondary to Epstein-Barr Virus (EBV) infection following the treatment with baricitinib. The patient has provided informed consent for the publication of their case report.

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### Disclosure of interest
None declared.

P082

## Monogenic mimics of Behçet's disease in childhood: a single-centre retrospective study

L. De Nardi[1,2,*], S. Federici[1], C. Celani[1], C. Perrone[3], C. Passarelli[3], F. De Benedetti[1], A. Insalaco[1]
[1]Rheumatology Department, IRCCS Ospedale Pediatrico Bambino Gesù; [2]Department of Systems Medicine, University of Roma 'Tor Vergata'; [3]Medical Genetics, IRCCS Ospedale Pediatrico Bambino Gesù, Rome, Italy
**Correspondence:** L. De Nardi
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### Introduction
Monogenic autoinflammatory diseases (AIDs) may present early in life with clinical features mimicking Behçet's disease(BD), posing major diagnostic challenges.
### Objectives
To characterize the clinical and genetic features of paediatric patients referred for suspected BD and assess the contribution of monogenic disorders.
### Methods
This single-centre retrospective study included paediatric patients referred for suspected BD. Next-generation sequencing(NGS) panels for AIDs or whole-exome sequencing(WES) were performed in patients with disease onset before 8 years and/or a positive family history of Behçet-like disease or autoimmunity.
### Results
Thirty-six out of 40 children underwent genetic testing(58.3% NGS panel, 41.7% WES). All patients were Caucasian, 35% female, with a

median disease onset of 5 years (IQR 2–9.5). A positive family history of autoimmunity or BD was reported in 53% of cases. Recurrent oral aphthosis occurred in 95% of patients (38/40), while recurrent fever in 68%(27/40), including SURF phenotype in 30%(12/40). Genital ulcers were reported in 35%(14/40), cutaneous manifestations in 53%(21/40) and ocular involvement in 15%(6/40). Arthritis and arthralgia were observed in 30% and 45% of patients, respectively. Gastrointestinal involvement resembling inflammatory bowel disease (IBD) was registered in 35%(14/40), whereas thrombosis/thrombophlebitis occurred in 18%(7/40). Additional manifestations included myocarditis/pericarditis (2/40), cytopenia (3/40), central nervous system involvement (4/40), myositis (3/40), and lymphadenopathy (13/40). Twelve patients(30%) fulfilled the PEDBD 2015 criteria and 20 (50%) met the ICBD 2014 criteria. HLA-B51 was positive in 6 of 16 tested patients. Pathogenic or likely pathogenic variants (ACMG classes 4-5) were identified in 22% of patients(8/36), involving*TNFAIP3* (*n*=2), *NFKB1* (*n*=1), *SOCS1* (*n*=2), mosaic trisomy 8 (*n*=1), *IFNAR2* (*n*=1), *RELA* (*n*=1). Variants of uncertain significance (ACMG class 3) in AID-related genes were detected in an additional 24 patients (66.7%). These included 2 de novo variants in *NLRP1* (*n*=1) and *TNFRSF1A* (*n*=1), two ELF4 hemizygosity, and 12 variants inherited from parents affected by AID-like phenotypes. Such variants involved *TNFAIP3* (*n*=3), *RELA* (*n*=1), *PLCG2* (*n*=2), *ERAP1* (*n*=1), *PSTPIP1* (*n*=1), *LPIN2* (*n*=1), *NFKB1* (*n*=1), *WDR1* (*n*=1) and*TNFRSF1A* (*n*=1), further supporting a genetic contribution in a substantial proportion of cases.

**Conclusion**

Monogenic disorders represent a significant and likely underrecognized cause of Behçet-like disease in childhood. The high prevalence of variants in genes previously associated with Behçet-like phenotypes highlights the importance of early genetic screening, particularly in patients with disease onset before 8 years of age and/or a positive family history of Behçet-like disease or autoimmunity.

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**Trial registration identifying number**

Not applicable.

**Disclosure of interest**

None declared.

## P084

### Genetic investigation of atypical PFAPA cases in a tertiary hospital: a five-year retrospective study

M. Michailou[1,*], M. Tsaroucha[1], V. Giatraki[1], E. Paraskakis[1], C. Perdikogianni[1]
[1]Department of Pediatrics, University Hospital of Heraklion, Heraklion, Greece
**Correspondence:** M. Michailou

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**Introduction**

PFAPA (periodic fever, aphthous stomatitis, pharyngitis, adenitis) syndrome is a benign and often underdiagnosed entity. Although the genetic basis of PFAPA remains unclear, clinical and genetic overlap with monogenic autoinflammatory syndromes has increasingly been recognized.

**Objectives**

To evaluate the frequency of PFAPA among children investigated for periodic fever and to characterize the clinical and genetic features of atypical PFAPA cases.

**Methods**

This retrospective study included all patients that were admitted in the Pediatric Clinic or followed at the Pediatric Immunology Outpatient Clinic from 12/2020 to 05/2026 due to periodic fever.

**Results**

Out of the 83 patients that were investigated for periodic fever in the past five years, a total of 59 patients were diagnosed with PFAPA based on clinical criteria. Genetic testing was performed in 42,3% (25/59) due to atypical presentation such as late age of onset, abdominal or thoracic pain, rash and diarrhea (Table 1). Fourteen patients (14/25, 56%) carried at least one gene variant associated with periodic fever syndromes and autoinflammatory disorders such as MEFV (*n*=6/14, 42,8%), TNFRSF11A (*n*=2), CARD 14, IL-10, IL10R4, SAA1.2, SLC7A9, PLG2 and AP1S3 (*n*=1). The most common MEFV variant was M694V (3/6, 50%). In our set of patients, atypical symptoms were most frequently encountered in the non-PFAPA patients (RR:2.24 for any atypical symptom, 95% CL 1.10-4.56 *p*=0.029, RR:2.88 for abdominal pain,95% CL 1.07-7.66 *p*=0.043).

**Table 1 (Abstract P084)** Clinical characteristics of the cohort of patients

| | PFAPA | non-PFAPA | *p* |
|---|---|---|---|
| Age, years (mean + SD) | 4.661+2.99 | 7.833+4.66 | <0.01 |
| Gender (M/F) | 26/33 | 16 /9 | 0.091 |
| Total number of patients | 59 | 24 | |
| Atypical Symptoms | 11 | 10 | **0.029** |
| (+) Family History | 12 | 3 | 0.403 |
| Abdominal Pain | 6 | 7 | **0.045** |
| Thoracic Pain | 0 | 1 | 0.289 |
| Rash | 3 | 1 | 1 |
| Diarrhea | 5 | 1 | 0.667 |

**Conclusion**

These findings support a possible association between PFAPA and variants in genes implicated in monogenic autoinflammatory disorders. However, the high frequence of MEFV carriage in Crete should be considered for the interpretation of our results. Larger multicenter studies are needed to further evaluate this association.

**Disclosure of interest**

None declared.

## P085

### Subclinical joint involvement in pediatric patients with familial Mediterranean fever

M. Tsaroucha[1,*], E. Drakonaki[2], M. Michailou[1], C. Perdikogianni[1]
[1]Department of Pediatrics, University Hospital of Heraklion; [2]Department of Anatomy, Medical School, University of Crete, Heraklion, Greece
**Correspondence:** M. Tsaroucha

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**Introduction**

Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease in childhood. Subclinical inflammation plays a crucial role in the long-term progression of FMF contributing to complications.

**Objectives**

To investigate the frequency of subclinical joint inflammation in pediatric patients with Familial Mediterranean fever in remission.

**Methods**

Eight pediatric patients with genetically confirmed FMF were evaluated for subclinical joint involvement using high resolution musculoskeletal ultrasound. Eligible participants were patients diagnosed before the age of 18 years who had remained attack-free and asymptomatic during the preceding year. The assessed joints included hips, knees, ankles, subtalar joints, metatarsophalangeal (MTP) joints, elbows, wrists, metacarpophalangeal (MCP) joints, proximal interphalangeal (PIP) joints, and distal interphalangeal joints.

**Results**

All patients demonstrated subclinical findings of arthritis in at least one joint. Findings included mild effusion in knees (5/8), ankles (4/8), elbows (1/8), wrists (4/8) MTPs (8/8) and finger joints (3/8 at least 1 MCP or PIP). Synovial thickening was present in knees (3/8), ankles (1/8), MTPs (5/8), wrist (2/8). Mild synovial vascularity was evident in 2 patients in knees and ankle joints bilaterally, whereas in 2/8 patients, unilateral Gruberi bursitis was found. No findings of arthritis were evident in hips and subtalar joints. All patients were adequately controlled with colchicine therapy and had negative inflammation markers (ESR, CRP).

**Conclusion**

Mild effusion and synovial thickening may be detected using US in asymptomatic peripheral joints of patients with FMF in remission. The study highlights the importance of longitudinal monitoring of these patients to determine the prognostic implications of subclinical joint findings.

**Disclosure of interest**
None declared.

## P086

### Secukinumab in juvenile idiopathic arthritis–associated uveitis: results of open-label phase II study

A. V. Ramanan[1,2,*] on behalf of The TURTLE Investigators , A. D. Dick[3,4], T. Jaki[5,6], J. Choi[7], K. Armon[8], H. Petrushkin[9], A. P. Jones[10], J. Green[10], B. Hardwick[10], S. Drake[10], C. Guly[11], M. W. Beresford[12,13]
[1]Bristol Royal Hospital for Children; [2]University of Bristol, Bristol; [3]NIHR Moorfields BRC; [4]Institute of Ophthalmology, University College London, London, UK; [5]University of Regensburg, Regensburg, Germany; [6]University of Cambridge, Cambridge; [7]Sheffield Children's NHS Foundation Trust, Sheffield; [8]Cambridge University Hospitals NHS Foundation Trust, Cambridge; [9]Great Ormond Street Hospital for Children NHS Foundation Trust, London; [10]Liverpool Clinical Trials Centre, Liverpool; [11]Bristol Eye Hospital, Bristol; [12]Alder Hey Children's Hospital NHS Foundation Trust; [13]The University of Liverpool, Liverpool, UK
**Correspondence:** A. V. Ramanan
*Pediatric Rheumatology 2026*, **24(S1):**P086

**Introduction**
Juvenile idiopathic arthritis (JIA) is the most common paediatric rheumatic disease, with uveitis affecting 15–25%, risking vision loss [1, 2]. Adalimumab plus methotrexate (MTX) improves outcomes, but ~27% do not respond [3,4]. Interleukin–17A (IL–17A) is implicated in its pathogenesis [5,6]. Secukinumab (anti–IL–17A) is effective in adult uveitis, though subcutaneous trials missed primary endpoints [7,8]. Limited paediatric data support a Phase II Bayesian approach [9,10].
**Objectives**
Primary: 12-week ocular response per SUN criteria (two-step decrease or zero) [9].
Secondary: Safety and tolerability.
**Methods**
This trial (EudraCT: 2022-003068-26) used a two-stage design. Stage-1 enrolled 10 adalimumab-refractory participants receiving subcutaneous secukinumab plus MTX/MMF; ≥3 responders at week-12 were required. If met, Stage-2 would randomise 40 participants to secukinumab or adalimumab using Bayesian framework with expert-derived priors [10]. Primary analysis was intention-to-treat. Secondary outcomes used standard frequentist methods, with sensitivity analyses for missing data.
**Results**
Five males/females each were each recruited: median age 10.0 years (IQR 7.0–13.0). Most weighed 25–50 kg (60%; 30% >50 kg; 10% <25 kg). Table 1 shows anterior chamber cell outcomes (SUN score) and response (+/- or not done/applicable). One of ten met 12–week primary endpoint, with no new safety findings. Forty-three non-serious adverse events occurred in 8/10 participants; none were new or unexpected. One serious adverse event (SUSAR) occurred in 1/10 participants.
**Conclusion**
Secukinumab showed limited efficacy, failing to meet progression threshold. Findings do not support further investigation without changes to population.

**Table 1 (Abstract P086)** Twelve-week response of anterior chamber cells (SUN) grade

| Number | Eligible eye(s) | Baseline (L/*R*) | Week-12 (L/*R*) | Step change (L/*R*) | Responded |
|---|---|---|---|---|---|
| 1 | Both | 1+/2+ | 2+/2+ | 1/0 | No |
| 2 | Both | 1+/1+ | 2+/2+ | 1/1 | No |
| 3 | Left | 2+/– | */– | */– | No |
| 4 | Left | 3+/– | 2+/– | -1/– | No |
| 5 | Left | 2+/– | */– | */– | No |
| 6 | Right | –/4+ | –/* | –/* | No |
| 7 | Left | 1+/– | 1+/– | 0/– | No |
| 8 | Left | 1+/– | 0/– | -2/– | Yes |
| 9 | Both | 3+/3+ | 2+/2+ | -1/-1 | No |
| 10 | Both | 3+/3+ | 1+/2+ | -2/-1 | No |

*Participants discontinuing before week-12 classified as non-responders

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## P087

### Characteristics and treatment outcomes of paediatric patients with chronic idiopathic uveitis in a tertiary centre

F. Pitet[1,*], C. Vinit[1], M. Labouret[1], G. Dingulu[1], A.-T. Nguyen[1], A. Faye[1], A. Clément[2], C. Titah[2], E. Bui Quoc[3], U. Meinzer[1], C. Dumaine[1]
[1]General Pediatrics, Infectious Disease and Internal Medicine Department, Robert Debre Hospital; [2]Department of Ophthalmology, Hôpital Fondation Adolphe de Rothschild; [3]Department of Ophthalmology, Robert Debre Hospital, Paris, France
**Correspondence:** F. Pitet
*Pediatric Rheumatology 2026*, **24(S1):**P087

**Introduction**
Chronic non-infectious paediatric uveitis is a rare but major cause of acquired visual acuity loss. Those related to juvenile idiopathic arthritis (JIA) are the most common and best studied, but there is less literature on idiopathic uveitis.
**Objectives**
The aim was to describe the clinical characteristics and treatment outcomes of idiopathic uveitis in children at our tertiary centre. Secondary objectives were to study the duration of treatment with oral corticosteroids according to the type of uveitis and associated sparing treatments.
**Methods**
This is a single-centre retrospective study covering all children diagnosed with idiopathic uveitis before the age of 18 who required at least one consultation or hospitalisation at Robert Debré Hospital, between August 2008 and May 2024. The data collected included clinical, biological, anatomopathological, ophthalmological, extra-ophthalmological and therapeutic information.
**Results**
We included 113 patients, with a total of 195 affected eyes. The median age at diagnosis was 10 years, and the median follow-up duration was 30 months. Uveitis was bilateral in 73% of patients, with 43% of cases being granulomatous. The location was anterior in 34% of cases, panuveitis in 34%, intermediate in 23% and posterior in 4%. 58% of patients had at least one extraocular symptom. 79% of patients had an ophthalmological complication at diagnosis or during follow-up, including loss of visual acuity, papillary oedema, or retinal vasculitis. Systemic corticosteroid therapy was used in 73% of patients, with a median duration of oral corticosteroid therapy of 8 months. Among other treatments, methotrexate was used in 83 patients with a median duration of 21 months, and biotherapy was used in 47% of cases. At the last follow-up, nearly half of the patients had inactive uveitis (47%), and 24% were considered to be in remission. However, 68% of all patients were still undergoing treatment at the last visit.

**Conclusion**
These data provide impetus for future prospective cohort studies and the development of treatment protocols, with a particular focus on reducing corticosteroid use.
**Disclosure of interest**
None declared.

## P088

### An audit of pediatric uveitis management in 5 network centres across Scotland

M. Tavendale[1], R. Oswald[2], B. Oates[3], J. Bamber[4], C. Lees[5], C. King[1], J. Steel[5], S. Hardy[5], D. Knight[6], A. Brown[7], M. Varikkara[8], A. Baxter[7], S. Hicks[9], N. Martin[10,*]
[1]Royal Hospital for Children, Glasgow; [2]Ninewells Hospital, Dundee; [3]Paediatrics, Crosshouse University Hospital, Kilmarnock; [4]Dumfries and Galloway Royal Infirmary, Dumfries; [5]Wishaw General Hospital, Wishaw; [6]Opthalmology, Raigmore Hospital, Inverness; [7]Opthalmology, Wishaw General Hospital, Wishaw; [8]Opthalmology, Crosshouse University Hospital, Kilmarnock; [9]Paediatrics, Raigmore Hospital, Inverness; [10]Rheumatology, Royal Hospital for Children, Glasgow, UK
**Correspondence:** N. Martin
*Pediatric Rheumatology 2026*, **24(S1):**P088

**Introduction**
Since 2010 Pediatric Rheumatology network centres in Scotland have followed a national guideline for treating uveitis aiming to ensure consistent approach and improve visual outcomes.
**Objectives**
To assess adherence to Scottish Uveitis Network (SUN) guideline in secondary care centres and compare centres with combined rheumatology-ophthalmology clinics with those that hold separate clinics.
**Methods**
Data from pediatric patients diagnosed with uveitis in 5 secondary care hospitals between 2012 and 2023 were compared with 5 key recommendations from the SUN guideline. (1) Patients with macular oedema should receive systemic steroids and start a DMARD. (2) Topical steroids for anterior uveitis should be weaned within 4 months. If uveitis not controlled or recurs patients should start a DMARD. (3) Failure to control uveitis and stop topical or systemic steroids within 12 weeks of starting DMARD should lead to starting Infliximab or Adalimumab. We recorded each patient's visual acuity at their first and most recent visit and defined poor visual outcome as developing cataract or visual acuity ≥0.3 on LogMAR scale at most recent visit as this is the minimum requirement to hold a driving license in the UK.
**Results**
Of 88 patients 41 were male. 48 had JIA associated uveitis, 37 idiopathic uveitis and 3 had other causes of uveitis. 8/14 with macular oedema at diagnosis were treated with systemic steroids and DMARD. 33/45 started DMARD if unable to stop topical steroids within 4 months. 33/57 started TNFi if unable to stop topical steroids within 12 weeks of starting DMARD. There was no statistically significant difference between adherence in centres with combined versus separate clinics. Methotrexate or MMF were used as first line DMARD in 69 of 70 patients. Adalimumab or Infliximab were used as first line biologic in 60 of 66 patients. Alternative first line biologics were used in 2 cases due to close relatives with demyelinating neuropathy. 6 eyes had poor visual outcomes. 1 had steroid induced cataract, 5 had corrected visual acuity ≥0.3 on LogMAR scale at most recent visit. There were no poor visual outcomes at centres with combined clinics.
**Conclusion**
This multi centre audit from non-tertiary centres within a national paediatric rheumatology network showed improved visual outcomes compared with historical cohorts. Compliance with SUN guidelines was not significantly different between centres holding combined or separate clinics. Some patients were still exposed to prolonged courses of topical steroids with delays in starting DMARD or TNFi. Where combined clinics are not practical, optimising communication between Ophthalmology and Rheumatology teams may be an effective alternative to reduce delays in starting systemic treatment. These results will be used to inform the update of national uveitis guidelines and delivery of pediatric uveitis care in Scotland.
**Disclosure of interest**
None declared.

## P089

### Endothelial dysfunction and clinical predictors of uveitis in children with rheumatic diseases: development of a multivariable risk model

I. Shevchenko[1,*], N. Koshel[2], O. Oshlianska[1]
[1]Department of Pediatrics, Neonatology, Pediatric Infectious Diseases, Immunology and Allergy, Shupyk National Healthcare University of Ukraine; [2]Laboratory of Mathematical Modeling of Aging Processes, State Institution "Institute of Gerontology named after D. F. Chebotaryov of the National Academy of Medical Sciences of Ukraine", Kyiv, Ukraine
**Correspondence:** I. Shevchenko
*Pediatric Rheumatology 2026*, **24(S1):**P089

**Introduction**
Uveitis is a severe and potentially vision-threatening complication in children with rheumatic diseases, particularly juvenile idiopathic arthritis (JIA). Current predictive approaches rely mainly on immunological markers and demonstrate limited accuracy. The role of endothelial dysfunction in uveitis pathogenesis remains insufficiently explored.
**Objectives**
To evaluate endothelial function in children with uveitis, assess its association with disease activity and quality of life, and develop a multivariable predictive model for uveitis risk.
**Methods**
A cross-sectional observational study (2022–2025) included 113 children divided into four groups: uveitis associated with rheumatic diseases (URD, $n$=26), idiopathic uveitis (IU, $n$=12), rheumatic diseases without uveitis (RD, $n$=31), and healthy controls ($n$=31). Endothelial function was assessed using flow-mediated dilation (FMD) of the brachial artery. Disease activity was evaluated using Juvenile Arthritis Disease Activity Score (27 joints), (JADAS27), Standardization of Uveitis Nomenclature (SUN), National Eye Institute, (NEI), and Uveitis Activity Score, (UAS) criteria. Quality of life was assessed with Childhood Health Assessment Questionnaire, (CHAQ), Disabilities of the Arm, Shoulder, and Hand, (DASH), and National Eye Institute 25-item Visual Function Questionnaire, (NEI VFQ-25). A multivariable logistic regression model was constructed.
**Results**
Endothelial dysfunction was more pronounced in children with uveitis, with significantly reduced flow-mediated dilation (FMD) (0.87±3.47% in idiopathic uveitis and 0.12±3.9% in uveitis associated with rheumatic diseases vs. 9.13±2.53% in JIA without uveitis and 14.6±2.9% in healthy controls; $p$<0.05). Diastolic blood flow parameters were lower in uveitis patients (EDV 2.63±0.14 vs. 3.1±0.08 mm; TAMX 18.94±3.04 vs. 27.09±2.8 cm/s; $p$<0.05), with predominance of hypoergic (28.94%) and paradoxical (31.57%) responses compared to normoergic responses in controls (79.54%). Hemoglobin levels were significantly lower in the uveitis group ($p$=0.008). Uveitis activity correlated negatively with quality of life scores (NEI VFQ-25: $r$=−0.73; $p$=0.007; DASH: $r$=−0.79; $p$=0.002). The predictive model demonstrated good performance (AUC=0.867; 95% CI 0.751–0.943), with sensitivity of 80.8% and specificity of 80.6%.
**Conclusion**
The proposed multivariable model demonstrates good predictive accuracy and represents a practical tool for early risk stratification. Integration of FMD assessment into clinical practice may enable earlier detection of uveitis and improve patient management.
**Disclosure of interest**
I. Shevchenko: None declared, N. Koshel Employee with: No conflict of interest, O. Oshlianska Employee with: No conflict of interest.

## P090

### Pediatric tubulointerstitial nephritis and uveitis: single-center case series

S. Correia[1], R. S. Pereira[1], J. B. Lima[2,*], L. Salazar[3], T. Costa[3], M. S. Faria[3], V. Miranda[4], S. Alves[2], C. Zilhão[2]
[1]Pediatric Department, Unidade Local de Saúde de Trás-os-Montes e Alto Douro, Vila Real; [2]Pediatric Rheumatology Unit; [3]Pediatric Nephrology Department, Centro Materno Infantil do Norte, Unidade Local de Saúde de Santo António; [4]Ophthalmology Department, Unidade Local de Saúde de Santo António, Porto, Portugal
**Correspondence:** J. B. Lima
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**Introduction**
Tubulointerstitial nephritis and uveitis (TINU) syndrome is a rare immune-mediated disorder characterized by the coexistence of tubulointerstitial nephritis and uveitis [1]. Diagnosis can be challenging due to the asynchronous onset of renal and ocular manifestations and the need to exclude alternative causes. Although TINU accounts for roughly 2% of all uveitis, it appears to be more common in children [1].
**Objectives**
Characterize the clinical course and outcomes of children diagnosed with TINU syndrome.
**Methods**
Retrospective review of pediatric TINU cases, diagnosed between 2015 and 2024, assessing clinical presentation, organ involvement, treatment and outcomes at diagnosis and 24 months.
**Results**
Seven patients were included, with a median age of 14.6 years (IQR 13.4–16.7). Uveitis was the initial manifestation in 5 patients and tubulointerstitial nephritis in 2 (interval between manifestations of 0–2.8 months). All patients had symptomatic anterior uveitis (2 unilateral, 5 bilateral). Renal impairment was present at diagnosis in 5 cases, with a median initial eGFR of 68 mL/min/1.73 $m^2$ (Bedside Schwartz Equation). Two patients underwent kidney biopsy. Treatment included topical/systemic corticosteroids and methotrexate, with biologic therapy used in 3 cases. At 24 months, renal function was preserved in all patients (median eGFR 97 mL/min/1.73 $m^2$), although 2 had persistent active uveitis. Patient-level data are summarized in Table 1.
**Conclusion**
Despite the small cohort, this study highlights the heterogeneous presentation of pediatric TINU, including silent renal involvement. Uveitis was the main presenting feature, reinforcing the need for renal–ophthalmologic screening and surveillance. Elevated urinary β2-microglobulin may be a useful marker of tubular involvement, even with preserved renal function. Persistent ocular activity in 2 patients underscores the importance of individualized long-term multidisciplinary follow-up to prevent irreversible damage.

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**Disclosure of interest**
None declared.

P091
**Therapeutic drug monitoring of adalimumab in paediatric non-infectious uveitis: a retrospective analysis**
J. Dehoorne[1,2,*], T. Renson[1,2], C. Bonroy[3], C. Sys[4], C. Park[5], L. De Coninck[1], E. Kreps[6,7]
[1]Department of Internal Medicine and Pediatrics, Department of Pediatric Rheumatology, University Hospital Gent; [2]European Reference Network for Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases, Gent University Hospital; [3]Department of Diagnostic Sciences, Ghent University; Department of Laboratory Medicine, University Hospital Gent, Gent; [4]Department of Ophthalmology, AZ Sint Jan, Brugge; [5]Department of Ophthalmology; [6]Department of Ophthalmology and Center for Medical Genetics Ghent, University Hospital Gent; [7]European Reference Network for Rare Eye Diseases at the Ghent University Hospital, Gent, Belgium
**Correspondence:** Joke Dehoorne
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**Introduction**
Adalimumab is currently considered the most efficacious anti-TNFα agent for childhood noninfectious uveitis (NIU). However, a substantial number of patients have a suboptimal response.Therapeutic drug monitoring (TDM) using measurement of adalimumab trough level (TL) and anti-adalimumab antibody (AAA) level holds potential to optimize management.
**Objectives**
To evaluate the usefulness of therapeutic drug monitoring (TDM) of adalimumab in children with chronic non-infectious uveitis (NIU).
**Methods**
A retrospective cohort study was conducted at the Department of Ophthalmology and Paediatric Rheumatology of Ghent University Hospital, Belgium. Children with chronic NIU diagnosed under the age of 16, treated with adalimumab, with (group 1) or without (group 2) concomitant immunomodulating treatment (IMT), with at least one available serum adalimumab trough level (TL) and anti-adalimumab antibody (AAA) level were included. Adalimumab TL were quantified using a bridging ELISA, which measures concentrations of active drug (ApDia Adalimumab kit, reference 710201) and free AAAs were detected using a drug-sensitive bridging ELISA (ApDia anti–Adalimumab kit, ref 710301). Controlled uveitis was defined as an anterior chamber (AC) activity ≤0.5 cells on <2 steroid eye drops per day. Clinical response at the time of TDM and subsequent therapeutic decisions were monitored.

**Table 1 (Abstract P090)** Baseline characteristics, treatment and 24-month outcomes

| Case | Sex | Age (years) | Initial Manifestation | Time to 2nd manifestation (months) | Initial eGFR (mL/min/1.73 $m^2$) | uB2M | Treatment | Uveitis 24 mo | eGFR 24 mo (mL/min/1.73 $m^2$) |
|---|---|---|---|---|---|---|---|---|---|
| 1 | F | 14.6 | Unilateral AU | 0.7 | 70 | High | TCS, MTX | No | 64 |
| 2 | F | 15.6 | Bilateral AU | 0.8 | 92 | High | TCS, SCS, MTX, ADA | No | 85 |
| 3 | F | 14.1 | Nephritis | 0.8 | 63 | High | TCS, SCS, MTX | No | 83 |
| 4 | F | 13.4 | Bilateral AU | 0.0 | 60 | High | TCS, SCS, MTX, ADA, IFX | Yes | 109 |
| 5 | M | 10.7 | Nephritis | 2.8 | 22 | High | SCS, MTX | No | 130 |
| 6 | M | 16.7 | Bilateral AU | 1.5 | 68 | N/A | TCS, SCS, MTX, ADA | Yes | 131 |
| 7 | F | 17.7 | Bilateral AU | 2.3 | 151 | High | TCS, SCS, MTX | N/A | N/A |

AU, anterior uveitis; uB2M, urinary β2-microglobulin; eGFR, estimated glomerular filtration rate; MTX, methotrexate; TCS, topical corticosteroids; SCS, systemic corticosteroids; ADA, adalimumab. IFX, infliximab; N/A, non-applicable

**Results**
A total of 37 children (73 eyes) with NIU were included, with a median age at diagnosis of 7.4 years (range 2.2-13.8). Most patients had anterior uveitis (73%). Adalimumab was administered for a median duration of 29 months (range 5-79) (cumulative 103.5 person-years). During this period, 200 TL were measured, with a median concentration of 13.4 µg/ml (range 0 – 33), and TDM-based therapeutic adjustments were performed in 22 patients (59.5%). In group 1 (*n*=28), treatment escalation was performed in 9 of 28 patients (32.1%), which resulted in increased adalimumab levels (median +4.1 µg/ml, range [-0.7; +15.6]) in 7 patients (87.5%), and regained uveitis control in 5 of 8 patients (62.5%). In group 2 (*n*=9), 4 patients developed primary treatment failure, with TL <0.5 µg/ml and elevated AAA levels (range 29.4–181 µg/ml). In 3 of the other 5 patients, treatment adjustments were made, including TDM-guided escalation in 1, taper in 1 and cessation of treatment in 1 patient as TL was 0 despite clinical inactivity.
**Conclusion**
In children with chronic uveitis, TDM of adalimumab is a promising tool to optimise the clinical outcome and limit treatment related side effects.
**Disclosure of interest**
None declared.

## P093

### Managing the unseen: an insight into treatment challenges in JIA-associated uveitis

N. Pai[1,*], M. Chatten[1], I. Nengroo[1], A. Pradeep[2], S. Rangaraj[1], K. Warrier[1]
[1]Paediatric Rheumatology; [2]Paediatric Opthalmology, Nottingham University Hospitals NHS Trust, Nottingham, UK
**Correspondence:** N. Pai
*Pediatric Rheumatology 2026*, **24(S1):**P093

**Introduction**
Juvenile Idiopathic Arthritis–associated Uveitis (JIA-U) is the most common extra-articular manifestation of JIA. It typically presents as asymptomatic chronic anterior uveitis, making diagnosis challenging in children. Early identification, timely initiation of therapy and close monitoring are essential to optimize long-term visual outcomes and reduce morbidity.
**Objectives**
To describe the demographics, complications, treatment patterns, and disease burden among paediatric patients with JIA-U.
**Methods**
A retrospective study was conducted from digital health records. 30 patients with JIA-U managed jointly by the paediatric ophthalmology and rheumatology teams at Queen's Medical Centre between 2023 and 2025 were identified.
**Results**
JIA-U predominantly affected females (22/30) and patients with oligoarticular JIA (23/30), with bilateral involvement observed in most cases (18/30). The median age at diagnosis for both JIA and uveitis was 6 years. 16.6% of patients initially presented with uveitis prior to the diagnosis of JIA. Drug-induced remission was achieved in 70% of patients (21/30) after a median duration of 42 months. Sustained remission beyond three years was achieved in only nine patients (40%). Five patients underwent a trial off therapy after a median of 45 months - all relapsed within six months. Complications at presentation were identified in 10 patients (42%). Prolonged use of topical corticosteroid (>3 months) was required in 43% patients and was associated with complications, including ocular hypertension and cataract. 47% patients needed systemic corticosteroid therapy. Most patients (27/30) required combination therapy with conventional synthetic disease-modifying antirheumatic drugs (cs-DMARDs) and biologic DMARDs (b-DMARDs), particularly those with bilateral disease. Methotrexate was used as standard first-line therapy. Despite combination treatment, almost half (13/27) the patients experienced disease flares, of whom 10/13 required a second biologic agent. Uveitis was severe in 17 patients and was more common in ANA-positive patients with bilateral disease. These patients had a high treatment burden, with 16/17 requiring combination therapy and one requiring triple Immunosuppressive therapy. No patients were discharged from follow-up during the study period. Seven patients transitioned to adult services, all of whom had been in remission for at least one year prior to transfer.
**Conclusion**
- JIA-U in this cohort demonstrated a chronic, relapsing disease course with substantial long-term treatment burden requiring prolonged systemic immunosuppression, often requiring multiple agents.

- Female sex, oligoarticular subtype, younger age at onset, delayed referral, ANA positivity, and bilateral ocular involvement were associated with more severe disease phenotypes.
- Continued follow-up into adulthood was necessary for all patients, reinforcing that JIA-associated uveitis requires long-term multidisciplinary team (MDT) management and structured transitional care services.

**Disclosure of interest**
None declared.

## P094

### Is the prevalence of uveitis in juvenile idiopathic arthritis decreasing?

T. Güzel[1,*], M. T. Karadoğan[1], B. Çinpolat[1], Z. Hazıroğlu Ökmen[1], E. Fırat[1], M. Erdoğan[2], F. Çakmak[1]
[1]Pediatric Rheumatology, Basaksehir Cam and Sakura City Hospital; [2]Ophthalmology, Kanuni Sultan Suleyman Training and Research Hospital, Istanbul, Türkiye
**Correspondence:** T. Güzel
*Pediatric Rheumatology 2026*, **24(S1):**P094

**Introduction**
Juvenile idiopathic arthritis(JIA) is a chronic rheumatic disease that begins before the age of 16. Although arthritis is the primary concern, attention should also be paid to extra-articular manifestations such as uveitis. Since uveitis can occur asymptomatically in JIA patients, patients should undergo regular eye examinations. The risk is higher in the oligoarticular type of JIA and in RF-negative polyarticular JIA subtypes.
**Objectives**
The aim of this study was to demonstrate that systemic treatment in patients diagnosed with JIA may prevent the development of uveitis.
**Methods**
The study included patients diagnosed with JIA who were followed for at least 6 months between 2022 and 2026. Patient data were retrospectively obtained from electronic health records.
**Results**
A total of 171 patients were included in the study; 89 (52%) were male, 82 (48%) were female. Among the JIA subtypes, oligoarticular JIA was the most common with 100 cases (58.5%), followed by 30 (17.5%) enthesitis-related, 13 (7.6%) systemic, 11 (6.4%) RF-negative polyarticular, 11 (6.4%) psoriatic, 4 (2.3%) RF-positive polyarticular, and 2 (1.2%) unclassified. The JADAS27 scores at diagnosis and at the final follow-up found to be statistically significant. Patients began cDMARD treatment an average of 34.7 days after receiving their diagnosis. Uveitis developed in only two of the patients during follow-up. One of these patients was a 6-year-old male diagnosed with oligoarticular JIA; uveitis developed while he was on MTX, so adalimumab was added to his treatment. The other patient was a 9-year-old female also diagnosed with oligoarticular JIA; she was on etanercept when uveitis developed, so her treatment was switched to adalimumab. Uveitis was brought under control in both patients during follow-up. Three of the patients had experienced a uveitis flare-up prior to their JIA diagnosis; one was a male patient with systemic JIA, and the others were a female and a male patient with oligoarticular JIA. No uveitis flare-ups were observed in these patients during follow-up.
**Conclusion**
This study emphasizes the importance of initiating systemic treatment after a JIA diagnosis in preventing significant extra-articular involvement such as uveitis. While the incidence of uveitis in JIA patients is reported to be 12.7% in previous studies, the fact that uveitis was observed in only 2 (1.16%) of the patients followed for JIA in this study is noteworthy. Additionally, three patients (1.75%) reported a history of uveitis. Therefore, patients diagnosed with JIA should be monitored at regular intervals for uveitis, even if they are asymptomatic. Furthermore, the early initiation of systemic treatment appears to reduce the incidence of uveitis.

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**Disclosure of interest**
None declared.

## P095

### Pediatric pars planitis: clinical characteristics and outcomes from a tertiary care center

A. Villarejo Perez[1,*], R. Gonzalez Preciado[1], A. I. Ortueta Olartecoechea[2], E. Enriquez Merayo[1], C. Lopez Lopez[1], P. Tejada Palacios[2], J. de Inocencio [1]
[1]Pediatric Rheumatology Unit; [2]Pediatric Ophthalmology Unit, University Hospital 12 de Octubre, Madrid, Spain
**Correspondence:** A. Villarejo Perez
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**Introduction**
Pars planitis (PP) is classified as an idiopathic intermediate uveitis and represents the second most common cause of pediatric uveitis. It predominantly affects males and typically presents bilaterally, characterized by the presence of snowbanks and snowballs within the vitreous cavity.
**Objectives**
To describe the demographic, clinical, ophthalmologic, therapeutic, and outcome-related characteristics of pediatric patients diagnosed with PP, as well as to evaluate treatment response and relapse rates following biologic therapy discontinuation.
**Methods**
A retrospective study was conducted including patients diagnosed with PP who had been followed at a tertiary care center since 2006.
**Results**
Twenty-five eyes from 14 patients (53.8% male) were analyzed. Median age at disease onset was 8.2 years (IQR 6.4-11.0), with a median follow-up duration of 7.6 years (IQR 5.3-12.3). Bilateral involvement was observed in 78.6% of patients. Nine of 14 patients (64.3%) were symptomatic at disease onset, with decreased visual acuity being the most common presenting symptom (76.5%). At the initial ophthalmologic examination, 76% of eyes exhibited vitritis, 52% anterior chamber cells (Tyndall phenomenon), and 44% snowballs. Cystoid macular edema was the most frequent complication (3/24 eyes, 12%). The total number of complications during follow-up was associated with female sex ($p = 0.03$), without significant differences in median follow-up duration ($p = 0.26$). Synthetic disease-modifying antirheumatic drugs were required in 86% of patients, with subcutaneous methotrexate (MTX) being the most commonly prescribed agent (86%). A total of 64.3% of patients required biologic therapy for disease control, with adalimumab (ADA) used as the first-line biologic agent in all cases. Median time to biologic initiation was 15.2 months (IQR 9.9-41.6), while the median interval between MTX initiation and ADA initiation was 3 months. ADA was administered on 15 occasions in 8 patients, achieving permanent discontinuation due to sustained remission in 46.7% of cases after a median treatment duration of 30.3 months (IQR 26.9-54.1). Relapse occurred in 87.5% of patients after a median of 9.1 months (IQR 4.0-14.9), with no association between relapse and total treatment duration ($r = -0.08$, $p = 0.87$).
**Conclusion**
Pediatric PP follows a chronic relapsing-remitting course characterized by alternating periods of inactivity and exacerbation. Most patients require systemic therapy, demonstrating a moderate initial response but high relapse rates following treatment discontinuation.
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**Disclosure of interest**
None declared.

## P097

### Juvenile idiopathic arthritis in a patient with 22q11.2 deletion syndrome: a case report

D. Kairatova[1,*], M. Assylbekova[1], Z. Mukusheva [1], Z. Albekova[1], A. Dildabayeva [1], A. Akhmambetova [1]
[1]Rheumatology Program, Clinical Academic Department of Pediatrics, "University Medical Center" Corporate Fund, Astana, Kazakhstan
**Correspondence:** D. Kairatova
*Pediatric Rheumatology 2026*, **24(S1):**P097

**Introduction**
22q11.2 deletion syndrome, known as DiGeorge syndrome, is a genetic disorder presenting with congenital heart defects, hypocalcemia, cleft palate, and characteristic facial features. Immune dysfunction is also observed and may predispose patients to autoimmune diseases, including juvenile idiopathic arthritis (JIA) [1].
**Objectives**
To report a case of JIA diagnosed six years prior to the diagnosis of 22q11.2 deletion syndrome highlighting the importance of considering underlying immune dysregulation in patients with persistent autoimmune arthritis.
**Methods**
We present the case of a 9-year-old girl of Kazakh origin with juvenile idiopathic arthritis who was later diagnosed with 22q11.2 deletion syndrome. Clinical manifestations, laboratory findings and treatment history were reviewed. Response to therapy, frequent infections, and associated clinical features were assessed over the follow-up period.
**Case report**
A 9-year-old girl was diagnosed with polyarticular JIA at the age of 3 years after presenting with pain, swelling, and restricted movement of the knee and ankle joints, as well as deformity of the toes. Laboratory tests showed elevated inflammatory markers, including ESR 45.00 mm/h and CRP 47.24 mg/L, with negative antinuclear antibody and rheumatoid factor.
During the first 2 years after the disease onset, she received NSAIDs, intra-articular glucocorticoid injections, methotrexate, and etanercept. Treatment was complicated by frequent infections requiring antibiotics, leading to temporary interruptions in therapy and recurrent disease activity. Two years later, she was switched to adalimumab and then back to etanercept due to persistent disease activity. After 2 years, she presented with high disease activity, for which tocilizumab was initiated.
Throughout the disease course, she had frequent infections, a cleft palate, dysmorphic facies, failure to thrive with low weight and height for age. She repeatedly underwent uranostaphyloplasty. Whole-exome sequencing was recommended but was delayed. During her last hospitalization, hearing loss was reported, prompting genetic testing. Three months later, a heterozygous pathogenic deletion was identified at 22q11.21 consistent with 22q11.2 deletion syndrome. No congenital heart defect or persistent hypocalcemia was detected. At the latest follow-up, she remained on tocilizumab with moderate disease activity. Written informed consent was obtained from the patient's legal guardian.
**Conclusion**
JIA may be the early autoimmune manifestation of underlying 22q11.2 deletion syndrome, and delayed recognition of immune dysregulation can affect treatment decisions.

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**Disclosure of interest**
None declared.

## P098

### Type 1 interferon activation in patients with trisomy 21 with autoimmunity and pediatric rheumatic diseases

E. Rosswog[1,*], T. Strauß[2], F. Röwekamp[1], C. Suchrow[1], A. Schnabel[2], N. Brück[2], M. Haller[3], M. A. Lee-Kirsch[2], K. Mönkemöller[1], S. Catharina[2]
[1]Kinderkrankenhaus Amsterdamer Straße, Cologne; [2]University Hospital Carl Gustav Carus, Dresden; [3]Kinderarztpraxis Haller Gundelfingen, Gundelfingen, Germany
**Correspondence:** E. Rosswog
*Pediatric Rheumatology 2026*, **24(S1):**P098

**Introduction**
Patients with Trisomy 21 (T21) are predisposed to both autoimmune disorders and immune dysregulation, including increased susceptibility to infections. Interferon overexpression leading to JAK/STAT activation may contribute to this phenotype and elevated type 1 interferon signatures hint at JAK inhibition as a potential targeted therapy. However, this tailored approach has not been described for T21 individuals with pediatric rheumatic disorders.
**Objectives**
To investigate the spectrum of autoimmune manifestations and interferon signatures in patients with T21 and rheumatic diseases.
**Methods**
Clinical phenotypes, autoimmune manifestations, infectious complications, immunophenotyping, and treatment regimens were retrospectively analysed in patients with T21 followed at two pediatric centres in Germany.
**Results**
Seven patients with T21 were included. The rheumatologic diagnoses comprised polyarticular juvenile idiopathic arthritis (JIA) ($n$ = 2), oligoarticular JIA ($n$ = 1), psoriatic arthritis ($n$ = 2), enthesitis-related arthritis ($n$ = 1) and Raynaud's disease ($n$=1). In addition, all patients exhibited organ-specific autoimmune or autoinflammatory manifestations, including Celiac disease, Crohn's disease, Type 1 diabetes, Hashimoto thyroiditis, acne conglobata and nephrotic syndrome.Recurrent infections included pneumonia, sinusitis, otitis media, gastrointestinal infections and recurrent herpes simplex virus type 1 infections. Immunophenotyping revealed abnormalities of the adaptive immunity. In selected patients, interferon signatures showed marked activation of type I interferon pathway. Immunosuppressive treatment included methotrexate, tumor necrosis factor inhibitors, JAK inhibitors and prednisolone. Additional therapies included immunoglobulin replacement.
**Conclusion**
Our small cohort shows that patients with T21 and rheumatic manifestations may exhibit a complex phenotype characterized by concomitant autoimmunity and immunodeficiency. Comprehensive immunologic evaluation including type 1 interferon signatures and immunophenotyping are helpful in guiding individualized treatment strategies.

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**Disclosure of interest**
None declared.

## P099

### Beyond platelets: ACTN1 variants drive immune dysregulation at the thrombocytopenia–autoimmunity interface

F. Zeco[1,*], V. Michel[1], S. Palmeri[1], F. Adam[2], A. Kauskot[3], J. E. Montero Hernandez[4], B. Durel[5], N. Goudin[6], T. Leblanc[7,8], F. Suarez[9,10], N. Costedoat-Chalumeau[11,12], Fe. Sepulveda[4], A. Magerus[13], F. Rieux-Laucat[13]
[1]Immunogenetics of pediatric autoimmune diseases, Institut Imagine, INSERM UMR 1163, Université Paris Cité, Paris; [2]Laboratory for Hemostasis, Inflammation and Thrombosis (HITh), Le Kremelin Bicetre Hospital, INSERM UMR S 1176, Le Kremelin Bicetre; [3]Laboratory for Hemostasis, Inflammation and Thrombosis (HITh), Le Kremelin Bicetre Hospital, INSERM UMR S 1176; [4]Laboratory of Molecular Basis of Altered Immune Homeostasis, Imagine Institute, INSERM UMR 1163, Université Paris Cité; [5]Cell Imaging Platform; [6]Necker Bio-image Analysis Platform, INSERM US24; [7]Departmant of Pediatric Immunology and Hematology, Robert-Debré Hospital, APHP, Université Paris Cité; [8]CEREVANCE (Reference center for pediatric autoimmune cytopenia), Université Paris Cité; [9]Department of Adult Hematology, Necker-Enfants Malades Hospital, APHP, Université Paris Cité; [10]CEREDIH network (French National Reference Center for Primary Immunodeficiencies, Hôpital Necker-Enfants Malades, Paris, and Associated Collaborative Centers, France); [11]Department of Internal Medicine; [12]Reference Center for Rare Systemic Autoimmune and Autoinflammatory Diseases of the Île-de-France, Eastern, and Western regions, Cochin Hospital, APHP, Université Paris Cité; [13]Laboratory of Immunogenetics of Pediatric Autoimmune Diseases, Imagine Institute, INSERM UMR 1163, Université Paris Cité, Paris, France
**Correspondence:** F. Zeco
*Pediatric Rheumatology 2026*, **24(S1):**P099

**Introduction**
Genes essential for platelet biology can, when mutated, also drive immune dysregulation—as exemplified by *NBEAL2* mutations causing gray platelet syndrome with autoimmunity. *ACTN1*, encoding an actin-binding protein critical for cytoskeletal organization, underlies inherited thrombocytopenia, yet its potential role in immune homeostasis remains unexplored. Cytoskeletal dynamics govern both platelet function and lymphocyte activation, and actinopathies have recently emerged as a distinct class of immune dysregulation disorders.
**Objectives**
We investigated whether *ACTN1* variants contribute to the immune phenotype of patients presenting with immune thrombocytopenic purpura (ITP).
**Methods**
We studied a cohort of 4 ITP patients carrying heterozygous *ACTN1* variants. Functional analyses included lymphocyte and platelet phenotyping, actin remodeling assessment by microscopy, cell migration and deformability assays, and surface expression of immunoregulatory molecules.
**Results**
Patients presented with autoimmune cytopenias and immunological abnormalities. Microscopy revealed altered cell morphology consistent with impaired actin remodeling, likely disrupting intracellular trafficking and cell activation. Lymphocytes showed increased migration speed and reduced deformability. Strikingly, patient platelets overexpressed CD40L, a key driver of loss of immune tolerance, suggesting that platelet-mediated immune activation may contribute to autoimmunity in these patients.
**Conclusion**
*ACTN1* joins a growing family of platelet-disease genes, including *NBEAL2*, whose loss-of-function engages a dual thrombocytopenic and autoimmune program. These findings reveal a previously unrecognized role for ACTN1 at the interface between platelet biology and immune tolerance, and highlight the power of genetic dissection to reframe ITP as a mechanistically heterogeneous disorder amenable to targeted intervention.
**Disclosure of interest**
None declared.

## P100

### Founder variant, expanded phenotype, and new insights from the largest single-country cohort of a rare inborn autoinflammatory syndrome due to arpc1b gene defect

D. Bhattarai[1,*], A. Z. Banday[2], P. K. Patra[3], R. Baral[4]
[1]Immunology, Rheumatology and Allergy, College of Medical Sciences, Bharatpur, Kathmandu, Nepal, [2]Pediatrics, Government Medical College, Srinagar, Srinagar, [3]Pediatrics, AIIMS, Patna, Patna, India, [4]Pediatrics, Pokhara Academy of Health Sciences, pokhara, Nepal
**Correspondence:** D. Bhattarai
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**Introduction**
Genetic alteration of the genes of cytoskeletal regulatory proteins [e.g., actin-related protein complex-1 (ARPC1)] results in severe immune and hematological defects. ARPC1B deficiency (ARPC1BD) is an immune-actinopathy with combined immunodeficiency, allergy, inflammation, and bleeding tendency. Only a few dozen cases have been reported worldwide.

**Objectives**
To study the changed phenotypic landscape, genetic and clinical insights from cohort of ARPC1B deficiency.
**Methods**
We established the core area of cases with a c.64+2T>A splice-site mutation in the ARPC1B gene. We analysed the data for our objectives.
**Results**
A single Nepalese centre has diagnosed 20 cases (including 14 previously reported cases and 6 new cases) of ARPC1B deficiency so far. As far as published evidence shows, this is the largest single-country or single-centre cohort. Eighteen cases had the same homozygous splice-site founder variant c.64+2T>A in intron 2 of the ARPC1B gene. Two cases had compound heterozygous mutation (c.64+2T>A and c.784-1G>A). This cohort proves this variant to be the most prevalent pathogenic variant resulting in ARPC1BD to-date. Most of the cases were from 4 provinces. Haplotype analysis and homozygosity mapping proved this variant to be situated within a unique shared region of homozygosity, and all cases had similar haplotype around the variant, suggesting a founder gene effect. All of our cases had severe infective, allergic, autoimmune, and autoinflammatory features. Our cases (with the same founder variant) manifested a peculiar set of features, including frontal protuberance, a higher incidence of otitis, arthritis (including Rheumatoid factor positivity), recurrent skin vasculitis, severe skin hyperpigmentation, hyperkeratosis, distal phalangeal enlargement, inflammatory bowel disease-like features, gastroenteritis, elevated anti-tissue transglutaminase antibodies, immunoglobulin (Ig) A, and very high IgE. Homozygosity mapping and haplotype analysis in eight patients identified the c.64+2T>A intronic splice-site mutation as a founder variant (of Nepalese origin).
**Conclusion**
Comparatively soaring diagnosis of the cases with same pathogenic variant of ARPC1BD in a small geographic area has provided many scientific insights about rare immune-actinopathy changing phenotypic landscape and many definitions.

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**Disclosure of interest**
None declared.

## P101

### A tale of two siblings: contrasting phenotypes of ITCH deficiency – from systemic autoinflammation responding to methotrexate, to relative clinical quiescence

I. Z. Turtsevich[1,*], P. Mikolajski[1], A. Kavirayani[1], D. Miller[1], J. A. Archer[1], S. Richardson[2], D. Shears[2]
[1]Paediatric Rheumatology; [2]Clinical Genetics, Oxford University Hospitals NHS Foundation Trust, Oxford, UK
**Correspondence:** I. Z. Turtsevich
*Pediatric Rheumatology 2026*, **24(S1):**P101

**Introduction**
ITCH deficiency is an ultra-rare autosomal recessive disorder caused by mutations in the *ITCH* gene encoding an E3 ubiquitin ligase involved in immune regulation [1]. The condition is characterised by a broad spectrum of multisystem autoimmune and autoinflammatory manifestations, including arthritis, inflammatory bowel disease, endocrinopathies, and organ involvement, often accompanied by dysmorphic features, short stature, and developmental delay [2]. Marked phenotypic variability, even within families, remains poorly understood [1].
**Objectives**
To describe intrafamilial phenotypic variability in two siblings with compound heterozygous *ITCH* mutations and highlight divergent clinical trajectories.
**Methods**
An 8-year-old girl with the background of hypothyroidism, presented with recurrent fevers, painful panniculitic lesions, and systemic inflammation. Examination revealed multiple dysmorphic features (webbed neck, wide spaced nipples, downward slanted palpebral fissures), short stature, developmental delay, and lipodystrophic changes. Laboratory tests showed elevated inflammatory markers, negative ANA/ANCA, undetectable IgA, and positive GAD65 antibodies. MRI demonstrated widespread subcutaneous inflammation, fasciitis, and evolving peripheral lipodystrophy. She was steroid-responsive and achieved sustained remission on methotrexate, remaining clinically stable at 10 months of follow-up. Whole genome sequencing identified compound heterozygous variants in *ITCH* gene: a maternally inherited missense likely pathogenic variant (c.2264A> G; p.His755Arg) and a paternally inherited pathogenic nonsense variant (c.1783A> T; p.Lys595Ter), confirming *ITCH* deficiency. Her 13-year-old sister shares overlapping features including dysmorphism, developmental delay, short stature, but has not developed clinical or biochemical evidence of systemic inflammation or overt autoimmune disease and remains under surveillance.
**Conclusion**
This report illustrates striking intrafamilial phenotypic variability in *ITCH* deficiency, ranging from multisystem autoinflammatory disease to a currently non-inflammatory phenotype and the first reported case successfully managed with methotrexate. This case highlights the importance of comprehensive genetic testing in establishing a unifying diagnosis and the need for longitudinal monitoring and further research into disease-modifying factors and targeted therapeutic strategies.

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**Disclosure of interest**
None declared.

## P102

### Severe peripheral microvascular ischemia and thrombotic vasculopathy in a child with Rubinstein–Taybi syndrome associated with persistent antiphospholipid antibodies

M. A. Aljuaid[1,*]
[1]Pediatric Rheumatology Department, Prince Sultan Military Medical City, Riyadh, Saudi Arabia
**Correspondence:** M. A. Aljuaid
*Pediatric Rheumatology 2026*, **24(S1):**P102

**Introduction**
Rubinstein–Taybi syndrome (RTS) is a rare congenital disorder caused by pathogenic variants or deletions involving the CREBBP gene and is characterized by distinctive craniofacial features, developmental delay, and multisystem involvement. Although the phenotypic spectrum of RTS is well established, vascular manifestations and thrombotic complications remain poorly described.
**Objectives**
To describe a rare presentation of severe peripheral microvascular ischemia and thrombotic vasculopathy in a child with RTS associated with persistent antiphospholipid antibodies and thrombotic microangiopathy–like features.
**Methods**
A 6-year-old boy with genetically confirmed RTS presented with acute painful bluish discoloration of the fingers, palms, and toes progressing to digital ischemia and dry gangrene despite preserved large-vessel pulses. Extensive investigations demonstrated elevated von Willebrand factor, markedly reduced ADAMTS13 antigen, thrombocytopenia, eosinophilia, and persistent antiphospholipid antibodies detected on two occasions 12 weeks apart. Autoimmune serologies including ANA, ENA, anti-dsDNA, and ANCA were negative, with preserved complement levels. CT angiography demonstrated attenuation and nonvisualization of distal small vessels in the hands and feet, while skin biopsy revealed endothelial injury without immune complex deposition, supporting a microvascular thrombotic process rather than systemic vasculitis. Genetic testing confirmed a heterozygous CREBBP exon 26–27 deletion. Management included anticoagulation, pulse methylprednisolone, intravenous immunoglobulin, mycophenolate mofetil, vasodilator ther-

apy, and multidisciplinary care. Partial clinical improvement occurred; however, irreversible ischemic injury with dry gangrene and flexion deformities persisted.

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**Disclosure of interest**
None declared.

## P103

### IL-12/IFN-Γ axis deficiency presenting with disseminated tuberculosis and autoimmune disease: two case reports

M. T. Terreri[1,*], A. Pereira[1], A. P. Sakamoto[1], M. Fraga[1], D. Gerent Petry Piotto[1], G. Clemente[1]
[1]Pediatrics, Universidade Federal de São Paulo, Sao Paulo, Brazil
**Correspondence:** M. T. Terreri
*Pediatric Rheumatology 2026*, **24(S1):**P103

**Introduction**
The relevance of the interleukin (IL)-12 / interferon-gamma (IFN-γ) axis in the immune response against mycobacteria has been widely demonstrated. Defects in this axis are recognized as a key molecular and immunological link between immunodeficiency and autoimmunity.
**Objectives**
To describe two clinical cases involving deficiency in the IL-12 / IFN-γ axis with progression to disseminated TB and immune dysregulation, in association with juvenile systemic lupus erythematosus (jSLE) and Takayasu arteritis (TA). We also discussed the complex interface between infection, immunodeficiency, and autoimmunity.
**Methods**
**Case 1**
NMF, female, 14 years old, with a prior diagnosis of disseminated TB (miliary and lymph node involvement) and IL-12/IFN-γ axis immunodeficiency. After three years of follow-up, she developed persistent daily fever, hyporexia, polyarthralgia, and significant cervical lymphadenopathy. Initial investigations ruled out TB reactivation and other infections. She progressed with leukopenia with lymphopenia, elevated inflammatory markers, and hyperferritinemia. Evaluation for lymphoproliferative disorders or other neoplasias was negative. Due to persistence of symptoms, investigation for rheumatic disease was initiated. She presented ANA 1:1280 (homogeneous nuclear pattern), positive anti-dsDNA, low C4 levels, positive direct Coombs test, and subnephrotic proteinuria. She subsequently developed bilateral pleural effusion and acute renal failure with rapid resolution. A diagnosis of jSLE was confirmed, and treatment was initiated.
**Case 2**
KAS, female, 17 years old, diagnosed with type V TA, with involvement of the descending and abdominal aorta, as well as bilateral renal artery stenosis. She had a prior history of treatment for latent TB at 4 years of age, having completed appropriate therapy. Initial treatment for TA included adalimumab and methotrexate. After 3 months of treatment, she was diagnosed with lymph node TB, adalimumab was suspended and received appropriate treatment, with good clinical response. Due to disease progression, tocilizumab was initiated after TB treatment had been completed. During outpatient follow-up, she experienced multiple infectious episodes, including cytomegalovirus, Bartonella, and herpesvirus infections. For this reason, an immunodeficiency workup was performed, leading to the diagnosis of IL-12/IFN-γ axis immunodeficiency. Rifampicine prophylaxis has been continued.
**Conclusion**
These cases highlight the complex intersection between infection, immunodeficiency, and autoimmunity. Disseminated tuberculosis in a patient with primary immunodeficiency may not only reflect an underlying immune defect but also act as a trigger for autoimmune diseases. Early recognition of this overlap is crucial to guide targeted therapies and improve outcomes in pediatric patients with systemic manifestations.

**Disclosure of interest**
None declared.

## P104

### What is the difference between macrophage activation syndrome in systemic juvenile idiopathic arthritis and multisystem inflammatory syndrome in children associated with COVID-19?

I. Avrusin[1,*], K. Belozerov[1], O. Efremova[2], D. Tambovtseva[1], A. Zayats[1], L. Firsova [3], O. Kalashnikova[1], L. Bregel[2,4], M. Kostik [1]
[1]Hospital Pediatrics, Saint Petersburg State Pediatric Medical University, Saint Petersburg; [2]Irkutsk State Medical Academy of Postgraduate Education, branch of Russian Medical Academy of Continuous Professional Education, Irkutsk; [3]Department of Propaedeutics of Childhood Diseases named after V. Yuriev, Saint Petersburg State Pediatric Medical University, Saint Petersburg; [4]Irkutsk Regional Children's Clinical Hospital, Irkutsk, Russian Federation
**Correspondence:** I. Avrusin
*Pediatric Rheumatology 2026*, **24(S1):**P104

**Introduction**
Macrophage activation syndrome (MAS) is a severe condition characterized by hemophagocytosis, pancytopenia, coagulopathy, and multiple organ dysfunction. MAS is a major complication of systemic juvenile idiopathic arthritis (sJIA), with approximately 10% incidence, and quite often (around 18%) can complicate multisystem inflammatory syndrome in children associated with COVID-19 (MIS-C).
**Objectives**
To compare the characteristics of patients with MAS complicating the course of sJIA and MIS-C.
**Methods**
This retrospective study included 204 patients with sJIA and 166 patients with MIS-C. Of these, 54 patients with MAS according to the EULAR/ACR/PRINTO 2016 criteria were selected: 36 (17.6%) with sJIA (23 girls, 13 boys), 18 (10,8%) with MIS-C (9 girls, 9 boys), who were examined and treated at the clinics of Saint Petersburg and Irkutsk in the period from 2007 to 2024 (for sJIA) and 2020-2022 (for MIS-C).
**Results**
The main clinical signs observed with similar frequency in both groups were respiratory system involvement (66.7% - sJIA; 72.2% - MIS-C), lymphadenopathy (75% vs. 80%), hepatomegaly (86.1% vs. 81.3%), and splenomegaly (81% vs. 81.3%). Rash was also very common in both groups, but more often in sJIA-MAS group (97.2% vs. 82.4%, $p$=0.056). On the other hand, neurological symptoms were more frequent in MIS-C (66.7% vs. 30.6%, $p$=0.011). Among laboratory parameters elevated inflammatory biomarkers, such as ESR, CRP, ferritin, ALT, AST, and triglycerides were seen in both groups, as well as decreased hemoglobin and fibrinogen levels. However, some parameters were significantly different. MIS-C patients more often had thrombocytopenia (83.3% vs. 58.3% in sJIA, $p$=0.066), and higher median CRP levels (190.1 vs. 109.5 mg/l, $p$=0.005). Patients with sJIA more frequently exhibited elevated LDH (100% vs. 70.6%, $p$=0.005), and had higher ferritin levels (6900 vs. 1075 µg/l, $p$=0.009). The Hscore was calculated in both groups. In patients with sJIA-MAS it was statistically significantly higher (217 vs. 154, $p$<0.001). Therapy for MAS in both conditions is based on controlling systemic inflammation using glucocorticosteroids and biologic disease-modifying antirheumatic drugs (bDMARDs). However, bDMARDs were required significantly more often in sJIA-MAS cases (68.8% vs. 16.7%, $p$=0.002). In both groups more than half of the patients required admission to the intensive care unit (52.8% for sJIA and 66.7% for MIS-C). There were no fatal outcomes among patients with MIS-C-MAS, whereas two patients died in the sJIA-MAS group.
**Conclusion**
The course of MAS in sJIA and MIS-C is quite similar. However, in sJIA, it is more severe and more often requires bDMARDs treatment. The main differences between these conditions pertain to laboratory parameters and diagnostic criteria. The HScore is an effective universal screening tool in patients with MAS, but for most effective use of it for hemophagocytic syndrome prediction, it is necessary to determine the thresholds for different nosologies.
**Disclosure of interest**
None declared.

## P105

### Macrophage activation syndrome secondary to pediatric rheumatologic diseases: a multicenter cohort study

B. Menentoğlu[1,*], G. Özdemir[1], S. Doğantan[2], T. Çeltik[3], E. N. Dizman[4], S. İmamoğlu[5], S. Şener[6], T. Güzel[2], A. Dudaklı[1], F. Demir[7], F. Çakmak[2], M. Çakan[8], G. Aksu[5], K. Öztürk[4], B. Bora[3], N. Aktay Ayaz[1]
[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine; [2]Pediatric Rheumatology, Başakşehir Çam And Sakura Cıty Hospital, Istanbul; [3]Pediatric Rheumatology, Dokuz Eylul University Faculty of Medicine, Izmir; [4]Pediatric Rheumatology, Medeniyet University Faculty of Medicine, Istanbul; [5]Pediatric Rheumatology, Ege University, Izmir; [6]Pediatric Rheumatology, Adana City Hospital, Adana; [7]Pediatric Rheumatology, Acıbadem Altunizade Hospital; [8]Pediatric Rheumatology, Zeynep Kamil Training and Research Hospital, Istanbul, Türkiye
**Correspondence:** B. Menentoğlu
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**Introduction**
Macrophage activation syndrome (MAS) is a severe hyperinflammatory condition secondary to rheumatic diseases that may lead to multiorgan dysfunction and mortality.Data regarding the clinical presentation, treatment, and outcomes of MAS according to the underlying rheumatic disease remain limited.
**Objectives**
This study aimed to investigate the clinical presentation, laboratory findings, treatment strategies, and outcomes of MAS secondary to pediatric rheumatic diseases.
**Methods**
MAS diagnosis was based on the 2016 EULAR/ACR/PRINTO, Parodi criteria and expert opinion. Patients with infections, malignancies, or other non-rheumatic causes of hyperinflammation were excluded. Demographic, clinical, laboratory, treatment, and outcome data were analyzed.
**Results**
A total of 59 patients with MAS secondary to rheumatologic diseases were included. The mean age at MAS diagnosis was 7.75 ± 0.62 years, and 25 patients(42.4%) were female.In 48 patients(81.4%), MAS was the initial manifestation leading to the diagnosis of the underlying rheumatologic disease.The most common underlying disease was sJIA($n$=48),followed by SLE($n$=8),Kawasaki disease($n$=2),and Aicardi–Goutières syndrome($n$=1). Fifty-three patients(89.8%) experienced a single MAS episode, while 6 patients had recurrent attacks.All patients presented with fever.Rash was observed in 84.7% of patients, hepatosplenomegaly in 69.5%,arthritis in 32.2%,and serositis in 28.8%.Median ferritin, CRP, and ESR levels at MAS onset were 12,525 µg/L (IQR 6,356–27,426),78 mg/L (IQR 14–145),and 38 mm/h (IQR 14–78),respectively.Pulse methylprednisolone therapy was administered in 91.5% of patients.Biologic therapy was required in 31 patients, anakinra in 29 patients, with concomitant ruxolitinib in one patient and emapalumab in one patient, while 2 patients received tocilizumab. IVIG and plasmapheresis were administered in 30 and 17 patients, respectively.Among patients with MAS secondary to sJIA, arthritis was present in 60.4%, whereas none of the patients with SLE-associated MAS had arthritis ($p$=0.04).No significant differences were observed between sJIA and SLE-associated MAS regarding other clinical or laboratory findings. IVIG use was more frequent in SLE-associated MAS than in sJIA-associated MAS (87.5%vs.43.8%,$p$=0.04), and plasmapheresis was also used more commonly in SLE-associated MAS (75%vs.22.9%,$p$=0.006).
**Conclusion**
MAS commonly presented at the onset of pediatric rheumatologic diseases in this cohort. While clinical and laboratory findings were largely similar between sJIA- and SLE-associated MAS, differences were observed in arthritis frequency and treatment modalities.

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**Disclosure of interest**
None declared.

## P106

### Macrophage activation syndrome in juvenile dermatomyositis: a comprehensive review of 39 reported cases

F. Baldo[1,*], M. Rossano[2], P. Vaienna[3], A. Marino[1], F. Minoia[2]
[1]Clinica Reumatologica Pediatrica, ASSt-G.Pini-CTO; [2]SCPediatria-Immunoreumatologia, Fondazione IRCCS CA' Granda Ospedale Maggiore Policlinico; [3]Università degli Studi di Milano, Milano, Italy
**Correspondence:** F. Baldo
*Pediatric Rheumatology 2026*, **24(S1):**P106

**Introduction**
Macrophage Activation Syndrome (MAS) is a rare but potentially life–threatening complication of Juvenile Dermatomyositis (JDM). Its recognition is challenging because signs of systemic hyperinflammation often overlap with manifestations of the underlying disease, and no MAS–specific diagnostic criteria exist for JDM-MAS.
**Objectives**
To investigate demographic, clinical, laboratory parameters, therapy and outcomes of JDM associated MAS.
**Methods**
A systematic literature review was performed to identify all published cases of MAS associated with JDM, and demographic, clinical, laboratory, immunological and therapeutic variables were extracted.
**Results**
Thirty–nine patients were identified. Age at JDM onset, was 8.95 ± 4.83 years. Among patients with known sex (34), 54.3% were female, and amyopathic JDM was reported in 20% of cases for whom information was availble. An infectious trigger was identified in 15% of evaluable cases, and in 68% of patients MAS presents at JDM onset. Before MAS, laboratory findings showed elevated muscle and liver enzymes and hyperferritinemia; during MAS, most markers worsened, whereas CPK levels improved (see Table 1). Most prevalent autoantibodies included MDA5 (11) and NXP2 (6) (autoantibodies were not reported in 17 patients). For JDM, treatment information was reported in 34 cases: intravenous immunoglobulin (17) and pulse methylprednisolone (17) were the most frequently used agents, followed by methotrexate and prednisolone in 7, cyclosporine and cyclophosphamide in 6; other treatments included mycophenolate (5), plasma exchange (5), tacrolimus(4) and tofacitinib (3). Biologic DMARDS were reported only occasionally. Only 20 patients had specific MAS–directed therapies including cyclosporine (14), followed by intravenous immunoglobulin (10), pulse methylprednisolone or lower dose IV methylprednisolone in 11 cases, while smaller numbers of patients received anakinra (4), tocilizumab (3), dexamethasone(4) or cyclophosphamide(4), plasma exchange (5), continuous renal replacement therapy(2), tofacitinib(2), mycophenolate (2) or etoposide (2). Nine patients required ICU admission. Mortality was substantial, with five deaths, twenty–four survivors, and nine cases with unavailable outcomes.

**Table ( Abstract P106)**

| | PreMAS Median (IQR) | MAS Median (IQR) |
|---|---|---|
| CPK (U/L) | 1094 (205–4265) | 312 (104–1322.5) |
| LDH (U/L) | 637 (486–903) | 1085 (780–1607) |
| AST (U/L) | 217 (60–286) | 696 (106–1179) |
| ALT (U/L) | 108 (75–179) | 187.5 (77.5–539.5) |
| Ferritin (ng/mL) | 878 (594–2945) | 1684 (1002–4514) |
| Platelets ($\times 10^3$/µL) | 147.5 (80.5–234) | 90 (72–243.5) |
| Hemoglobin (g/dL) | 11.95 (11.95–12.55) | 10.2 (9.35–66.5) |

**Conclusion**
MAS in JDM frequently presents at disease onset and is characterized by severe hyperinflammation, cytopenias and clinically significant morbidity, including frequent need for intensive care. Interestingly, we observed lower CPK levels during MAS episodes. Hyperferritinemia remains a key diagnostic marker. The combination of relevant mortality and marked heterogeneity in diagnostic and therapeutic approaches highlights the need for standardized criteria and shared management strategies to improve patient outcomes.
**Disclosure of interest**
None declared.

## P107

### Interferon responsive gene expression differentiates macrophage activation syndrome complicating childhood-onset systemic lupus erythematosus (cSLE) from active cSLE alone

M. Heshin-Bekenstein[1,2,3,*], B. Jebb[4], M. Vinker-Shuster[1], Z. Ding[5], D. Dominguez[1,5], D. M. Levy[1], A. Knight[1,6], R. S. Yeung[1,4,7], D. Dissanayake[1,4], L. T. Hiraki[1,5]
[1]Rheumatology, The Hospital for Sick Children, University of Toronto, Totornto, Canada; [2]Rheumatology, Dana Children's Hospital of Tel Aviv Medical Center; [3]Grey Faculty of Medical and Health Sciences, Tel Aviv University, Tel Aviv, Israel; [4]Cell and Systems Biology Research Program; [5]Genetics and Genome Biology Program; [6]Neurosciences and Mental Health Program, The Hospital for Sick Children Research Institute; [7]Department of Pediatrics, Immunology and Institute of Medical Science, University of Toronto, Totornto, Canada
**Correspondence:** M. Heshin-Bekenstein
*Pediatric Rheumatology 2026*, **24(S1):**P107

**Introduction**
Macrophage activation syndrome (MAS) is a severe hyperinflammatory complication of rheumatic diseases that is increasingly recognized in childhood-onset systemic lupus erythematosus (cSLE). While MAS is often considered to be associated with interferon-γ (IFNγ)-driven inflammation based on Still's disease, this may not apply to SLE, where underlying interferon pathways may differ.
**Objectives**
To evaluate interferon-responsive gene (IRG) expression in treatment-naïve patients with new diagnosis of cSLE, with and without MAS.
**Methods**
This is an observational study of a treatment-naïve inception cohort of children with newly diagnosed cSLE. Patients were recruited from the SickKids clinic, fulfilled 2019 SLE classification criteria and had RNA samples at time of diagnosis prior to glucocorticoid initiation. SLE and MAS classification was based on expert pediatric rheumatologist diagnosis, with retrospective application of proposed criteria[1,2]. cSLE and MAS features were extracted from the dedicated Lupus Database, supplemented by chart review. cSLE+MAS patients were age, sex, ancestry and disease activity matched to patients without MAS (cSLE-only). Baseline demographic and disease characteristics were compared between groups, including age at diagnosis, sex, genetic ancestry, SLE classification features, and SLEDAI scores at sampling. Whole blood RNA analysis was completed using a NanoString platform. Individual gene expression was examined, and a 28-gene IRG score was calculated. We compared differential gene expression in treatment-naïve samples collected at diagnosis from cSLE+MAS versus cSLE-only (R 4.4.3 ($P$<0.05)).
**Results**
We included 9 cSLE+MAS and 9 cSLE-only patients. The majority were female (67% for both groups) and the most prevalent ancestry was East Asian (55% vs. 33%, $P$=0.60), followed by South Asian (22% in both groups). Median age at diagnosis was 13 years [IQR 11–14] vs. 13 years [IQR 10–14], ($P$=0.66), with a total of 44% with lupus nephritis in both groups and a median SLEDAI score at time of sampling of 13 [IQR 11.5–20.5] vs. 13 [IQR 10–26], ($P$=0.90). Differential gene expression was observed in several IRGs, with higher expression of *LY6E* ($P$=0.005), *IFI44L* ($P$=0.009), *IFI44* ($P$=0.012), *HERC5* ($P$=0.031), and *IFIT1* ($P$=0.039) in the non-MAS SLE only group. There was no significant difference in the 28-gene IRG score between the groups.
**Conclusion**
In this treatment-naïve inception cohort of children with new diagnosis cSLE, with and without MAS, we identified several differentially expressed interferon-responsive genes. These findings indicate differences in IRG expression in the context of MAS complicating cSLE, distinct from active cSLE alone. Our future work will expand the analysis to include additional IRG and pathway-specific approaches.

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**Disclosure of interest**
None declared.

## P109

### A teenager with hodgkin's lymphoma, polyarthritis, and osteonecrosis: diagnostic and therapeutic challenges

L. Martínez[1], A. Remesal [2,*], R. Alcobendas [2], C. Udaondo [2], A. Hercberg[3], V. Courel [2], C. Plasencia [2]
[1]Hospital Severo Ochoa , [2]Hospital La Paz , [3]Hospital de Getafe , Madrid , Spain
**Correspondence:** A. Remesal
*Pediatric Rheumatology 2026*, **24(S1):**P109

**Introduction**
Musculoskeletal manifestations in pediatric oncology patients may arise from infection, malignancy, treatment-related toxicity, or inflammatory processes. Their evaluation and management can be particularly challenging in patients with underlying bone lesions and persistent pain despite adequate antimicrobial and oncologic treatment.
**Objectives**
To describe the clinical presentation, diagnostic workup, and therapeutic management of severe polyarthritis associated with osteonecrotic bone lesions in an adolescent with Hodgkin lymphoma.
**Methods**
We report the case of a 16-year-old adolescent with a previous diagnosis of Hodgkin lymphoma (HL) and a history of multifocal osteoarticular infection caused by Proteus mirabilis, for which prolonged antibiotic therapy had been initiated.

**References**

1. Despite appropriate antimicrobial treatment and control of the infectious process, the patient continued to experience severe diffuse pain predominantly affecting the joints of the lower limbs. Pain was refractory to standard analgesia, prevented even minimal passive mobilization, and required intensive pain management including opioid therapy. Progressive muscle wasting and functional impairment secondary to prolonged immobility were also observed.
2. Magnetic resonance imaging and computed tomography demonstrated multiple lytic bone lesions involving the pelvis and lower extremities, interpreted as areas of osteonecrosis secondary to previous infection superimposed on metastatic involvement from the primary malignancy.
3. Multiple diagnostic procedures, including biopsies and repeated arthrocenteses, were performed. Synovial aspirates showed thick fatty fluid with abundant cellularity, interpreted as bone marrow contamination due to extensive bone destruction. No microorganisms were isolated, and no evidence of persistent malignancy was identified on histopathological analysis.
4. Given the persistence of severe pain and inflammatory symptoms despite antimicrobial therapy and supportive care, a multidisciplinary team including pediatric rheumatology, oncology, infectious diseases, radiology, and pain specialists decided to initiate treatment with intravenous pamidronate and intra-articular triamcinolone injections. Due to incomplete clinical response and persistent diffuse pain, intravenous tocilizumab (interleukin 6 inhibitor) was subsequently added.
5. After combined therapy, the patient showed marked clinical and radiological improvement, with significant reduction in pain, progressive recovery of mobility, and improvement in muscle mass and functional status.
6. This case highlights the complexity of musculoskeletal manifestations in pediatric oncology patients with extensive bone involvement. In the absence of established clinical guidelines, therapies such as bisphosphonates, intra-articular corticosteroids, and interleukin 6 inhibitors agents may represent useful therapeutic options for controlling pain and inflammation and improving quality of life.

**Disclosure of interest**
None declared.

## P110

### Biomarkers of bone metabolism in juvenile idiopathic arthritis compared to controls based on a Norwegian observational study

A. Lundestad[1,2,*], L. Cetrelli [3,4], O. W. Angenete[5,6], K. Rosendahl[7,8], M. Hoff[9,10], G. B. Hoftun[2], K. Tylleskär[11], E. Nordal[12,13], C. Aulin[14], P. R. Romundstad[15], M. Rygg[2,3]

[1]Department of Clinical and Molecular Medicine, NTNU- Norwegian University of Science and Technology; [2]Department of Pediatrics, St.Olavs Hospital; [3]Department of Clinical and Molecular Medicine, NTNU; [4]Mid-Norway (TkMidt), Center for Oral Health Services and Research; [5]The Department of Circulation and Medical Imaging, NTNU; [6]Department of Radiology and Nuclear Medicine, St.Olavs Hospital, Trondheim; [7]Department of Clinical Medicine, UiT The Artic University of Norway; [8]Section of Paediatric Radiology, University Hospital of North Norway, Tromsø; [9]Department of Neuromedicine and Movement Science, NTNU; [10]Department of Rheumatology, St.Olavs Hospital, Trondheim; [11]Department of Pediatrics and Adolescent Medicine, Haukeland University Hospital, Bergen; [12]Department of Clinical Medicine, UiT; [13]Department of Pediatrics, University Hospital of North Norway, Tromsø, Norway; [14]Department of Medicine Solna, Center for Molecular Medicine, Karolinska Institutet, and Division of Rheumatology, Karolinska University Hospital, Stockholm, Sweden; [15]Department of Public Health and Nursing, NTNU, Trondheim, Norway

**Correspondence:** A. Lundestad

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**Introduction**

There is currently little knowledge about biomarkers connecting inflammation and bone metabolism in juvenile idiopathic arthritis (JIA). Studies on biomarkers reflecting bone health and long-term outcomes in JIA are needed.

**Objectives**

To increase our understanding of bone metabolism in children with JIA, and to identify biomarkers associated with reduced bone health to facilitate intervention strategies.

**Methods**

Data on 187 children with juvenile idiopathic arthritis (JIA) according to the International League of Associations for Rheumatology (ILAR) criteria [1], along with age- and sex-matched controls from the general population, were obtained from the NorJIA study, http://norjia.com,a longitudinal, multi-center cohort study. Clinical examination, validated patient/parent-reported questionnaires, blood tests, and bone density measurements were performed. Serum levels of Tartrate-resistant acid phosphatase (TRAP-5b) and Cross-linked N-telopeptide of type I collagen (NTX-I) were analyzed using enzyme-linked immunosorbent assay (ELISA) [2]. The results are presented as geometric mean (GM), mean of log-transformed values, then exponentiated. Bone mineral density (BMD) was measured with dual-energy X-ray absorptiometry (3). BMD z-scores were adjusted for bone age. We used standard descriptive statistics and linear regression.

**Results**

Median age of the 187 children with JIA (60% girls) was 15 (IQR 12-17) years, and 15 (12-17) years in the control group. Median disease duration was 7 years, 51% had used or were currently using biologic disease-modifying anti-rheumatic drugs (bDMARDs), 25% had ever used systemic steroids, and 35% had active disease. GM TRAP levels were 9.5 (95% CI 8.6, 10.3) U/ml in JIA and 10.4 (9.2, 11.6) U/ml in controls. GM NTX-I levels was 48.7 (40.6, 56.7) ng/ml and 52.5 (43.0, 61.9) ng/ml, respectively. Both TRAP and NTX-I levels decreased progressively with age. TRAP declined from 13.6 (11.7, 15.5) U/ml at ages 4–12 to 6.9 (6.1, 7.6) U/ml at ages 13–16, and further to 3.9 (3.2, 4.5) U/ml at ages 17–18. NTX-I showed a similar drop, from 64.3 (42.9, 85.8) ng/ml to 21.0 (15.3, 26.8) ng/ml and 5.8 (3.5, 8.2) ng/ml across the same age groups. A similar trend was observed in controls. When assessing factors known to affect bone health such as parenteral education, ISO-BMI, physical activity, daily intake of vitamin D and calcium, with the corresponding serum levels, we did not observe any clear pattern of changes in TRAP and NTX-I values. However, these markers were marginally elevated in children with JIA with more unfavorable disease characteristics, including those who had ever used bDMARDs or systemic steroids. These groups also demonstrated a slightly lower adjusted BMD (L1–L4), Z-score.

**Conclusion**

Bone turnover markers declined with age and were largely similar between children with JIA and controls, although marginally higher in those with more severe disease, together with slightly lower BMD. Due to marked age-related variability, TRAP and NTX-I are currently of limited value for assessing bone health in children with JIA.

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**Trial registration identifying number**

NCT03904459.

**Disclosure of interest**

None declared.

## P111

### Mandibular involvement in pediatric CNO: retrospective analysis of treatment and outcome in a multicenter cohort

A. Theobold[1,*], H. Morbach[2], H. Lausmann[3], C. Van Quekelberghe[4], F. Weller-Heinemann[5], A. Schnabel[6], K. Tenbrock[7], T. Keller[8], M. Laaths[9], J. Kümmerle-Deschner[1], H. Girschick[10], A. Skrabl-Baumgartner[11], A. Mráz[12], C. Reiser[1,12]

[1]Pediatric Rheumatology, Faculty of Medicine and University Hospital Tübingen, Tübingen; [2]Pediatric Rheumatology and Immunology, Children's Hospital, University of Würzburg, Würzburg; [3]Pediatric Rheumatology, Children's Hospital St. Marien, LA Regio Kliniken gKU AdöR, Landshut; [4]Pediatric Rheumatology, Clementine Children's Hospital, Frankfurt am Main; [5]Department of Pediatrics, Prof. Hess Parent-Child Center, Bremen-Mitte Hospital, Bremen; [6]Department of Pediatrics, Faculty of Medicine and University Hospital Carl Gustav Carus, Technische Universität Dresden, Dresden; [7]Dept of Pediatrics, Pediatric Rheumatology, RWTH Aachen University, Aachen; [8]Department of Pediatrics, KJF Josefinum Children's Hospital, Augsburg; [9]Department of Pediatrics, St. Marien Hospital, Amberg; [10]Department of Pediatrics, Vivantes Clinic Friedrichshain, Berlin, Germany; [11]Department of Pediatrics, Medical University Graz, Graz; [12]Department of Pediatrics, Bregenz Regional Hospital, Bregenz, Austria

**Correspondence:** A. Theobold

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**Introduction**

Involvement of the mandible in pediatric chronic nonbacterial osteomyelitis (CNO) is associated with a severe disease course [1] and high rates of second-line therapy [1, 2].

**Objectives**

This retrospective multicenter study included pediatric CNO patients from 12 centers in Germany and Austria with mandibular involvement.

**Methods**

Multiple non-overlapping therapy episodes per patient were analyzed. Clinical and radiological inactive disease were defined as absence of clinical lesions and MRI lesions, respectively.

**Results**

Of 34 included patients, 32 were eligible for longitudinal analysis. All patients initially received NSAIDs, but 75% (24/32) required treatment escalation and 47% (15/32) received two or more non-NSAID therapeutic classes. Treatments included TNF inhibitors ($n$=14), pamidronate ($n$=13), csDMARDs ($n$=9), and prednisolone ($n$=14). TNF inhibitors achieved the highest rates of clinical inactive disease at months 6, 9, and 12 (70-78%), whereas no relevant differences were observed at month 3 (33–43%). CsDMARDs were associated with the highest rates of clinical flares during the first 9 months (14-50%). Radiological inactive disease was rare in the first year (Pam: 2/6, TNFi: 3/16, csDMARDs: 0/5) but increased in the second year across all groups (Pam: 2/4, TNFi: 5/7, csDMARDs: 1/2). Kaplan–Meier analysis indicated a shorter time to clinically inactive disease with pamidronate (median 4.8 months) compared to csDMARDs (5.7 months) and TNF inhibitors (6.2 months), without reaching statistical significance ($p$=0.51). Several disease activity (DA) scores, including physician/patient global assessment, PedCNO, and MRI based composite scores, decreased during the first 18 months of follow-up, followed by increasing DA between month 24 and 36.

**Conclusion**
In this cohort, a high proportion of patients required treatment escalation beyond NSAIDs, consistent with previous reports [1, 2]. TNF-α inhibitors, particularly adalimumab, showed the highest rates of clinically inactive disease, whereas pamidronate showed the highest rates of radiological response. Radiological improvement was delayed, emphasizing the importance of prolonged follow-up. Increasing disease activity after 24 months suggests that short-term improvement may not reflect sustained disease control. Although pamidronate showed a numerically shorter time to clinical inactive disease, this did not reach statistical significance, as reported previously (2, 3).

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**Disclosure of Interest**: None declared.

## P112

### Characteristics of children with mandibular chronic non-bacterial osteomyelitis

I. Avrusin[1,*], M. Alimov[1], Y. Vlasova[1], M. Alimov[1], L. Sorokina [1], E. Isupova [1], M. Kostik [1]
[1]Hospital Pediatrics, Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation
**Correspondence:** I. Avrusin
*Pediatric Rheumatology 2026*, **24(S1):**P112

**Introduction**
Chronic nonbacterial osteomyelitis (CNO) is a non-infectious autoinflammatory skeletal disease, characterized by single or multifocal bone tissue lesions, manifesting predominantly in childhood and adolescence and prone to recurrence. Mandibular CNO (MCNO) is typical, but relatively rare form of pediatric CNO, whom dental specialists are not-familiar. The rare incidence of MCNO limits large-scale studies, leading to the absence of standardized diagnostic criteria and hindering the development of effective treatment protocols. Improving knowledge of jaw CNO requires a clear understanding of current strategies for its diagnosis and treatment.
**Objectives**
To evaluate the clinical, laboratory and instrumental characteristics of pediatric MCNO.
**Methods**
The retrospective single-center study included 10 children (8 girls, 2 boys) with MCNO who were observed and treated in the clinic of St. Petersburg State Pediatric Medical University from 2019 to 2025. Median age of patients was 12 years (from 9 to 16).
**Results**
The main MCNO features included jaw pain (80%), swelling (90%), and facial asymmetry due to mandibular overgrowth (80%). Half of the patients had skin hyperemia over the affected area. Enlarged submandibular lymph nodes (40%), restricted mouth opening (40%), and non-persistent low-grade fever (40%) were less frequently observed signs. Imaging features included cortical mandibular thickening (60%) due to hyperostosis and swelling of the adjacent muscles (50%), mandibular osteosclerosis (40%) and periostitis (40%). A comprehensive examination of the musculoskeletal system revealed lower limb involvement in 40% of patients, which was multifocal in nature. Bone biopsy showed thinning and fragmentation in the cortical layer in 50% of the patients. Laboratory blood parameters did not show significant deviations. The majority of patients (60%) received empirical antibacterial therapy prescribed by the dental specialists during the period of initial clinical manifestations, which was proved to be ineffective. After MCNO diagnosis a combination of NSAIDs with bisphosphonates, which yields a positive effect, was used.
**Conclusion**
MCNO remains a little-known disease for non-rheumatologists with delayed diagnosis and an overly invasive examination that is unnecessary. The most effective therapy for this condition seems to be a combination of bisphosphonates and NSAIDs.
**Disclosure of interest**
None declared.

## P113

### Acute phase reactants in pediatric chronic nonbacterial osteomyelitis: implications for diagnosis and correlation with disease phenotype

B. Tetik Dinçer[1,*], P. Ö. Avar Aydın[1], F. Aydın[1], D. Sarısoy[1], E. Erorhan[1], O. Bahçeci[1], Z. B. Özçakar[1]
[1]Pediatric Rheumatology, Ankara University School of Medicine, Ankara, Türkiye
**Correspondence:** B. Tetik Dinçer
*Pediatric Rheumatology 2026*, **24(S1):**P113

**Introduction**
The diagnostic relevance of acute phase reactants (APRs) in chronic nonbacterial osteomyelitis (CNO) remains uncertain. While many patients present with mild-to-moderate elevations, some exhibit normal values. In contrast, markedly elevated APRs may point toward CNO mimickers and can lead to lower classification scores.
**Objectives**
This study aimed to evaluate the impact of elevated APRs on CNO classification at diagnosis and to explore their association with clinical and disease characteristics.
**Methods**
In this retrospective single-center study, patients diagnosed with CNO were stratified based on APR levels at diagnosis into two groups: markedly elevated APRs (defined as CRP ≥30 mg/L and/or ESR ≥60 mm/hour according to classification criteria) and normal-to-moderately elevated APRs. Clinical features, imaging findings, and treatment approaches were compared between the groups.
**Results**
A total of 51 patients with CNO were included, of whom 31% ($n$=16) had markedly elevated APRs. This group demonstrated lower classification scores, lower hemoglobin levels, and higher platelet and ferritin levels. They also showed more widespread skeletal involvement and a greater frequency of CNO-associated conditions. The median number of bone lesions was significantly higher in patients with markedly elevated APRs, and a moderate correlation was found between CRP levels and lesion count. Involvement of the pelvis, clavicle, and sacroiliac joints was significantly more frequent in this group.
**Conclusion**
A considerable proportion of CNO patients present with markedly elevated APRs, which may limit the accuracy of current classification criteria. These patients tend to have more pronounced systemic inflammation, more extensive disease involvement—particularly affecting the clavicle, pelvis, and sacroiliac joints—and a higher likelihood of associated conditions.
**Disclosure of interest**
None declared.

## P114

### Neridronate in chronic nonbacterial osteomyelitis (CNO): clinical and standardized radiological efficacy evaluation

G. Martini[1,*], S. Pastore[2], F. Biscaro[3], E. Cattaruzzi[4], A. Taddio[2], A. Florenzano[5], C. Giraudo[6], L. Cereser[5]
[1]Pediatrics, University Hospital, Udine; [2]Pediatric rheumatology, IRCCS Burlo Garofolo, Trieste; [3]Pediatrics, Ca' Foncello Hospital, Treviso; [4]Radiology, IRCCS Burlo Garofolo, Trieste; [5]Radiology, University Hospital, Udine; [6]Radiology, University Hospital, Padova, Italy
**Correspondence:** Giorgia Martini
*Pediatric Rheumatology 2026*, **24(S1):**P114

**Introduction**
CNO presents a wide spectrum of severity ranging from mild unifocal to extensive multifocal bone inflammation. Whole-body MRI (WBMRI) is increasingly used both at time of diagnosis to identify clinically silent

lesions and in follow-up to monitor the response to treatment. First-line therapeutic agents are NSAIDs but also corticosteroids, DMARDs, bisphosphonates and anti-TNF agents have been suggested.

**Objectives**

To evaluate efficacy of Neridronate in children with CNO by clinical response and by assessment of changes in radiological disease activity using a modified radiological index for non-bacterial osteitis (mRINBO), recently proposed to standardize reporting and quantification of WBMRI findings [1].

**Methods**

WBMRI of children with CNO before and after receiving neridronate iv infusions were evaluated by two expert radiologists blinded to the phase of the disease and to the treatment. They independently assessed each WBMRI and filled out the mRINBO score. Evaluated parameters were number and anatomic site of radiologically active bone lesions (RAL), size of RAL, extramedullary and spinal involvement and relative changes. Bone pain intensity and lab tests were evaluated before starting and after treatment.

**Results**

Eight patients (3 M, 5 F) with mean age at onset 8.4 years were included. Before treatment bone pain was the main symptom in all patients, fever was present in 5/8. Patients received 3 to 6 iv infusions of neridronate at mean 3.1 months interval (range 0.5-4). All patients experienced disappearance of pain, ESR and CRP significantly reduced from baseline (mean 43 to 16.5 mm/h with $p$ 0.01 and 9.4 to 3.2 mg/L with $p$ 0.05 respectively). Inter-observer agreement between the two radiologists was excellent for the number of RAL, with an intraclass correlation coefficient (ICC, 95% CI) of 0.87 and a weighted Cohen's kappa of 0.88 (95% CI) and was very good for the total mRINBO score with an ICC of 0.78 (95% CI) and a weighted Cohen's kappa of 0.77 (95% CI). The mRINBO score improved in all patients from a median of 5 (range 2–7) at baseline to 2.5 (0–4) after treatment ($p<0.05$). Neridronate was well tolerated with 3/8 patients reporting mild flu-like syndrome after the first infusion.

**Conclusion**

Our data suggests Neridronate as a promising and well-tolerated treatment for CNO as it induced a significant clinical improvement and reduction of radiological disease activity. mRINBO confirms as a reliable and reproducible tool for disease activity evaluation based on WBMRI. According to our knowledge, this is the first report in which efficacy of neridronate was demonstrated by standardized evaluation of radiological changes.

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**Disclosure of interest**

None declared.

## P117

### Persistent radiologic activity despite clinical improvement in chronic non-bacterial osteomyelitis: longitudinal whole-body MRI assessment using CROMRIS

K. Liontou[1], S. Proutzos[2], K. Chiotopoulou[3], K. Kourtesi[3], M. Gkezepi [3], O. Papakonstantinou[2], L. Fotis[3,*]
[1]"General Pediatrics and Pediatric Subspecialties: Clinical Practice and Research", School of Medicine; [2]Department of Radiology, ATTIKON General University Hospital; [3]Department of Pediatrics, Division of Pediatric Rheumatology, ATTIKON General University Hospital, National and Kapodistrian University of Athens, Athens, Greece
**Correspondence:** L. Fotis
*Pediatric Rheumatology 2026*, **24(S1):**P117

**Introduction**

Whole-body MRI (WB-MRI) is increasingly used for disease monitoring in chronic nonbacterial osteomyelitis (CNO). However, the relationship between clinical improvement and radiologic remission remains incompletely understood. Chronic Recurrent Multifocal Osteomyelitis MRI Scoring system (CROMRIS) provides a standardized quantitative assessment of inflammatory burden and may offer a more sensitive evaluation of disease activity during longitudinal follow-up.

**Objectives**

To evaluate longitudinal radiologic evolution in pediatric CNO using serial WB-MRI and CROMRIS, and to assess the relationship between clinical improvement and persistent radiologic inflammatory activity.

**Methods**

We retrospectively evaluated pediatric CNO patients followed at a tertiary pediatric rheumatology center who underwent serial WB-MRI examinations during follow-up. Clinical manifestations, treatment exposure, and CROMRIS scores were recorded. Baseline and final WB-MRI examinations were compared, and subgroup analyses according to treatment intensity were performed.

**Results**

Twenty-three pediatric CNO patients underwent longitudinal WB-MRI assessment with serial CROMRIS evaluation. Median CROMRIS decreased from 46.0 at baseline to 25.0 at final follow-up, while 22/23 patients demonstrated a longitudinal reduction in CROMRIS scores. Despite radiologic improvement, residual MRI inflammatory activity remained detectable in all patients at final follow-up, with no patient achieving complete radiologic remission (CROMRIS=0). Patients requiring treatment escalation with bisphosphonates and/or biologic therapy demonstrated higher baseline radiologic burden compared with patients managed without intensified therapy. In the intensified-treatment group ($n$=10), median CROMRIS decreased from 53.5 to 22.5 during follow-up. In contrast, patients managed without bisphosphonates or biologic therapy ($n$=13) demonstrated lower baseline radiologic burden, with median CROMRIS decreasing from 46.0 to 26.0. Clinical improvement, including reduction in pain, fatigue, and functional limitation, was observed in all patients during follow-up. However, persistent MRI inflammatory activity frequently remained detectable despite substantial clinical improvement.

**Conclusion**

Clinical improvement in CNO does not necessarily correspond to complete radiologic remission. Serial WB-MRI assessment using CROMRIS identified persistent inflammatory activity despite substantial longitudinal improvement and reduction in radiologic disease burden. Patients with more severe baseline radiologic involvement demonstrated marked improvement following treatment escalation, although complete MRI remission remained uncommon. CROMRIS may represent a valuable longitudinal imaging outcome measure for disease monitoring and therapeutic assessment in pediatric CNO.

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**Disclosure of interest**

None declared.

## P118

### Paediatric osteoporosis in a tertiary paediatric rheumatology unit: a retrospective case series

I. Cardoso[1], R. Nicolau[2], M. C. Natal[3], F. Gomes[4], D. M. Mikiel[5], P. J. Pereira[6], B. F. Esteves[3], S. Ganhão[7], F. Aguiar[7], M. Rodrigues[7,*], I. Brito[7]
[1]Pediatrics Department, ULS Gaia/Espinho, Gaia/Espinho; [2]Rheumatology Department, ULS Trás-os-Montes e Alto Douro, Vila Real; [3]Rheumatology Department, ULS São João, Porto; [4]Pediatrics Department, ULS Braga, Braga; [5]Pediatrics Department, ULS Matosinhos, Matosinhos; [6]Rheumatology Department, ULS Braga, Braga; [7]Pediatric and Young Adult Rheumatology Unit, ULS São João, Porto, Portugal
**Correspondence:** M. Rodrigues
*Pediatric Rheumatology 2026*, **24(S1):**P118

**Introduction**

Paediatric osteoporosis is a rare and heterogeneous condition, frequently secondary to chronic disease, corticosteroid exposure and reduced mobility, requiring combined clinical, densitometric and radiological assessment for diagnosis and management.

**Objectives**

To characterise the clinical presentation, aetiology, densitometric findings, treatment and outcomes of children and adolescents with osteoporosis followed in a tertiary paediatric rheumatology unit.

**Methods**
Retrospective case series including patients followed between 2008 and 2025 at a tertiary paediatric rheumatology centre. Demographic, clinical, laboratory, radiological and densitometric data, treatment and outcomes were collected. Osteoporosis was defined according to International Society for Clinical Densitometry (ISCD) criteria. One additional patient with markedly reduced BMD and high fracture risk requiring antiresorptive therapy despite not fulfilling ISCD criteria was also included.
**Results**
Nine patients were included (66.7% male), with a median age at diagnosis of 12 years (range 7–17). Eight fulfilled ISCD criteria for paediatric osteoporosis. Most cases were secondary and multifactorial, mainly associated with chronic corticosteroid exposure and reduced mobility, occurring in the context of complex neurological, oncological and endocrine disorders, while one patient presented probable primary idiopathic osteoporosis despite negative next-generation sequencing testing for osteogenesis imperfecta. Five patients (55.6%) presented vertebral fractures, all involving multiple vertebral compression fractures; asymptomatic vertebral fractures were identified in three patients, either incidentally or during targeted skeletal evaluation. Three patients presented long-bone fractures fulfilling ISCD criteria. Median minimum BMD Z-score was -2.9 (range -6.0 to 1.3). Vitamin D deficiency was identified in 55.6% of patients. Eight patients required antiresorptive therapy, most commonly intravenous zoledronic acid (55.6%), while pamidronate was used in 22.2% of cases. One patient subsequently received denosumab following fracture recurrence after zoledronic acid discontinuation. Oral risedronate was used in the only patient not fulfilling ISCD criteria.
**Conclusion**
Paediatric osteoporosis in this cohort was predominantly secondary and largely multifactorial, although one patient presented probable primary idiopathic osteoporosis. Vertebral fractures represented a major feature of disease burden and were often asymptomatic. The low number of identified patients in a tertiary centre managing complex chronic diseases suggests substantial underdiagnosis of paediatric bone fragility, particularly among high-risk populations followed in paediatric neurology, oncology and endocrinology. Greater awareness and systematic evaluation of bone health may improve early diagnosis and management.
**Disclosure of interest**
None declared.

## P119

### Acne-dominant phenotype, axial involvement, and treatment outcomes in pediatric SAPHO spectrum disease: a nationwide multicenter study from Türkiye

M. O. Erkan[1,*], E. Sag[1], V. Cam[1], E. Tunce[2], E. Kilic Konte[3], S. D. Arik[4], M. Kacmaz[5], M. Cakan[6], H. Kisaoglu[7], H. Tastanoglu[8], B. Acun[9], S. M. Toprak[10], M. Sezer[11], Z. Ekici Tekin[12], F. Gundogmus[13], O. A. Akin[14], G. Oguz[15], S. Cevizbas[16], S. E. Unsal[16], S. Turkucar[15], S. Demir[14], M. Kalyoncu[13], F. Haslak[10], S. A. Bakkaloglu[9], A. Pac Kisaarslan[8], N. Aktay Ayaz[4], O. Kasapcopur[3], B. Sozeri[2], O. Basaran[1], Y. Bilginer[1], S. Ozen[1]
[1]Pediatric Rheumatology, Hacettepe University, Ankara; [2]Pediatric Rheumatology, Ümraniye Training and Research Hospital; [3]Pediatric Rheumatology, Cerrahpaşa University; [4]Pediatric Rheumatology, İstanbul University, İstanbul; [5]Pediatric Rheumatology, Diyarbakır Children's Hospital, Diyarbakır; [6]Pediatric Rheumatology, Zeynep Kamil Training and Research Hospital, İstanbul; [7]Pediatric Rheumatology, University of Health Sciences, Kayseri City Hospital; [8]Pediatric Rheumatology, Erciyes University, Kayseri; [9]Pediatric Rheumatology, Gazi University, Ankara; [10]Pediatric Rheumatology, İstanbul Medeniyet University, İstanbul; [11]Pediatric Rheumatology, Ankara Training and Research Hospital; [12]Pediatric Rheumatology, University of Health Sciences, Ankara Bilkent City Hospital, Ankara; [13]Pediatric Rheumatology, Karadeniz Technical University, Trabzon; [14]Pediatric Rheumatology, Eskişehir Osmangazi University, Eskişehir; [15]Pediatric Rheumatology, Pamukkale University, Denizli; [16]Pediatric Rheumatology, Dokuz Eylül University, İzmir, Türkiye
**Correspondence:** M. O. Erkan
*Pediatric Rheumatology 2026*, **24(S1):**P119

**Introduction**
SAPHO spectrum disease is a rare autoinflammatory osteoarticular disorder with heterogeneous dermatologic and musculoskeletal manifestations. Pediatric data remain limited, particularly on lesion distribution, isotretinoin exposure, and real-life treatment outcomes.
**Objectives**
This study aimed to describe the demographic, clinical, dermatologic, osteoarticular, imaging, treatment, and outcome characteristics of patients with pediatric SAPHO spectrum disease.
**Methods**
We retrospectively analyzed 58 patients from a nationwide multicenter cohort. Demographic, clinical, imaging, treatment, isotretinoin exposure, and longitudinal outcome data were collected. Continuous variables were summarized as median and interquartile range, and categorical variables as numbers and percentages. Categorical variables were compared using chi-square or Fisher's exact test, and paired longitudinal changes using the Wilcoxon signed-rank test.
**Results**
A total of 58 patients were included; 13 (22.4%) were female. Median age at first manifestation was 14.2 (11.8–15.2) years, median age at diagnosis was 15.3 (13.8–16.2) years, and median diagnostic delay was 11.9 (3.6–23.0) months. Kahn criteria were fulfilled in 52 patients (89.7%). The most common cutaneous manifestations were acne conglobata (67.2%), acne fulminans (50.0%), and palmoplantar pustulosis (24.1%). Isotretinoin exposure before diagnosis/treatment was recorded in 25 patients (43.1%); among them, arthralgia, sacroiliitis, and acne fulminans following exposure were observed in 60.0%, 56.0%, and 44.0%, respectively. Musculoskeletal pain was present in 57 patients (98.3%). The most frequent osteoarticular sites were the sacroiliac joints (56.9%), anterior chest wall (51.7%), femur (44.8%), thoracic spine (31.0%), and tibia (25.9%). Patients with sacroiliitis more frequently had spinal involvement [31/33 (93.9%) vs. 4/25 (16.0%), $p<0.001$]. Anti-TNF therapy was administered to 39 patients (67.2%) and was more common in patients with sacroiliitis [26/33 (78.8%) vs. 13/25 (52.0%), $p=0.031$] and prior isotretinoin exposure [21/25 (84.0%) vs. 18/33 (54.5%), $p=0.018$]. Among patients with available treatment-response data, TNF inhibitors accounted for most treatments associated with best clinical response (65.5%), mainly adalimumab (32.7%) and etanercept (30.9%). CRP, ESR, pain VAS, and skin VAS significantly improved from baseline to last visit (all $p<0.001$).
**Conclusion**
Pediatric SAPHO spectrum disease in this nationwide cohort was characterized by an acne-dominant phenotype, frequent sacroiliac/axial involvement, and substantial anti-TNF use. The association between sacroiliitis and spinal involvement suggests an axial-dominant pattern in a subset of patients. High isotretinoin exposure and its association with anti-TNF use warrant further investigation.
**Disclosure of interest**
None declared.

## P120

### Beyond CNO: multifocal bone lesions as the first clue to Hodgkin lymphoma

P. Mikolajski[1], V. Manohar[2], G. Thompson[3], I. Elbeshlawi[4], S. L. Chung[3], I. Turtsevich[1,*]
[1]Paediatric Rheumatology; [2]Histopathology; [3]Radiology; [4]Paediatric Oncology, Oxford University Hospitals, Oxford, UK
**Correspondence:** I. Turtsevich
*Pediatric Rheumatology 2026*, **24(S1):**P120

**Introduction**
Chronic nonbacterial osteomyelitis (CNO) is a rare autoinflammatory bone disorder affecting children and adolescents. It is characterised by sterile, multifocal inflammatory bone lesions, commonly involving long bones, pelvis, clavicle, and vertebrae(1). Clinical features include bone pain, swelling, and variable functional impairment, often without systemic illness(1).There are no definitive diagnostic biomarkers. CNO remains a diagnosis of exclusion, requiring careful evaluation to rule out infection or malignancy. Hodgkin lymphoma (HL) accounts for 10–15% of childhood malignancies and is the third most common paediatric cancer(2). It typically affects adolescents and is characterised by Reed–Sternberg cells. HL usually presents with painless lymphadenopathy,

often cervical or mediastinal, and may be accompanied by systemic "B symptoms": fever, night sweats, and weight loss(3). Although extranodal involvement can occur, primary skeletal disease is rare and may complicate diagnosis(4). The overlap in clinical presentation and imaging findings between HL with skeletal involvement and CNO can create diagnostic challenges, particularly when initial imaging demonstrates multifocal marrow lesions without obvious lymphadenopathy(4).

**Objectives**

To emphasise the importance of thorough evaluation and exclusion of malignancy in children presenting with multifocal bone lesions suggestive of CNO, especially in the presence of systemic or atypical lab features.

**Methods**

We report a case of a 15 year old girl presenting with fever and progressive multifocal pain in her hips and lower back. Lab investigations revealed anaemia, thrombocytosis, and markedly elevated inflammatory markers, suggesting inflammatory or infectious process. Targeted magnetic resonance imaging (MRI) demonstrated multifocal marrow based lesions of the pelvis and spine, initially interpreted as CNO. Given the diagnostic uncertainty, a whole-body MRI was performed identifying previously unrecognised mediastinal and cervical lymphadenopathy. Lymph node biopsy confirmed classical HL, nodular sclerosis subtype. Staging with positron emission tomography–computed tomography (PET-CT) demonstrated stage IV disease with extensive skeletal involvement, consistent with advanced HL. This case illustrates limitations of relying on imaging and highlights the role of comprehensive assessment, including systemic evaluation and tissue diagnosis in differentiating CNO from malignancy.

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**Disclosure of interest**

None declared.

## P121

### Infantile cortical hyperostosis mimicking osteomyelitis: an atypical case in a 1-month-old infant


K. Temelkova[1,*], M. Ganeva[1], T. Vasilev[1], D. Kibarova-Hristova[1], S. Stefanov[2]
[1]Pediatric rheumatology, SHAT of children diseases - Sofia; [2]Department of pediatrics, Medical University, Sofia, Bulgaria
**Correspondence:** K. Temelkova

*Pediatric Rheumatology 2026*, **24(S1):**P121

**Introduction**

Infantile cortical hyperostosis (Caffey disease) is a rare inflammatory disorder of infancy characterized by fever, soft tissue swelling, irritability, and cortical hyperostosis. It is classically associated with pathogenic variants in *COL1A1*[(1)] .We report an atypical neonatal presentation associated with a homozygous *AHSG* variant.

**Objectives**

To describe a diagnostically challenging case of infantile cortical hyperostosis mimicking osteomyelitis and to highlight a possible alternative genetic background of the disease.

**Methods**

Clinical data, laboratory findings, imaging studies, microbiological investigations, and genetic analyses were retrospectively reviewed.

**Results**

A 1-month-old boy presented with progressive bilateral periorbital edema and low grade fever. Laboratory evaluation demonstrated marked systemic inflammation with leukocytosis, thrombocytosis, ESR 82 mm/h, and CRP 232 mg/L. Extensive microbiological investigations were negative and initial imaging studies were unrevealing. Bone marrow aspiration excluded malignancy.Further diagnostic evaluation with CT demonstrated a lesion involving the left iliac bone with periosteal reaction and adjacent soft tissue changes. Pelvic MRI showed bone marrow edema, lamellar periosteal reaction, and surrounding soft tissue edema initially suggestive of osteomyelitis. Despite empirical antibiotic therapy, the patient developed recurrent febrile episodes and swelling in the right parotid region, later becoming bilateral. Repeat imaging revealed massive periosteal new bone formation of the mandible with preserved cortex and absence of bone destruction, highly suggestive of Caffey disease. Genetic testing did not identify pathogenic variants in COL1A1. However, a homozygous variant of uncertain significance, c.4A> T (p.Lys2Ter), was detected in AHSG, encoding fetuin-A, a protein involved in bone mineralization and inflammatory regulation. Both parents were heterozygous carriers. To our knowledge, this is the second reported case linking an AHSG variant to a Caffey disease phenotype (2).

**Conclusion**

Infantile cortical hyperostosis should be considered in infants presenting with sterile inflammation, periosteal reaction, and pseudo-osteomyelitis. Mandibular hyperostosis with preserved cortex was the key diagnostic finding. The absence of COL1A1 mutations together with a homozygous AHSG variant suggests possible genetic heterogeneity in Caffey disease.

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**Trial registration identifying number**

**Disclosure of interest**

None declared.

## P122

### Hearing impairment in patients with fibrodysplasia ossificans progressiva: real-world data from a Russian cohort


V. Matkava[1,*], S. Arsenyeva[1], O. Inozemtseva[1], I. Nikishina[1]
[1]Paediatric, V.A. Nasonova Scientific Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** V. Matkava

*Pediatric Rheumatology 2026*, **24(S1):**P122

**Introduction**

Fibrodysplasia ossificans progressiva (FOP) is an ultra-rare genetic disorder characterized by congenital skeletal malformations and progressive heterotopic ossification due to mutation in the ACVR1 gene. Although hearing impairment has been described as the most common extra-skeletal manifestation of FOP, systematic audiological data remain limited, especially in real-world cohorts with longitudinal follow-up.

**Objectives**

To analyze frequency, type of audiological abnormalities in genetically verified FOP patients.

**Methods**

Pure-tone audiometry was performed in patients with genetically verified FOP followed at our center from May 2023 to May 2026. Audiological assessment was performed approximately once a year in patients aged ≥5 years who were able to cooperate with the examination. Audiometry reports were retrospectively reviewed.

**Results**

A total of 29 patients with FOP were assessed with pure-tone audiometry. Audiometry was done once in 19/29 pts (65.5%), twice in 8/29 (27.6%) and three times in 2/29 (6.9%). The mean age at first audiological examination was 12.6±6.4 y.o., median age was 12 [IQR 8;16] y.o. (min 5; max 32). The most pts (27/93.1%) had the typical c.617G> A (p.Arg206His) variant in the *ACVR1* gene; 2 pts had ultra-rare non-classic variants p.Gly328Glu and p.Gly356Asp. Hearing loss was registered in 13/29 pts (44.8%). Conductive hearing loss was revealed predominantly (in 7/13/53.8% pts); mixed/combined hearing loss was observed in 4/13 (30.8%) pts and sensorineural hearing loss in 2/13 (15.4%) pts. Three pts (10.3%) had isolated threshold elevation without established

hearing loss. Overall, audiological abnormalities were detected in 16/29 pts (55.2%). Among 10 pts with annual repeated audiograms, new hearing loss was reported in 4 pts (2 – conductive, 1 – combined, 1 - threshold elevation); 1 pt demonstrated positive dynamic (bilateral hearing loss to unilateral); 4 pts had stable status during follow-up (1 – without abnormalities, 2 – conductive hearing loss, 1 – sensorineural hearing loss). In one patient with tubotitis-associated conductive hearing loss was registered normal audiogram after recovery. One pt with bilateral combined hearing loss stage 3 used a hearing aid.

**Conclusion**

Audiological abnormalities were detected approximately in half of assessed by audiometry patients. Repeated audiograms showed that hearing abnormalities may occur after initially normal assessment. Regular pure-tone audiometry should be included in multidisciplinary monitoring of FOP patients.

**Disclosure of interest**

None declared.

## P123

### Paradoxical interferonopathy-like phenotype following anifrolumab administration in a patient with dermatomyositis

R. M. Alcobendas[1], A. Remesal[1,*], C. Udaondo[1], A. HERCBERG[1], L. Martínez[2], S. Barreiro[1]
[1]Pediatric rheumatology; [2]Hospital La Paz, Madrid, Spain
**Correspondence:** A. Remesal
*Pediatric Rheumatology 2026*, **24(S1):**123

**Introduction**

Anifrolumab is a fully human monoclonal antibody targeting the type I interferon receptor. Its potential benefit has been reported in different conditions, including lupus, dermatomyositis and monogenic interferonopathies.

**Objectives**

To describe the paradoxical development of an interferonopathy-like clinical phenotype following initiation of anifrolumab therapy in a patient with dermatomyositis, and its subsequent resolution after drug withdrawal.

**Methods**

Clinical chart review.

**References**

**Results**

An 11-year-old boy was referred with a diagnosis of anti-TIF1-positive dermatomyositis. At presentation, he exhibited extensive cutaneous involvement, including erythematous-necrotic facial rash involving the nasal tip, thoracic and dorsal rash, Gottron papules on the hands, morphea-like plaques, generalised lipoatrophy, fingertip ulcers, and elbow calcinosis. Previous treatments included methotrexate, intravenous immunoglobulins, tofacitinib, baricitinib, and hydroxychloroquine. He was additionally receiving intravenous anifrolumab. Following initiation of anifrolumab, muscle strength, transaminases and creatine kinase normalised. However, facial lesions, including the nasal tip lesion, significantly worsened and clinically resembled SAVI and Episcleritis (similar to that seen in CANDLE syndrome) developed. Microbiological testing only identified *S. aureus* and *S. pyogenes* in skin swabs, although antibiotics and empirical aciclovir failed to improve the condition. As symptoms consistently worsened following infusions, anifrolumab was discontinued, resulting in complete resolution of ocular and facial lesions. One month later, the patient experienced a disease flare with typical malar rash, muscle weakness, and marked elevation of transaminases and CK, improving after systemic corticosteroid therapy. He is currently receiving intravenous immunoglobulins, hydroxychloroquine, pamidronate, bosentan, nifedipine, cyclophosphamide, rituximab, and tapering corticosteroids, with good clinical evolution and no further relapses. Genetic testing for autoinflammatory diseases is pending. No recurrence of ocular or nasal involvement has been observed since discontinuation of anifrolumab, and chest computed tomography imaging was unremarkable.

**Conclusion**

The limited experience with recently introduced therapies highlights the need for caution in their administration and the importance of reconsidering both diagnosis and therapeutic strategies in cases of unexpected clinical evolution.

**Disclosure of interest**

None declared.

## P125

### Anti-HMGCR positive juvenile dermatomyositis: a physiotherapy case study

A. O'Hare[1,*], S. Ward[1], K. Gallagher[2]
[1]Physiotherapy; [2]Paediatric Rheumatology, National Centre for Paediatric Rheumatology, Children's Health Ireland, Dublin, Ireland
**Correspondence:** A. O'Hare
*Pediatric Rheumatology 2026*, **24(S1):**P125

**Introduction**

Myositis-Specific Autoantibodies (MSAs) are found almost exclusively in patients with idiopathic inflammatory myopathies (IIMs) and can help aid diagnosis and categorise phenotypes. Juvenile dermatomyositis (JDM) is a rare multi-systemic IIM. Documented clinical characteristics of anti-HMGCR JDM include distal muscle weakness, muscle atrophy, falls and fatigue. An individualised physiotherapy management approach is recommended for JDM however it is not currently tailored according to patients' MSAs.

**Objectives**

This case report reviews the physiotherapy assessment and management of a patient with anti-HMGCR positive JDM at diagnosis, during initial rehabilitation phase and at one year follow-up.

**Methods: Case Presentation**

13 year-old male, 6-month history of progressive muscle weakness and regression in physical activity abilities. Proximal muscle weakness, positive Gower's, minimal cutaneous features and no significant arthritis on exam. Initial investigations: significantly elevated creatinine kinase >9,561, elevated liver enzymes, myositis on STIR MRI pelvis, positive anti-HMGCR MSA. Initial Childhood Myositis Assessment Scale (CMAS) score: 34/52 indicating moderate physical disability. Initial Manual Muscle Test 8 (MMT8) score: 43/80. Inpatient length of stay: 32 days. Treated with intravenous steroids followed by oral steroids and Methotrexate. Rituximab and immunoglobulin therapy added due to refractory disease. Patient received daily inpatient physiotherapy and weekly outpatient physiotherapy initially. Physiotherapists re-assessed CMAS and MMT8 weekly during admission, bi-monthly then monthly in outpatients, and at one year follow-up. Psychosocial measures of fatigue and quality of life (QOL) were assessed at diagnosis and at one year follow-up using the Peds QL Multidimensional Fatigue Scale and the Peds QL QOL Inventory. CMAS score of 48/52 indicating no physical disability achieved four months from diagnosis and sustained at one year follow-up. CMAS score of 24/52 with incidence of falls at home prompted readmission for medical and physiotherapy management during initial rehabilitation phase. At one year follow-up, MMT8 scores increased to 69/80. Peds QL Multidimensional Fatigue Scale and Peds QL QOL Inventory scores were 76.38 and 67.45 respectively at diagnosis where higher scores indicate less fatigue and better QOL. Scores at one year follow-up were 84.72 and 84.78 respectively indicating improved physical-psycho-social outcomes. This patient demonstrated several recognised clinical features of anti-HMGCR JDM. Improvements in physical function, fatigue and QOL were observed following combined physiotherapy and medical management. Consideration of MSA profile during rehabilitation may support more individualised physiotherapy management. Further research into MSA-specific clinical phenotypes may help guide future rehabilitation approaches.

**Disclosure of interest**

None declared.

## P126

### Experience with IFNAR1–blocking antibody (anifrolumab) in a patient with treatment–refractory juvenile dermatomyositis

A. Kozlova[*,1], M. Leontieva[1]
[1]Center for the diagnosis and treatment of autoinflammatory diseases, Ilyinskaya Hospital, Moscow, Russian Federation
**Correspondence:** A. Kozlova
*Pediatric Rheumatology 2026*, **24(S1):**P126

**Introduction**

Juvenile dermatomyositis (JDM) is a rare idiopathic inflammatory myopathy of childhood; first–line treatment usually includes corticoste-

roids and conventional immunosuppressants, but complete and sustained remission is not always achieved.

**Objectives**

We present a 10–year–old girl with treatment–refractory JDM. Disease onset occurred at 3.5 years with facial erythema, periorbital edema and telangiectasia, and nasolabial involvement; subsequently she developed periungual capillary changes, progressive proximal muscle weakness, intermittent fever up to 38.0 °C, Gottron's papules over the elbows and knees, and palatal erythema. Laboratory tests showed elevated creatine phosphokinase (CPK), lactate dehydrogenase (LDH) and transaminases. Despite sequential therapy with systemic corticosteroids, methotrexate, intravenous immunoglobulin (IVIG), mycophenolate mofetil, tofacitinib, adalimumab, upadacitinib and abatacept, clinical symptoms progressed with development of numerous subcutaneous calcifications that limited mobility and caused skin breakdown. Whole–genome sequencing (trio) identified a heterozygous frameshift variant in TRPV3 (c.1132del, p.Thr378Serfs*41); the variant was also present in the asymptomatic father. Interferon–I stimulated gene (ISG) expression was markedly elevated at 8.7 (normal range 0.57–1.9).

**Methods**

Treatment with rituximab and anifrolumab was initiated while continuing tofacitinib and periodic IVIG. Following introduction of anifrolumab, the patient demonstrated substantial clinical improvement: increased muscle strength, marked regression of skin disease and reduction of symptomatic calcifications; ISG expression decreased to 1.85 at six months.

**Conclusion**

In this patient with treatment–refractory JDM and marked IFN–I pathway activation, blockade of the IFNAR1 receptor with anifrolumab was associated with significant clinical and biomarker improvement. Anifrolumab may represent a promising therapeutic option for selected refractory JDM cases with elevated IFN–I signalling.

**Disclosure of interest**

None declared.

## P127

### Heterogeneity of serological markers in juvenile dermatomyositis: correlation with clinical features

D. T. Francheshkova[1,*], K. Temelkova[1], T. Vasilev[1], D. Kibarova-Hristova[1], S. Stefanov[1], A. Dasheva[1]
[1]Department of Pediatrics, Medical University of Sofia, Sofia, Bulgaria
**Correspondence:** D. T. Francheshkova
*Pediatric Rheumatology 2026*, **24(S1):**P127

**Introduction**

Juvenile dermatomyositis (JDM) is a rare autoimmune inflammatory myopathy, with a typical female predominance. Antinuclear antibodies (ANA), Myositis-specific antibodies (MSAs) and Myositis-associated antibodies (MAAs) play important roles in the disease heterogeneity and prognosis.

**Objectives**

To assess the serological profiles and the associated clinical features of a small cohort of JDM patients.

**Methods**

A five year retrospective single-center analysis of nine JDM patients (7 males, 2 females) was conducted. Serum concentrations of ANA, MSAs and MAAs were measured by enzyme-linked immunosorbent assays. Clinical symptoms and laboratory data were obtained from clinical records. Clinical characteristics were compared between seropositive and seronegative patients.

**Results**

MSAs positivity was identified in 4 patients (44.4%), MAAs positivity in 1 patient (11.1%), yielding an overall antibody-positive rate of 55.56%. 44.44% lacked detectable MSAs or MAAs, consistent with reported seronegative incidence in JDM [1]. ANA positivity was observed in all but one case. Antibody distribution comprised of: anti-TIF1-γ (2/9, 22.2%), anti-MDA5 (2/9, 22.2%), anti-Mi-2 (1/9, 11.1%), anti-NXP2 (1/9, 11.1%) and anti-Ro52, anti-PM-Scl (1/9, 11.1%), paralleling frequencies reported in larger cohorts [2]. The anti-NXP2-positive patient displayed severe muscle weakness, dysphagia and calcinosis. The anti-TIF1-γ-positive patients had classic dermatological findings. Among anti-MDA5-positive individuals, one exhibited predominant cutaneous involvement with arthritis and mild myopathy, while the other (also anti-Mi-2-positive) showed severe muscle and skin disease. No interstitial lung disease was detected in anti-MDA5- or MAA-positive patients. The MAA-positive patient (anti-Ro52, anti-PM-Scl) presented with severe myopathy and typical cutaneous features without clinical features suggesting scleroderma overlap. Among seronegative patients one had severe muscle weakness with minimal skin involvement and three exhibited mild myopathy with prominent rashes. Two of the latter developed calcinosis during follow-up. These findings align with global antibody-phenotype correlations in JDM [2].

**Conclusion**

This cohort reveals a male predominance not typically reported in JDM, suggesting potential population-specific variations. The serological status and clinical features are consistent with international findings, emphasizing the utility of comprehensive serological profiling for personalised prognosis and management.

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**Disclosure of interest**

None declared.

## P128

### JAK inhibitors in refractory juvenile dermatomyositis: a single-center case series of patients treated with baricitinib and TNF inhibitor

M. Manojlović[1,*], M. Jakovljevic[1], T. Stankovic[1,2], D. Lazarevic[1], J. Vojinovic[1,2]
[1]Department of Rheumatology, Clinic for Pediatrics, University Clinical Center of Nis; [2]University of Nis, Faculty of Medicine, Nis, Serbia
**Correspondence:** M. Manojlović
*Pediatric Rheumatology 2026*, **24(S1):**P128

**Introduction**

Juvenile dermatomyositis (JDM) is characterized by muscle inflammation and skin involvement, with heterogeneous phenotypes and potentially severe refractory courses despite conventional sDMARDs and anti-TNF therapy. Increasing evidence supports the type I interferon pathway activation, providing a rationale for Janus kinase (JAK) inhibition. However, data on baricitinib in pediatric patients remain limited.

**Objectives**

To evaluate the efficacy and safety of baricitinib in a single-center cohort of patients with refractory JDM treated with anti-TNF, methotrexate and baricitinib.

**Methods**

We present a case series of three patients with refractory JDM treated with anti TNF and methotrexate, in tertiary referral center. All patients had long-standing disease and prior exposure to corticosteroids, methotrexate, intravenous immunoglobulins, and biologic agents (anti-TNF) with inadequate response. Phenotypes included: one patient with extensive calcinosis, one with severe corticosteroid-related toxicity (Cushingoid features, hypertension, obesity and cataract), and one with overlap JDM/scleromyositis phenotype with anti-PM/Scl 75 positivity, severe skin involvement, contractures and high risk for interstitial lung disease. Baricitinib was used off-label based on emerging evidence supporting JAK inhibition in interferon-driven myopathies. It was initiated at 2 mg daily in one patient and 4 mg daily in two patients. Clinical outcomes, including muscle strength, cutaneous involvement, and functional status, were followed up.

**Results**

All three patients demonstrated significant clinical improvement following baricitinib initiation. Median treatment duration was 12 (range 10-30) months, with a median age of 9.4 (range 7.8-11.8) years, female-to-male ratio of 1:2. Cutaneous disease activity was assessed using CDASI, demonstrating a marked reduction of approximately 15-25 points (60-80%), corresponding to a shift from moderate-severe to mild disease. Steroid-free disease control was achieved across all cases, despite different phenotypes. Despite multiple prior flares and treatment

failures, baricitinib led to improved muscle strength, functional status, and cutaneous disease control in all patients. No serious adverse events were observed.

**Conclusion**

Baricitinib may represent an effective and well-tolerated therapeutic option in refractory JDM, enabling steroid-free disease control across diverse severe phenotypes. Further prospective and multicenter studies are warranted to confirm these findings, including long-term efficacy and safety.

**Disclosure of interest**

None declared.

## P129

### Spectrum of renal involvement in anti-MDA-5 antibody-positive juvenile dermatomyositis: a case series

P. Kaur[1,*], M. Kulandaisamy [1], A. Kollana [1], J. Fartiyal[2], A. W. Barwad[2], G. Singh[2], S. Paraselli [1], M. Yadav[1], S. Mehaboob[3], V. Gupta [3], N. Bagri[1]
[1]Pediatrics; [2]Pathology; [3]Dermatology, All India Institute of Medical Sciences, New Delhi, New Delhi, India
**Correspondence:** P. Kaur
*Pediatric Rheumatology 2026*, **24(S1):**P129

**Introduction**

Anti-melanoma differentiation-associated gene 5 (anti-MDA5) antibody-positive juvenile dermatomyositis (JDM) is characterised by a distinct clinical phenotype, including ulcers, amyopathy, and interstitial lung disease (ILD). Barring anecdotal reports[1–3]. Renal involvement in JDM is rare and has not been described in children with anti-MDA-5 antibody-positive JDM.

**Objectives**

To review clinical features and renal involvement in children with anti-MDA5 antibody-positive JDM.

**Methods**

Children with anti-MDA5 antibody-positive JDM with renal involvement were screened from the Pediatric Rheumatology Clinic Register (Jan 2020-March 2026), All India Institute of Medical Sciences, New Delhi.

**Results**

Four children were identified. All presented with cutaneous features, muscle weakness, elevated muscle enzymes, ILD, and renal manifestations (Table). All tested negative for anti-nuclear antibodies (except patient 4) and anti-dsDNA antibodies, with normal complement levels at presentation. Kidney biopsies revealed thrombotic microangiopathy (TMA) in patients 1 and 2 and IgA nephropathy in 3. Biopsy was deferred in patient 4 as the sub-nephrotic proteinuria resolved. All patients were initially treated with IV steroids. After initial treatment, patient 1 presented with TMA, in hypertensive emergency, with acute kidney injury (AKI), hemolysis, and thrombocytopenia, and additionally required therapeutic plasma exchange (PEX). After five sessions, she had clinical improvement.

**Table ( Abstract P129)**

| Parameter | Patient 1 | Patient 2 | Patient 3 | Patient 4 |
|---|---|---|---|---|
| Age at onset, years | 15 | 11 | 16 | 9 |
| Gender | Female | Female | Male | Female |
| Disease duration, months | 12 | 9 | 22 | 6 |
| CMAS at presentation | 42 | 39 | 35 | 13 |
| Extramuscular features | ILD (organising pneumonia) | | | |
| Lung | | | | |
| Renal | Anasarca, hypertensive emergency (seizures), oliguric AKI, proteinuria, microscopic hematuria, hematologic TMA | Hypertension, proteinuria | Proteinuria, microscopic hematuria | Hypertension, proteinuria |
| Baseline CPK (U/L) | 460 | 352 | 323 | 456 |
| Urine protein | 3+ | Nil | 2+ | Nil |
| Urine RBC | 2–3/hpf | Nil | 5–10/hpf | Nil |
| 24-hour urine protein (mg/day) | 8542 | 1683 | 904 | 425 |
| Renal biopsy | Subacute TMA with segmental sclerosis | Chronic TMA with FSGS | IgA nephropathy | Deferred |
| Immunomodulation | IVMP, IVCP, PEX | IVMP, IVCP | IV dexamethasone, azathioprine | IVMP, IVCP |
| Follow-up duration, months | 3 | 2 | 3 | 3 |
| CMAS | 52 | 45 | 38 | 40 |
| 24-hour urine protein (mg/day) | 442 | 260 | 21 | 250 |

**Conclusion**

Children with anti-MDA5-positive JDM may experience a spectrum of renal involvement, ranging from mild IgA nephropathy to life-threatening TMA. The presence of hypertension, hemolysis, AKI, proteinuria, or hematuria should prompt evaluation for renal involvement for timely therapy.

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**Disclosure of interest**

None declared.

## P130

### Extramuscular and extracutaneous manifestations of juvenile dermatomyositis: vasculopathy beyond the muscles

P. Kaur[1,*], N. Shaikh[2], R. Chawla[2], N. Bagri[1]
[1]Pediatrics; [2]Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India, All India Institute of Medical Sciences, New Delhi, New Delhi, India
**Correspondence:** P. Kaur
*Pediatric Rheumatology 2026*, **24(S1):**P130

**Introduction**

Juvenile Dermatomyositis (JDM) is an inflammatory vasculopathy that presents with characteristic muscular and cutaneous features in more than 95% of patients. However, its potential for systemic involvement is less recognised and underreported.

**Objectives**

To investigate the frequency and types of extramuscular and extracutaneous manifestations in children with JDM enrolled in the pediatric rheumatology clinic of AIIMS, New Delhi.

**Methods**
Case records of children with juvenile dermatomyositis (JDM) enrolled in the pediatric rheumatology clinic of our tertiary care hospital (2012-2025) were screened to identify children with extramuscular/cutaneous disease manifestations.
**Results**
Of the 123 children with JDM enrolled between 2012 and 2025, four extramusculocutaneous manifestations were identified: cardiac, gastrointestinal, macrophage activation, and ocular.
**Case 1**
A 9-year-old boy who presented with dilated cardiomyopathy and cardiogenic shock, with a 30% ejection fraction. He was supported with decongestive measures and inotropic support. IV methylprednisolone (IVMP), methotrexate, escalated to mycophenolate mofetil (MMF), were administered for disease. Six years into follow-up, he is now off any cardioactive drugs with a normal biventricular function (EF=65%).
**Case 2**
A 9-year-old girl with severe ulcerative lesions, abdominal pain, and upper gastrointestinal bleeding attributed to mesenteric ischemia and acute pancreatitis secondary to vasculopathy. The course was further complicated by macrophage activation syndrome. Hence, she was intensively managed with IVMP pulse, IV cyclophosphamide (IVCP), and subsequently monthly IVIg. She had a severe refractory disease course requiring readmission and further intensification with Rituximab seven months into follow-up. She developed persistent lymphopenia, macrophage activation syndrome (MAS) and severe gastrointestinal bleeding thereafter. Endoscopic evaluation revealed oesophagal ulcers, with candida growth on ulcer biopsy culture. Sepsis and MAS improved with antifungals and cyclosporine therapy, respectively.
**Case 3**
An 11-year-old girl with progressive painless blurring of vision. Evaluation revealed pseudobulbar palsy, elevated creatine kinase and bilateral optic atrophy with maculopathy. She was managed with IVMP pulse and methotrexate. Two years into follow-up, her muscular weakness improved and visual acuity stabilized, with no further deterioration.
**Conclusion**
Juvenile Dermatomyositis is an autoimmune inflammatory myositis, perceived to characteristically involve striated muscles and skin. However, it is indeed a multisystemic vasculopathy that may present with potentially life-threatening extramuscular and extracutaneous manifestations, including myocardial dysfunction, macrophage activation syndrome and neurological deficits, which are possibly amenable to timely immunomodulation.

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**Disclosure of interest**
None declared.

## P131

**Early onset juvenile dermatomyositis: phenotype disparities and novel genetic findings**

Z. Hilal[1,*], A. Alasmari[1], A. Alsonbul[1], S. M. Al-Mayouf[1,2], A. Alsaleem[1]
[1]Pediatric Rheumatology, King Faisal Specialist Hospital and Resarch Center; [2]Pediatric Rheumatology, College of Medicine, Alfaisal University, Riyadh, Saudi Arabia
**Correspondence:** Z. Hilal
*Pediatric Rheumatology 2026*, **24(S1):**P131

**Introduction**
Juvenile dermatomyositis is a rare, chronic multisystem inflammatory disease of unclear origin. Characterized by proximal muscle weakness and characteristic cutaneous manifestations. Early-onset disease may represent a distinct clinical subset, but data remain limited. Previous studies suggested that early onset JDM may have diagnosis delay and more sever disease course.
**Objectives**
To determine whether early onset JDM differ from late onset JDM in terms of clinical features, treatment, and outcome measures.Also, to report the genotype findings in the early onset group.
**Methods**
A retrospective, observational, cross-sectional study was conducted. using data from our pediatric Rheumatology myositis clinic between June 1997 and February 2026. All patients fulfilled Bohan and peter criteria; were categorized into early onset (≤5years) and late onset (> 5 years) groups based on age of disease onset. Medical records were reviewed for demographic, family history, clinical and genetic data, therapy and outcome measures.
**Results**
Total of 35 patients were included: 18 with early onset and 17 with late-onset. Median age of disease onset was of 4.0 years (IQR 2.0-3.0) in the early onset group and 10.5 years (IQR 6.0-8.25) in the late-onset group. Female predominance was observed in both groups. Constitutional symptoms, lipodystrophy, cutaneous ulceration, and calcinosis were more frequent in the early onset group, whereas Raynaud's phenomenon was seen more in the late onset. Facial swelling, heliotropic rash, and Gottron papules were comparable in both groups. Both groups showed comparable Serologic profiles of myositis specific autoantibodies. Corticosteroids, and methotrexate were the most frequent medications used in both groups. While IVIG was more commonly used in early-onset disease. Patients with early onset disease required biological agents, more frequent. Eleven patients with early onset disease completed genetic testing, and multiple candidate variants of uncertain significance were identified. Genetic testing in the early-onset group identified several candidate variants, including (*HLA-DQB1*, *ERCC5*, *HADHB*, *VCAN*, *FLG*, *HFE*, *UFC1*, *ACADSB)*; supporting the possibility that genetic factors may contribute to phenotypic heterogeneity in a subset of patients. Early onset had higher mean myositis damage index (related to calcinosis and lipodystrophy) at last follow up visit. There was no death reported in Early onset group however, a single mortality occurred in the late-onset group.
**Conclusion**
Our study showed that early onset JDM exhibit prominent systemic inflammation, more cutaneous manifestations, lipodystrophy, calcinosis, and the need of higher treatment escalation. We have identified novel genetic findings with secondary candidate variants which may explain the phenotypic disparity.

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**Disclosure of interest**
None declared.

## P132

**Interferon signature in juvenile dermatomyositis: a single-center**

E. Aslan[1,*], S. Firtina[2], Z. Torunoglu[1], N. Akay[1], U. Gul[1], E. Kilic Konte[1], M. Yildiz[1], S. Sahin[1], K. Barut[1], O. Kasapcopur[1]
[1]Pediatric Rheumatology; [2]Medical Genetics, Istanbul University- Cerrahpasa, Istanbul, Türkiye
**Correspondence:** E. Aslan
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**Introduction**
Juvenile dermatomyositis (JDM) is a heterogeneous autoimmune disease in which type I interferon (IFN) activation plays a pivotal role in its pathogenesis.
**Objectives**
Aim of this study is, to evaluate the expression of interferon-related genes in patients with JDM and to investigate their association with disease activity scores.
**Methods**
Eleven patients with a diagnosis of JDM were included in the study. The expression of four interferon-related genes (IRG) was assessed (ISG15, IFI27, IFIT1, and IFI44L) by real-time quantitative-PCR from peripheral blood; Ct values were normalized to the ACTB reference gene. Patient

**Table 1 (Abstract P132)** Correlation of IFN signature and Disease Activity Scores

| | ISG15 | IFI27 | IFIT1 | IFI44L | Total IFN score |
|---|---|---|---|---|---|
| PhG VAS (0-10) | +0.08 | +0.16 | +0.04 | +0.14 | +0.13 |
| PaG VAS (0-10) | +0.39 | +0.35 | +0.37 | +0.40 | +0.38 |
| Skin VAS (0-10) | +0,30 | **+0,62*** | **+0,66*** | **+0,64*** | **+0,71*** |
| Skin DAS (0-9) | +0,28 | +0,59 | **+0,63*** | **+0,61*** | **+0,68*** |
| CMAS (0-52) | **-0.66*** | **-0.63*** | **-0.70*** | **-0.68*** | **-0.72*** |
| MMT8 (0-150) | **-0.64*** | **-0.61*** | **-0.69*** | **-0.66*** | **-0.71*** |
| JDMAI1 (0-30) | **+0.71*** | **+0.67*** | **+0.69*** | **+0.73*** | **+0.76*** |
| JDMAI2 (0-30) | **+0.66*** | **+0.63*** | **+0.65*** | **+0.68*** | **+0.72*** |

data were converted to z-scores relative to healthy control references. Disease activity scores were recorded at the time of sample collection.

**Results**

Among 11 JDM patients, 27% were female. Median age at diagnosis was 10.3 years. 73% were myopathic, 27% were clinically amyopathic JDM. Calcinosis was present in 36% ($n$=4) and interstitial lung disease in 18% ($n$=2). Disease activity measures at the time of sampling showed a median skin VAS of 5 (IQR 4–8), skin DAS of 4 (IQR 3–6), CMAS of 52 (IQR 47.5–52), MMT8 of 150 (IQR 96–150). Mean z-scores were elevated for all IRGs in the JDM cohort: 8.6 for ISG15, 71.6 for IFI27, 36.8 for IFIT1, and 43.8 for IFI44L. The total interferon z-score was calculated as 40.2.

In correlation analyses between IFN gene expressions and disease activity measures, the total interferon z-score demonstrated strong correlations with skin VAS ($\rho = +0.71$), skin DAS ($\rho = +0.68$), CMAS ($\rho = -0.66$), and MMT8 ($\rho = -0.71$). All IRGs showed strong negative correlation with CMAS and MMT8. All IRGs except ISG15 showed at least moderate positive correlation with skin VAS and DAS. (Table 1)

**Conclusion**

In this study, the IFN signature was markedly elevated in all patients with JDM. In this cohort, where cutaneous involvement was more prominent than myopathy, IFN activation correlated with both muscular and cutaneous disease activity scores. Notably, while ISG15 expression correlated with myositis parameters such as CMAS and MMT8, the absence of correlation with skin disease activity suggests this gene may reflect a muscle dominant interferon response. Our findings support the potential utility of the IFN signature as a tool for defining biological subgroups and monitoring disease activity in JDM.

**Disclosure of interest**

None declared.

## P134

### Treatment-free remission in juvenile dermatomyositis: a single-center experience

H. K. Zora[*,1], S. S. Kilic[1]
[1]Department of Pediatric Rheumatology, Faculty of Medicine, Bursa, Türkiye
**Correspondence:** H. K. Zora
*Pediatric Rheumatology 2026*, **24(S1):**P134

**Introduction**

Juvenile dermatomyositis (JDM) is a rare inflammatory myopathy of childhood with a heterogeneous clinical course and treatment response (1). Predictors of treatment-free remission in JDM remain unclear.

**Objectives**

To evaluate the clinical and laboratory characteristics of JDM patients and identify features associated with treatmen-free remission.

**Methods**

Medical records of 26 patients with JDM followed between January 2016 and February 2024 were retrospectively reviewed. All patients fulfilled the Bohan and Peter criteria. Demographic, clinical, laboratory, treatment, and remission data were recorded. Treatment-free remission was defined as inactive disease for at least 6 months after discontinuation of all systemic immunosuppressive therapies. Patients were divided into two groups: treatment-free remission and remission on treatment/active disease. Muscle strength was assessed using the Childhood Myositis Assessment Scale (CMAS).

**Results**

Of the 26 patients, 69.2% were female. Treatment-free remission was achieved in 11 patients (42.3%). Female sex was significantly more common in the treatment-free remission group compared to the remission on treatment/active disease group (90.9% vs. 53.3%, $p$=0.040). No significant differences were found regarding age at symptom onset, age at diagnosis, or diagnostic delay. Clinical manifestations, organ involvement, muscle enzyme levels, CMAS scores, and relapse rates were similar between groups. Electromyography positivity was significantly more frequent in the remission on treatment/active disease group (86.7% vs. 45.5%, $p$=0.029). Myositis-specific autoantibody positivity tended to be lower in patients achieving medication-free remission. Adalimumab use was significantly more common in the remission on treatment/active disease group (40% vs. 0%, $p$=0.017), while no differences were observed for other immunosuppressive or biologic therapies.

**Conclusion**

Patients with JDM who achieved treatment-free remission were characterized by a higher female predominance, while clinical and laboratory features were largely similar between groups. Higher rates of electromyography positivity and biologic treatment use in patients without treatment-free remission may reflect a more severe or treatment-resistant disease phenotype. Myositis-specific autoantibody positivity may also be associated with lower rates of treatment-free remission.

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**Disclosure of interest**

None declared.

## P138

### Natural killer cell profile at the onset of juvenile dermatomyositis: immunological correlations and functional implications in a monocentric pilot study

L. Capra Mattioli[1,*], S. Mazzitelli[2], F. Vianello[2], F. Di Stasio[2], P. Vaienna[1], F. Minoia[2], D. Montin[2], G. Filocamo[2], M. Rossano[2]
[1]Università degli studi di Milano; [2]Pediatric Immuno-Rheumatology Unit, Fondazione IRCCS Cà Granda Ospedale Maggiore Policlinico, Milan, Italy
**Correspondence:** L. Capra Mattioli
*Pediatric Rheumatology 2026*, **24(S1):**P138

**Introduction**

Juvenile Dermatomyositis (JDM) is a rare systemic autoimmune disease characterized by muscle and skin involvment. While the pathogenic role of Natural Killer cells (NK) in adult dermatomyositis is well-documented, evidence in JDM remains limited. Recent studies have shown that reduced peripheral NK cell counts are associated with muscle weakness in untreated pediatric patients, suggesting their potential role as biomarkers. Furthermore, it has been observed that the reduction of circulating NK cells is linked to interferon activation. However, the role of NK cells at disease onset and its relationship with clinical features and disease course remain poorly defined.

**Objectives**

To characterize peripheral NK cell counts at disease onset in a monocentric cohort of patients with JDM and to investigate their association with clinical, laboratory, and therapeutic parameters.

**Methods**

A retrospective analysis was conducted on 13 treatment-naïve JDM patients at the time of diagnosis. Absolute counts and percentages of lymphocyte subpopulations (CD3+; CD3+CD4+; CD3+CD8+; CD19+; CD3-CD16+CD56+) were measured alongside inflammatory markersindices of myocytolysisand muscle strength assessed via the hMC score. Additionally, the number of treatments received by the patient during follow-up was analyzed, as marker of therapeutic complexity. Statistical analysis was performed using non-parametric tests (Kruskal-Wallis, Mann-Whitney) and Spearman's correlation.

**Results**

A peripheral depletion of NK was observed at JDM onset in 11 out of 13 patients (mean 61.6/uL ± 34.7). Patients without overt muscle involve-

ment had higher mean NK counts than those with myositis (78 vs. 58.6 cells/μL), although the difference was not statistically significant ($p$ = 0.118). Furthermore, no significant differences emerged between NK values and skin involvement ($p$ > 0.6). On the other hand, a correlation was highlighted between elevated NK values and the severity of skin disease at onset, expressed as skin VAS ($r$ = 0.71; $p$ < 0.05). The trend in which higher CK values correlate with lower NK values was confirmed ($r$ = -0.32, $p$ = 0.29). Additionally, the analysis highlighted a trend indicating that patients with lower NK counts required more lines of treatment ($r$ = -0.40, $p$ = 0.18).

**Conclusion**

Peripheral NK cell depletion appears to be a consistent immunological feature of JDM at disease onset. The significant correlation with skin severity and the trends observed with muscle involvement and therapeutic complexity suggest that the NK profile may represent an early biomarker of activity and prognosis. Our preliminary data support the integration of immunological monitoring into JDM management, to identify distinct subsets and risk stratifications. Larger prospective studies in well-characterized cohorts are needed to confirm these observations and better define the pathogenic and prognostic role of NK cells in JDM.

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**Disclosure of interest**

None declared.

## P139

### Anti-HMGCR antibody associated immune-mediated necrotizing myopathy triggered by **Salmonella spp** infection in a 12-year-old girl

K. Zarra[1], M. Tsinti[1,*], E. Tsitsami[1]
[1]Pediatric Rheumatology Unit, First Department of Pediatrics, National and Kapodistrian University of Athens Medical Schoo, "Agia Sofia" Children's Hospital, Athens, Greece
**Correspondence:** M. Tsinti
*Pediatric Rheumatology 2026*, **24(S1):**P139

**Introduction**

Immune-mediated necrotizing myopathy (IMNM) is a rare, severe antibody-defined subset of idiopathic inflammatory myopathies (IIM) characterized by rapidly progressive proximal muscle weakness and markedly elevated creatine kinase (CK) levels, resulting to disability if left untreated. Two principal autoantibody subtypes-anti-3-hydroxy-3-methylglutaryl-coenzyme-A-reductase (anti-HMGCR) and anti-signal recognition particle (anti-SRP) define distinct clinical phenotypes, representing 4% and 1% of juvenile IIM respectively. Anti-HMGCR myopathy is associated with statin exposure in adults but not in children. MRI usually reveals diffuse, symmetric edema of the thigh and gluteal muscles, correlating with myofiber necrosis.

**Objectives**

To highlight the role of unique laboratory and imaging features that lead to early diagnosis of a**nti-HMGCR associated** IMNM.

**Methods**

Case presentation.

**References**

A 12-year-old girl was referred for fatigue, proximal muscle weakness, CK increase 16,906 IU/L and ESR 60 mm/hr, 15 days after febrile *Salmonella spp enteritis.* MRI revealed edema in the thigh muscle groups. Diagnosis of IMNM was corroborated by detection of anti-HMGCR autoantibodies (160 IU/ml). Despite reporting mild dysphagia, no gastrointestinal dysmotility was detected by functional and imaging studies. Respiratory distress was absent however plethysmography was suggestive of respiratory muscle weakness. Cardiac function was unaffected. Prednisolone 60 mg/day and monthly IVIG resulted in immediate and significant improvement of muscle strength. CK levels decreased at ~2,000-4000 IU/L. Testing for Myositis-specific/associated autoantibodies revealed anti-HMGCR myopathy. Congenital dystrophies and metabolic causes of necrotizing myopathy were excluded. Follow up MRI 3 months later revealed resolution of edema and presence of muscle atrophy. Muscle biopsy revealed muscle fiber necrosis. Intravenous rituximab 1 gr 2 doses fortnightly was added and physiotherapy was initiated. Prednisolone was weaned off in 5 months. Muscle strength and daily function returned to near normal, no swallowing difficulties were reported again and respiratory function tests stabilized, however CPK remains at ~2,000 IU/ml.

In this case Salmonella infection probably triggered anti-HMGCR myopathy. P**roximal muscle weakness** significantly elevated CK levels and MRI findings supported the diagnosis of IMNM early in the disease course. Early initiation of immunomodulation lead to. to significant improvement of muscle strength.

**Disclosure of interest**

None declared.

## P140

### Characterization of muscle histology subgroups in juvenile dermatomyositis

M. Matt[1,*], S. O'Connor [1], L. Auld[1], H. Brunner[1], G. Schulert[1], S. Angeles-Han[1]
[1]Rheumatology, Cincinnati Children's Hospital Medical Center, Cincinnati, USA
**Correspondence:** M. Matt
*Pediatric Rheumatology 2026*, **24(S1):**P140

**Introduction**

Few studies in children with juvenile dermatomyositis (JDM) have reported whether histological findings on muscle biopsy are associated with myositis-specific antibody (MSA) status, muscle enzyme levels, and muscle strength testing.

**Objectives**

Evaluate the diversity of muscle biopsy findings in the Cincinnati Myositis Registry (CMR) and correlate muscle histologic pattern with MSAs and muscle enzyme levels.

**Methods**

This retrospective study included children with JDM enrolled in the CMR who underwent a muscle biopsy at time of diagnosis. Relevant data from 2010 to 2026 were extracted from the CMR REDCap database, including laboratory findings, Childhood Myositis Assessment Scale (CMAS) scores, and histology and immunohistochemistry from muscle biopsies. One way ANOVA with subsequent Tukey HSD test was performed to evaluate for differences in CMAS and muscle enzyme levels by histologic subtype. Fisher's exact test was utilized for categorical data. Statistical analysis was performed in R, version 4.4.0.

**Results**

Muscle biopsy data was available for 44/124 children (35%) enrolled in the CMR. This cohort was 73% female. 85% of patients reported white race, alongside 10% black, 4% Asian, and 1% Hispanic. The mean age at diagnosis was 6.3 years. The mean baseline CMAS score was 27.6. The predominant histologic patterns included perifascicular atrophy (PFA: $n$=22, 50%), minimal change ($n$=14, 32%), predominant endomysial and perimysial macrophage infiltration (EMI: $n$=5, 11%), and necrosis/degeneration of muscle fibers ($n$=3, 7%). Immunohistochemical changes of increased MHC-1 expression on myofibers ($n$=8) and reduced capillary density ($n$=4) were seen only in patients with a PFA histological pattern. Electron microscopy data were available from 22 biopsies, and tubuloreticular inclusions in endothelial cells ($n$=9) were only present in patients with PFA pattern. MSA testing was performed in 24 patients and MSAs were detected in eight children; four patients had anti-p155 autoantibodies, two anti-Jo-1, one anti-NXP2, and one patient had anti-HMGCR. No specific pattern of association between MSA type and histologic pattern was appreciated. There was no significant difference in baseline CMAS based on histological pattern. No significant differences in mean baseline levels of muscle enzymes (CK, AST, ALT, aldolase, LDH) were seen among patients with different muscle biopsy histological patterns.

**Conclusion**

Compared to other large studies of muscle histology in children with JDM, we report a greater proportion of patients with predominant perifascicular atrophy (PFA) and fewer patients with EMI. In contrast to

prior work, no significant differences in baseline muscle enzyme levels or MSAs were observed among histologic subtypes. Similar to prior studies, however, there was no significant difference in CMAS score between histologic subtypes.

**Disclosure of interest**
None declared.

P141

**Nailfold capillaroscopy for disease activity assessment in juvenile dermatomyositis**

M. Matt[1,*], H. Wilson[1], L. Auld[1], S. O'Connor [1], T. Ting[1], H. Brunner[1], G. Schulert[1], S. Angeles-Han[1]
[1]Rheumatology, Cincinnati Children's Hospital Medical Center, Cincinnati, USA
**Correspondence:** M. Matt
*Pediatric Rheumatology 2026*, **24(S1):**P141

**Introduction**
Abnormalities in nailfold capillary density and morphology are seen in children with juvenile dermatomyositis (JDM) and may correlate with disease activity.
**Objectives**
Investigate the correlation between nailfold capillaroscopy (NFC) measurement parameters and disease activity level in children with JDM from the Cincinnati Myositis Registry (CMR).
**Methods**
Patients with JDM enrolled in the CMR were approached for NFC. Images of the nailfold capillaries were obtained at 200X magnification using a digital capillaroscope. Two images were obtained from each nailfold from fingers 2-5. A 1 mm square grid was applied to each image, and images were then scored for capillary density, dilated apical limb diameter (> 20 µm), giant capillaries (apical limb diameter > 50 µm), number of abnormal shapes/ramifications, and the presence of microhemorrhages. Nailfold scoring was conducted according to the protocol of the EULAR Study Group on Microcirculation in Rheumatic Diseases. For analysis, we separated patients based on disease activity: active patients had Childhood Myositis Assessment Scale (CMAS) score < 48, elevated muscle enzymes, corticosteroid treatment, or calcinosis; patients in medicated remission had CMAS >48, normal muscle enzymes, and no corticosteroid use. Clinical data were extracted from the CMR REDCap database and statistical analysis was performed in R version 4.4.0. Wilcoxon signed-rank tests were utilized to assess numerical data and Fisher's exact test for categorical variables.
**Results**
Seventeen children with JDM had available NFC images and were included in this study. Ten patients were female. Eight patients met criteria for active JDM, and nine were in medicated remission. The mean age was similar between the two groups. Five patients had calcinosis. The median CMAS score was 50 in active patients and 52 in patients with medicated remission. NFC abnormalities were observed in both groups (Table 1); however, patients with active JDM had reduced capillary density compared to patients in medicated remission. Significantly more giant capillaries were observed in patients with active disease. Active JDM patients also had a greater frequency of dilated and ramified/abnormal blood vessels and more analysis fields with microhemorrhage when compared to patients in medicated remission, but these differences were not statistically significant.

**Table 1 (Abstract P141)** Quantitative and qualitative NFC parameters in JDM patients. Normal capillary density is ≥ 7 capillaries per mm$^3$. Ψ = Wilcoxon signed rank test $p$ <0.05; * = Fisher's exact test $p$<0.05

| | Active JDM | JDM in Medicated Remission |
|---|---|---|
| Capillary Density per mm$^3$; mean (SD) | 4.37 (1.32)Ψ | 5.91 (0.93) |
| **Dilated Apical Loops** per mm$^3$; median (SD) | 0.98 (0.48) | 0.53 (0.73) |
| **Abnormal Shapes** per mm$^3$; median (SD) | 1.06 (0.50) | 0.67 (0.61) |
| **Giant Capillaries** (number of patients) | 5* | 1 |
| **Microhemorrhage** (number of patients) | 6 | 3 |

**Conclusion**
In this single-center study of JDM patients, NFC abnormalities were observed in all patients regardless of disease severity. However, patients with active JDM had significantly reduced capillary density and significantly more giant capillaries when compared to JDM patients in medicated remission. Quantitative and qualitative NFC parameters represent an important, non-invasive means of assessing disease activity in children with JDM.
**Disclosure of interest**
None declared.

P142

**Prevalence of lipodystrophy in juvenile dermatomyositis: a systematic review and meta-analysis**

H. M. Natour[1,*], G. Filocamo[1], J. Cunningham[2], K. Chiu[1], E. Jones[1], K. Whitney[1], B. M. Feldman[1]
[1]Division of Rheumatology; [2]Learning Institute, The Hospital for Sick Children, University of Toronto, Toronto, Canada
**Correspondence:** H. M. Natour
*Pediatric Rheumatology 2026*, **24(S1):**P142

**Introduction**
Juvenile dermatomyositis (JDM) is a rare autoimmune disorder associated with various long-term complications, including acquired lipodystrophy (LD). The reported prevalence of LD in JDM patients varies significantly across individual studies, ranging from 8% to 40%.
**Objectives**
This systematic review and meta-analysis aim to synthesize data from all published studies to provide a more accurate estimate of the overall prevalence and associated metabolic abnormalities.
**Methods**
A comprehensive systematic search of medical databases (e.g., PubMed, EMBASE, Cochrane) was conducted for all studies published in English from inception to October 2025 that reported on the prevalence of LD in patients with JDM. Data from eligible studies were extracted, and a meta-analysis using a random-effects model was performed to determine the pooled prevalence estimate. Subgroup analyses were planned to explore sources of heterogeneity, such as study design (retrospective vs. prospective), geographic location, and specific patient characteristics (e.g., autoantibody status, treatment regimens).
**Results**
A total of 12 studies, comprising 542 JDM patients, met the inclusion criteria. The pooled estimated prevalence of acquired lipodystrophy among patients with JDM was 18.5% (95% CI, 12.1%–26.9%). The prevalence varied between studies, with reported rates ranging from a low of 5% (5/22 patients) to a high of 40% (8/20 patients). Lipodystrophy was more frequently partial (approximately 53%) than generalized (approximately 24%). Metabolic abnormalities were common in the lipodystrophy subgroup; specifically, hypertriglyceridemia was reported in 60% of patients with available data (9/16 patients), and diabetes mellitus/impaired glucose tolerance was found in 40% of cases (6/16 patients). The mean time from JDM diagnosis to LD onset was approximately 34.2 months (range 29-291 months), and LD development was significantly associated with a chronic continuous disease course, calcinosis, and anti-TIF1-γ autoantibody positivity.
**Conclusion**
Lipodystrophy is a significant, yet likely under-reported, long-term complication affecting approximately one-fifth of JDM patients. Its strong association with severe disease features and metabolic comorbidities highlights the need for routine screening as part of standard JDM follow-up. Early and aggressive immunosuppressive therapy may reduce the risk of developing this debilitating complication.

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**Disclosure of interest**
None declared.

## P143

### Regression of severe interstitial lung disease after anti-CD20 therapy in pediatric antisynthetase syndrome

J. Ożga[1,2,*], Z. Żuber[1,2]
[1]Department of Pediatrics, Andrzej Frycz Modrzewski Krakow University; [2]Clinical Department of Pediatrics and Rheumatology, St. Louis Regional Specialised Children's Hospital, Kraków, Poland
**Correspondence:** J. Ożga
*Pediatric Rheumatology 2026*, **24(S1):**P143

**Introduction**
Antisynthetase syndrome is a subtype of inflammatory myopathy characterised by a heterogeneous clinical presentation, including interstitial lung disease (ILD), myositis, arthritis and 'mechanic's hands', associated with the presence of antibodies against tRNA synthetases (ARS).
**Objectives**
To present pediatric patient with antisynthetase syndrome, severe pulmonary involvement, and positive anti-Jo-1 and anti-Ro52 antibodies.
**Methods**
The first symptoms, manifested as red, papular lesions on the skin of the hands, appeared at the age of 8. After several months, the child was admitted to hospital, where the parents reported significant weakness and a marked reduction in exercise tolerance. The physical examination revealed a mechanic's hand and tender, swollen knees and height below the 3rd percentile for age and sex. The CMAS (Childhood Myositis Assessment Scale) was 48/52. Blood tests revealed normal CRP and ESR levels, while muscle enzymes were elevated (CPK 823 U/l, aldolase 19 U/l). Anti-Jo-1 and anti-Ro52 antibodies were present. A whole-body magnetic resonance imaging (MRI) scan revealed signs of inflammation of the chest wall muscles. A high-resolution computed tomography (HRCT) scan of the chest revealed ground-glass opacities and reticular changes consistent with interstitial lung disease, leading to a diagnosis of antisynthetase syndrome with pulmonary involvement. Given the poor prognosis due to severe findings on HRCT, treatment was immediately initiated with pulse glucocorticoids, followed by oral therapy and rituximab, followed by methotrexate as maintenance therapy. Follow-up HRCT scans six months after the initiation of treatment showed regression of the interstitial changes, while in long-term follow-up only minimal residual changes remained, with no signs of active disease. Muscle enzyme levels normalised. Short stature remains under ongoing endocrinological surveillance. During subsequent follow-up, skin lesions and musculoskeletal symptoms resolved completely, accompanied by significant improvement in respiratory function. The favorable clinical and radiologic response suggests high effectiveness of glucocorticoids and anti-CD20 therapy in controlling severe organ involvement. This case highlights the importance of early recognition and prompt initiation of immunosuppressive therapy in pediatric antisynthetase syndrome with pulmonary involvement.

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**Disclosure of interest**
None declared.

## P144

### Treat-to-target outcomes in juvenile dermatomyositis: a single-centre retrospective cohort

P. Šeferna[1,*], P. Doležalová[1], K. Šingelová[1]
[1]Department of Pediatrics and Inherited Metabolic Disorders, General University Hospital in Prague, Prague, Czech Republic
**Correspondence:** P. Šeferna
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**Introduction**
Recent treat-to-target (T2T) recommendations identify inactive disease as the preferred therapeutic target, ideally achieved within 12 months, with discontinuation of glucocorticoids (GC) within 12 months. Real-world data are needed to assess how often these targets are achieved in and to describe treatment burden before transition to adult services.
**Objectives**
To describe presentation, treatment burden, and achievement of T2T outcomes in a single-centre JDM cohort.
**Methods**
Retrospective electronic hospital record review of all patients followed between April 2016 to April 2026.
**Results**
From 35 patients 23 (65.7%) were girls, 27 had classic JDM (77.1%), JDM without myositis and polymyositis were present in 4 patients (11.4%) each. Median age at onset and at diagnosis was 7.2 and 7.8 years, respectively, with the median diagnostic delay of 3 months. Initial manifestations included: fatigue and muscle weakness (88.6%), Gottron's papules (77.1%), nailfold changes (71.4%), malar rash (62.9%), heliotrope erythema (42.9%), and myalgia, gingivitis, arthritis in 40% each. Baseline CMAS was 37/52 (IQR 28–44) and MMT 68/80 (IQR 53–74.5). Myositis-specific antibodies were positive in 6/26 tested patients (23.1%), likely reflecting historical heterogeneity of testing methods and incomplete antibody panels. Muscle MRI was compatible with myositis in 27/35 (77.1%) cases. Typical nailfold capillaroscopy pattern was present in 24/29 assessments (82.8%). All patients received GC, methotrexate, hydroxychloroquine and IVIG were used in 97.1, 68.6 and 54.3%, respectively. Additional immunosuppression included mycophenolate mofetil, JAK inhibitors and cyclosporine A in 28.6, 20.0 and 17.1%, respectively while rituximab and cyclophosphamide were both used in 2 (5.7%) most severe patients. While clinically inactive disease (CID) was achieved in 28/35 patients (80.0%) after a median of 1.7 years from diagnosis, at 12 months it was noticed only in 7/35 patients (20.0%). CID off GC was reached in 25/35 patients (71.4%) after 2.7 years, and CID off all systemic treatment in 13/35 patients (37.1%) after 4.5 years. GC exposure was 31.0 months (IQR 20.5–59.5), with discontinuation within 12 months achieved in 5/35 patients (14.3%). At the last follow-up, 17/35 patients (48.6%) remained under paediatric care, 14/35 (40.0%) had transitioned to adult care, and 4/35 (11.4%) were lost to follow-up.
**Conclusion**
In this cohort most patients achieved CID during paediatric follow-up, but only minority did so within the recommended 12-month T2T timeframe. GC burden has been substantial. More progressive GC dose reduction is warranted with help of potential new treatments. Treatment-free CID was recorded in just over one third of the cohort before the last follow-up. These findings highlight the gap between T2T recommendations and real-world outcomes, while also reflecting limitations of retrospective assessment of early interim T2T milestones.

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**Disclosure of interest**
None declared.

P146

**Severe calcinosis in juvenile dermatomyositis: clinical response to tofacitinib – a case report**

T. R. Vasilev[1,*], K. Temelkova[1], D. Kibarova-Hristova[1], S. Stefanov[1]
[1]Department of Pediatrics, Medical University of Sofia, Sofia, Bulgaria
**Correspondence:** T. R. Vasilev
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**Introduction**
Calcinosis remains one of the most disabling complications in juvenile dermatomyositis (JDM), often refractory to conventional immunosuppressive therapy. Emerging evidence suggests a potential role of Janus kinase (JAK) inhibitors in modulating interferon-driven inflammation and improving cutaneous manifestations.
**Objectives**
To present the clinical course of a pediatric patient with JDM and progressive calcinosis, and to evaluate the therapeutic effect of tofacitinib.
**Methods**
We report a 12-year-old boy with clinically diagnosed JDM (heliotrope rash, Gottron-like lesions, mild proximal muscle weakness), negative myositis-specific antibodies, and normal muscle MRI. Initial treatment included glucocorticoids, methotrexate and hydroxychloroquine. Clinical, laboratory and imaging data were retrospectively reviewed. Disease activity and calcinosis progression were assessed clinically and photographically.
**References: Results** Despite ongoing conventional therapy with glucocorticoids, methotrexate and hydroxychloroquine, the patient demonstrated progressive and extensive calcinosis, with development of large periarticular deposits, most prominently in the region of the first metatarsophalangeal joint, as well as along the dorsum of the foot, accompanied by marked local inflammation, erythema and episodic drainage suggestive of secondary superinfection. Clinically, these lesions were associated with pain, local tenderness and functional impairment. Notably, laboratory parameters remained largely non-inflammatory throughout the disease course, with persistently normal creatine kinase levels and low inflammatory markers, supporting a predominantly cutaneous and vasculopathic disease phenotype rather than active myositis. Given the refractory progression of calcinosis and insufficient response to standard immunosuppressive therapy, treatment with tofacitinib was initiated. Following introduction of the JAK inhibitor, a clear and clinically meaningful improvement was observed, including reduction of local inflammatory changes, gradual softening of calcific deposits and partial regression of lesion size. In addition, the patient experienced a decreased frequency of inflammatory exacerbations and improved functional capacity, with reduced pain and better tolerance to daily activities. No significant adverse events were recorded during the follow-up period, and the therapy was well tolerated.
**Conclusion**
This case highlights the significant therapeutic challenge posed by calcinosis in juvenile dermatomyositis, particularly in patients with low systemic inflammatory activity, where conventional immunosuppressive treatment may be insufficient. It also underscores the potential role of JAK inhibition as a targeted approach addressing interferon-mediated pathogenic pathways. In the presented patient, treatment with tofacitinib was associated with a clinically meaningful improvement of calcinosis, including reduction of inflammation and partial regression of lesions. These findings support the growing body of evidence in the literature that JAK inhibitors may represent a promising therapeutic option in refractory cases of JDM with severe calcinosis, warranting further investigation in larger studies.
**Disclosure of interest**
None declared.

P147

**Liver transplantation in systemic lupus erythematosus and antiphospholipid syndrome complicated by Budd–Chiari syndrome**

R. M. Alcobendas[1], C. Udaondo[1], A. Remesal[1,*], L. Martínez[1], A. HERCBERG[1], S. Barreiro[1], C. Plasencia[1], E. Frauca[1], M. J. Quiles[1]
[1]Pediatric Rheumatology, Hospital La Paz, Madrid, Spain
**Correspondence:** A. Remesal
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**Introduction**
Systemic lupus erythematosus is associated with antiphospholipid antibodies in approximately 30–40% of cases, with a proportion of these patients subsequently developing antiphospholipid syndrome and an increased risk of both arterial and venous thrombosis. Hepatic vein thrombosis secondary to antiphospholipid syndrome may lead to Budd–Chiari syndrome, which can occasionally represent the first manifestation of the underlying autoimmune disease.
**Objectives**
To describe a clinical case of systemic lupus erythematosus and Budd–Chiari syndrome secondary to associated antiphospholipid syndrome who underwent liver transplantation.
**Methods**
Clinical chart review.
**References: Results** A 20-year-old female with a diagnosis of systemic lupus erythematosus and Budd–Chiari syndrome secondary to associated antiphospholipid syndrome was receiving treatment with mycophenolate mofetil, prednisolone, acenocoumarol, intravenous immunoglobulins and rituximab. However, due to chronic and irreversible thrombosis of all three hepatic veins, as well as chronic thrombosis of the portal vein extending to the junction with the right atrium, the patient was listed for orthotopic liver transplantation with ascending portal vein replacement. During surgery, owing to donor–recipient size mismatch, both the pleura and pericardium were affected. Following transplantation, immunosuppressive therapy with tapering corticosteroids, tacrolimus, and mycophenolate mofetil commenced, with progressive improvement in liver function. Nevertheless, the patient developed a persistent right-sided pleural effusion lasting for several weeks despite repeated thoracocenteses, ultimately requiring insertion of a permanent pleural drainage tube. In addition, she developed haemolytic anaemia with progressive deterioration requiring red blood cell transfusions. Upper and lower gastrointestinal endoscopy were performed to exclude variceal bleeding secondary to portal hypertension, while infectious and immunological causes — including suspected disease relapse presenting with serositis and cytopenia — were ruled out. Given the suspicion of a drug-related aetiology, tacrolimus was discontinued and replaced with ciclosporin, resulting in complete resolution of symptoms within a few days. After a hospital stay of two and a half months, the patient was discharged home without further complications.
**Conclusion**
In complex, heavily pre-treated patients, the development of new complications should prompt not only a thorough review of potential medical causes, but also careful consideration of possible drug-related adverse effects. In particular, haemolytic anaemia is a recognised complication in patients receiving tacrolimus.
**Disclosure of interest**
None declared.

P148

**Rare SAT1 variant in early-onset childhood systemic lupus erythematosus: case report and case-control functional validation**

A. A. Alasmari[1,*], T. Mukherjee[2], A. Knight[1], D. Dominguez[3], D. M. Levy[1], D. J. Philpott[4], L. T. Hiraki[1]
[1]Division of Rheumatology, The Hospital for Sick Children (SickKids), University of Toronto; [2]Department of Immunology, Laboratory Medicine and Pathobiology, University of Toronto; [3]Division of Rheumatology, SickKids Research Institute; [4]Department of Immunology, University of Toronto, Toronto, Canada
**Correspondence:** A. A. Alasmari
*Pediatric Rheumatology 2026*, **24(S1):**P148

**Introduction**
The X-linked gene, *SAT1* encodes spermidine/spermine $N^1$-acetyltransferase, a key regulator of polyamine metabolism. Rare *SAT1* variants have been reported in males with young-onset childhood systemic lupus erythematosus (cSLE) in males.
**Objectives**
We describe a male diagnosed with cSLE at age 5 years with a rare, predicted damaging *SAT1* variant and present clinical and functional correlates.

**Methods**
Trio whole-genome sequencing was performed in the proband and both parents, followed by variant annotation and prioritization. Functional assessment used patient-derived fibroblasts compared with two healthy male controls. STING pathway activation was assessed using interferon-stimulatory DNA and the STING agonist diABZI. Signaling responses were evaluated by time-course Western blot and interferon-stimulated gene (ISG) qPCR.
**Results**
The patient presented at age 5 years of age with absence seizures followed by a severe inflammatory episode with Kawasaki-like features complicated by mild coronary artery dilation and macrophage activation syndrome. At 5.5 years of age he developed proteinuria and was found to be hypocomplementemic, with positive ANA, anti-dsDNA, anti-Sm, anti-RNP, and DAT-positive hemolytic anemia. A kidney biopsy class IV lupus nephritis. He was treated with pulse glucocorticoids, hydroxychloquine and mycophenolate. At 10 years of age he developed neuropsychiatric SLE with cognitive and behavioral changes, along with hallucinations. He was treated with cyclophosphamide and increased glucocorticoids with symptom improvement. The patient was born to non-consanguineous, healthy parents of Indo-Guyanese ancestry, without a history of autoimmune disease. Trio sequencing identified a hemizygous X-linked *SAT1* missense variant (c.26C> A; p.Ala9Asp) inherited from his mother, with a high pathogenicity prediction (CADD 31). Functional studies demonstrated reduced *SAT1* protein expression and diminished STING pathway activation following diABZI stimulation compared with controls.
**Conclusion**
We report a novel *SAT1* variant in early-onset cSLE along with functional studies. Reduced *SAT1* expression in the proband and impaired STING signaling suggest disruption of polyamine-mediated innate immune regulation, highlighting variants in the *SAT1* as a potential contributor to immune dysregulation in SLE.
**Disclosure of interest**
None declared.

## P150

**Vaccination practices in jSLE: a survey of pediatric rheumatologists**
A. Dudakli[1,*], B. Menentoğlu[1], P. Prencuva Akyürek[1], Ö. Akgün[1], N. Aktay Ayaz[1]
[1]Department of Pediatric Rheumatology, Istanbul University, Istanbul, Türkiye
**Correspondence:** A. Dudakli
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**Introduction**
Children with juvenile systemic lupus erythematosus (jSLE) are at increased risk of infections because of immune dysregulation and immunosuppressive therapies. Although updated EULAR/PReS recommendations provide guidance for vaccination in pediatric autoimmune inflammatory rheumatic diseases, their implementation in routine practice remains uncertain.
**Objectives**
This study aimed to assess the knowledge, attitudes, and clinical practices of pediatric rheumatologists regarding vaccination in patients with jSLE.
**Methods**
This cross-sectional survey included pediatric rheumatologists and fellows. An anonymous questionnaire based on the 2021 EULAR/PReS recommendations evaluated vaccination knowledge, practices, attitudes, and clinical decision-making.
**Results**
A total of 49 physicians completed the survey: fellows (46.9%), specialists (16.3%), associate professors (32.7%) and professors (4.1%). Most worked at university (61.2%) or training and research hospitals (34.7%); 32.7% followed 26–50 jSLE patients and 14.3% followed ≥50. Knowledge of non-live vaccines was high; all participants correctly identified that these vaccines are generally safe in immunosuppressed jSLE patients and that immunosuppressive treatment should not be delayed for vaccination. In contrast, uncertainty was greater for live vaccines, especially varicella vaccination in selected immunosuppressed patients (49.0% correct).Most participants routinely evaluated vaccination status before immunosuppressive treatment (81.6%) and recommended influenza vaccination (73.5%). Disease activity (95.9%) and immunosuppressive treatment type (98.0%) were commonly considered in live-vaccine decisions, but frequent consideration of varicella vaccination in seronegative patients was limited (46.9%). Familiarity with EULAR/PReS recommendations was higher among physicians with >5 years versus ≤5 years of experience (61.9% vs. 32.1%, $p$=0.048). Correct identification of MMR booster eligibility in selected immunosuppressed patients was also higher among more experienced physicians (85.7% vs. 42.9%, $p$=0.007). A similar pattern was observed for varicella vaccination knowledge without reaching statistical significance (66.7% vs. 35.7%, $p$=0.069).
**Conclusion**
Pediatric rheumatologists demonstrated high awareness of non-live vaccines and general vaccination principles in jSLE. Persistent uncertainty about live vaccines, particularly among less experienced physicians, highlights the need for improved implementation of EULAR/PReS vaccination recommendations.

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**Disclosure of interest**
None declared.

## P151

**Drug-induced lupus erythematosus in children**
E. Turginekova[1], I. Avrusin[1,*], S. Budennaya[1], M. Kostik [1]
[1]Hospital Pediatrics, Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation
**Correspondence:** I. Avrusin
*Pediatric Rheumatology 2026*, **24(S1):**P151

**Introduction**
Drug-induced lupus erythematosus (DILE) is an autoimmune phenomenon that is similar to idiopathic lupus erythematosus and occurs in patients during treatment with certain medications. Given the variability of DILE manifestations, the absence of universal diagnostic criteria, and the limited number of publications on DILE in children, this topic is particularly relevant.
**Objectives**
Retrospective analysis of 9 pediatric DILE cases.
**Methods**
Data from 9 patients (8 girls, 1 boy) aged 10 to 17 years with a confirmed diagnosis of drug-induced lupus, were retrospectively analyzed. The following parameters were assessed: medical history, clinical symptoms, laboratory markers, the timing of symptom onset relative to the start of the triggering drug, and the effectiveness of the DILE therapy administered.
**Results**
The majority of patients had an underlying diagnosis of Juvenile Idiopathic Arthritis (88%, 8/9) with comorbid depression in one of them and the remaining patient had focal epilepsy. The main triggers of DILE were tumor necrosis factor inhibitors (TNFi) – adalimumab, etanercept, infliximab (in 5 patients); other cases were associated with sulfasalazine, oxcarbazepine, lamotrigine, and quetiapine. The median time from drug initiation to symptom onset was 5.23 months (range: 0.72-34.23 months). The most common manifestations were: cutaneous and mucosal lesions (subacute and discoid lupus, alopecia, oral ulcers – 44%, 4/9), Coombs' positive hemolytic anemia (22%, 2/9). Low C3 and C4 complement levels were noted in 22% (2/9) of patients. Anti-dsDNA antibodies were detected in 78% (7/9) of patients, antiphospholipid antibodies in 22% (2/9), anti-nucleosome antibodies in 22% (2/9). In patients with JIA, joint involvement as part of DILE was defined as flare and/or involvement of new joints during the drug treatment period (6/8, 75%). Capillaroscopy was performed in 56% (5/9) of patients. Characteristic lupus changes were observed in 80% (4/5) of

these, presenting as polymorphous, variable-caliber, relatively long, disorganized capillaries with areas of reduced density and perivascular zone edema. Treatment of DILE included withdrawal of the trigger drug in all cases, systemic glucocorticoids in 67% (6/9), hydroxychloroquine in 44% (4/9), rituximab in 22% (2/9), and mycophenolic acid in 11% (1/9). A positive response was achieved in 88% (8/9) of patients. In one patient with uveitis associated with JIA, DILE signs resolved following administration of hydroxychloroquine and rituximab; however, an exacerbation of ocular involvement was noted.

**Conclusion**

DILE in children remains an understudied problem, underscoring the need to increase awareness among physicians of all specialties. The development of DILE requires timely diagnosis and drug withdrawal. Capillaroscopy might be used in all cases JIA, there DILE highly suspected.

**Disclosure of interest**

None declared.

## P152

### Use of obinutuzumab in an adolescent with refractory proliferative and membranous lupus nephritis and a history of rituximab-induced anaphylaxis: a case report

C. Udaondo[1,*], N. C. Fuertes[2], R. Alcobendas Rueda[1], G. Moral Garcia[3], A. Hercberg Moreno[1], A. Remesal Camba[1], L. Martinez Marcelo[1], S. Barreiro Perez[1], E. Fernandez Pulido[1]

[1]Pediatric Rheumatology, Hospital La Paz, Madrid; [2]Pediatric, Hospital Puerta del Mar, Cadiz; [3]Rheumatology, Hospital Universitario Virgen Macarena, sevilla, Spain

**Correspondence:** C. Udaondo

*Pediatric Rheumatology 2026*, **24(S1):**P152

**Introduction**

Lupus nephritis is one of the most severe manifestations of pediatric systemic lupus erythematosus and is associated with high morbidity and mortality. In patients with refractory disease or inadequate response to conventional immunosuppressive therapy, B-cell targeted therapies represent a relevant therapeutic alternative (1). However, severe hypersensitivity reactions to rituximab may limit available treatment options. Obinutuzumab, a type II humanized anti-CD20 monoclonal antibody, has shown promising results in refractory lupus nephritis, although experience in the pediatric population remains limited (2).

**Objectives**

To describe the clinical course and tolerability of obinutuzumab in an adolescent with refractory class III + V lupus nephritis and a history of rituximab-induced anaphylaxis.

**Methods**

A 16-year-old male with class III + V lupus nephritis in the setting of pediatric systemic lupus erythematosus was receiving combined immunosuppressive therapy with tacrolimus, mycophenolate mofetil, hydroxychloroquine, and prednisone together with belimumab, with persistent clinical activity and poor disease control. As a relevant medical history, he had experienced an episode of rituximab-induced anaphylaxis six years earlier. Due to clinical refractoriness, belimumab was switched to obinutuzumab (3). The patient showed good treatment tolerance, without hypersensitivity reactions or immediate adverse events, along with subsequent clinical improvement following the therapeutic change.

**Conclusion**

Obinutuzumab may represent an effective and safe therapeutic alternative in pediatric patients with refractory lupus nephritis and a history of severe hypersensitivity to rituximab. This case highlights the potential benefit of humanized anti-CD20 blockade in complex clinical scenarios where therapeutic options are limited. Further studies in pediatric populations are needed to better define its long-term efficacy and safety profile.

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**Disclosure of interest**

None declared.

## P153

### Proteomic profiling identifies novel biomarkers for diagnosis and disease activity assessment in juvenile systemic lupus erythematosus

C. Simu[1,*], M. Badii[2,3], I. Szabo[1,4], O. Gaal[2,3], C. Pamfil[1,4], M. M. Tamas[1,4], L. Damian[1], M. Sparchez[5,6], T. O. Crisan[2,3], L. Joosten[2,3], S. Rednic[1,4]

[1]Rheumatology, Emergency County Hospital; [2]Medical Genetics, "Iuliu Hatieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania; [3]Internal Medicine, Radboud Institute for Molecular Life Sciences, Nijmegen, Netherlands; [4]Rheumatology, "Iuliu Hatieganu" University of Medicine and Pharmacy; [5]2nd Pediatric Clinic, Emergency Clinical Hospital for Children; [6]Mother and Child, "Iuliu Hatieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania

**Correspondence:** C. Simu

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**Introduction**

Juvenile-onset systemic lupus erythematosus (jSLE) is characterized by greater disease activity, more severe organ damage, and increased treatment requirements compared to adult-onset disease. Despite numerous autoantibodies associated with SLE, age-specific biomarkers for disease activity monitoring in jSLE remain limited.

**Objectives**

To identify novel protein biomarkers associated with disease activity in jSLE using proximity extension immunoassay technology.

**Methods**

Serum samples from 19 jSLE patients and 9 age-matched healthy controls were analyzed using proximity extension immunoassay (Olink) to quantify 92 inflammation-related proteins. Disease activity was assessed using the SLE Disease Activity Index (SLEDAI). Associations between differentially expressed proteins and serum vitamin D levels were evaluated to determine potential molecular contributors to disease course.

**Results**

Proteomic profiling identified two distinct patient clusters with low-inflammatory and high-inflammatory signatures. Eight proteins were upregulated in jSLE patients, with CXCL6 and CST5 showing statistically significant elevation compared to healthy controls (p0.05). Six additional proteins—TNF, HGF, FGF-5, CD244, MMP-10, and TNFRSF9—demonstrated upregulation without reaching statistical significance. Differentially expressed proteins included interferon-inducible chemokines, B cell receptor signaling molecules, and cytokines involved in leukocyte, neutrophil, and macrophage trafficking. jSLE patients exhibited significantly elevated disease activity with prominent renal and hematologic manifestations at diagnosis and during flares, though no specific protein profile correlated with these clinical features. Pearson correlation analysis revealed a significant positive association between CX3CL1 and serum vitamin D levels in the jSLE cohort.

**Conclusion**

This proteomic analysis identified novel candidate biomarkers reflecting distinct molecular patterns in jSLE patients with active disease. The observed proteomic signatures, particularly the high-inflammatory cluster, support the need for age-specific diagnostic and therapeutic strategies in juvenile-onset lupus.

**Disclosure of interest**

None declared.

## P155

### The effect of diagnostic criteria, gender, and genetics on low disease activity and remission in patients with juvenile systemic lupus erythematosus

D. Alkan[1,*], E. Arslanoglu Aydin[1]
[1]Paediatric Rheumatology, Etlik City Hospital, Ankara, Türkiye
**Correspondence:** D. Alkan
*Pediatric Rheumatology 2026*, **24(S1):**P155

**Introduction**
Juvenile-onset Systemic Lupus Erythematosus (jLSE) is a chronic, systemic autoimmune and inflammatory disease. Unlike adult-onset SLE, children and adolescents with jSLE typically experience higher levels of disease activity, a higher drug burden, and more severe internal organ involvement. SLE is generally a polygenic disease, but an increasing number of functional rare monogenic variants have been identified through whole-exome sequencing and whole-genome sequencing studies.
**Objectives**
In this study we aimed to determine the effect of demographic, clinical and laboratory findings alongside diagnostic tools on low disease activity and/or remission criterias of children with jSLE.
**Methods**
In this study, 26 patients followed up with a diagnosis of jSLE at the pediatric rheumatology clinic of Ankara Etlik City Hospital between October 2022 and February 2026 were retrospectively evaluated. For jSLE diagnosis ACR1997, SLICC 2012 and ACR/EULAR 2019 criteria were used; for disease activity SLEDAI-2K and for low disease activity and remission, cLLDAS and DORIS criterias were used. The time to cLLDAS was abbreviated as tLLDAS, the time to DORIS as tDORIS, the percentage of the follow-up period that passed cLLDAS as rLLDAS, and the percentage that passed DORIS as rDORIS. If rLLDAS or rDORIS >50%, the abbreviations r50LLDAS or r50DORIS were used.
**Results**
Our cohort consisted 22 girls (84.6%) and 4 boys (15.4%). The median age of onset of symptoms was 144.5 (IQR:98.75-160.25) months. The median age at diagnosis was 150.5 (IQR:113-160.25) months. All patients met the ACR/EULAR 2019 diagnostic criteria, while 92.3% met the ACR 1997 criteria and 76.9% met the SLICC 2012 criteria. Genetic analysis for monogenic lupus was sent in 11 (42.3%) patients, and at least one significant gene mutation was detected in 5 (45.5%) (C1Q in two patients, DNASE1L3, UNC93B1, and C7 in one patient each). DORIS was observed in 75% of boys and 22.7% of girls, but this difference was statistically insignificant ($p$=0.925). In contrast, r50DORIS was 50% in boys and 4.5% in girls. The difference between them was statistically significant after applying binary logistic regression, rDORIS50 (B = 3.045, Wald = 4.527, $p$ = 0.033). Patients who had hypocomplementemia and who did not receive IVIG in their treatment had higher rDORIS ($p$=0.028).
**Conclusion**
Our study showed no difference in remission, low disease activity, and time to inclusion in these classifications among patients diagnosed with monogenic lupus. The similar gender distribution among patients diagnosed with monogenic lupus, unlike other SLE patients, is consistent with the nature of the disease. Significant genetic variants for monogenic lupus were observed more frequently, particularly in patients with early onset, a history of consanguineous marriage, and recurrent infections. Although low disease activity was achieved in most patients, remission rates remained limited, with a higher rate of remissional state observed in whom hypocomplementemia at diagnosis, not receiving IVIG and males. Our findings support the importance of early genetic assessment and long-term disease control in jSLE.

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**Disclosure of interest**
None declared.

## P156

### Lupus nephritis presenting as acute pyelonephritis and renal vein thrombosis in an adolescent girl

O. Altug Gucenmez[1,*], B. E. Kosali[1]
[1]Department of Pediatric Rheumatology, University of Health Sciences, Dr. Behçet Uz Pediatric Diseases and Surgery Training and Research Hospital, Izmir, Türkiye
**Correspondence:** O. Altug Gucenmez
*Pediatric Rheumatology 2026*, **24(S1):**P156

**Introduction**
Systemic lupus erythematosus (SLE) is a multisystem autoimmune disease with a broad clinical spectrum and potentially severe organ involvement (1). Renal involvement is one of the major determinants of morbidity and long-term outcome in pediatric patients. Because fever, abdominal pain, urinary abnormalities, and elevated inflammatory markers may overlap with infectious diseases, SLE can occasionally mimic common pediatric infections at initial presentation. Thrombotic complications, although less frequent, may further obscure the diagnosis and delay appropriate immunosuppressive treatment (2).
**Objectives**
We aimed to present an adolescent girl who was initially managed with a presumptive diagnosis of acute pyelonephritis, subsequently developed renal vein thrombosis, and was ultimately diagnosed with lupus nephritis. Through this case, we sought to emphasize the importance of considering underlying autoimmune diseases in atypical or treatment-resistant renal infections, particularly when thrombotic and hematological abnormalities are present.
**Methods**
We retrospectively reviewed the clinical, laboratory, imaging, and histopathological findings of a 16-year-old female patient. She presented with right lower quadrant pain. Initial evaluation revealed pyuria, microscopic hematuria, and increased renal parenchymal echogenicity, leading to hospitalization with a presumptive diagnosis of acute pyelonephritis. Contrast-enhanced computed tomography showed a non-enhancing area in the right kidney, suggesting severe pyelonephritis with possible parenchymal injury or necrosis. Doppler ultrasonography raised suspicion of renal vein thrombosis, and anticoagulation was initiated. Extended-spectrum beta-lactamase–producing Escherichia coli was isolated, and antibiotic therapy was escalated. However, despite appropriate antimicrobial treatment, clinical deterioration and the development of hemolytic anemia prompted further investigation. Immunological work-up revealed ANA, anti-dsDNA, anti-Sm, and anti-nucleosome antibody positivity with hypocomplementemia, consistent with active SLE. Renal biopsy demonstrated class V + II lupus nephritis, with a modified NIH activity index of 1/24 and chronicity index of 1/12. Immunofluorescence showed a characteristic "full-house" pattern. This case highlights that childhood-onset SLE may initially mimic infection and may be complicated by thrombotic events such as renal vein thrombosis. In atypical or treatment-resistant infections, particularly when accompanied by thrombotic or hematological abnormalities, an underlying autoimmune disease should be considered early.

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**Disclosure of interest**
None declared.

## P157

### Early aortic stiffness and ventricular–vascular coupling in children with connective tissue diseases: insights from LEAP

S. Govindarajan[1,*], C. Ciculete[2], S. Dyball[3], B. Parker[2,3], I. N. Bruce[3], A. Chieng[3]
[1]Paediatric Rheumatology, Royal Manchester Children's Hospital; [2]Kellgren Centre for Rheumatology; [3]University of Manchester, Manchester, UK
**Correspondence:** S. Govindarajan
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**Introduction**
Patients with connective tissue diseases (CTD) are at increased risk of cardiovascular involvement in adulthood 1, but subclinical alterations in ventricular–vascular coupling remain poorly defined in paediatric population.
**Objectives**
To assess aortic stiffness and ventricular–vascular coupling in pediatric CTD compared with age- and sex-matched controls.
**Methods**
Thirty-three CTD patients (12-16 years), (27 juvenile systemic lupus erythematosus [JSLE]; others systemic sclerosis, UCTD/MCTD, Sjögren's) and five matched healthy controls were evaluated. Aortic pulse wave velocity (PWV, gold standard for arterial stiffness), augmentation index (AIx) reflecting the impact of peripheral pressure waves on the central aorta and left ventricular ejection time (LVET) were measured non-invasively (TensioMed). Data are mean ± SD. Associations were assessed using Spearman correlation and linear regression. The paediatric lupus arm of the LEAP study had prior ethical approval.
**Results**
CTD patients (87.8% female; ethnicity White/Asian/Other: 17/11/5; 64% on steroids) showed higher PWV and AIx than controls (PWV 6.7±1.1 vs. 6.2±0.65 m/s; AIx 9.8±9.4% vs. 4.5±5%); PWV correlated with AIx (ρ=0.476, $p$=0.01). Higher PWV was independently associated with shorter LVET (B=−20.3 ms per 1 m/s, $p$<0.001), revealing a **novel arterial stiffness–ventricular timing link** . No associations were observed with age, blood pressure, disease duration, ethnicity, CTD subtype, or steroid exposure.
Subgroup analyses showed heterogeneous stiffness: neuropsychiatric SLE had the highest PWV/AIx and shortest LVET; renal involvement was intermediate; mucocutaneous and hematological subgroups had near-control PWV with elevated AIx; other CTD including systemic sclerosis also had elevated PWV.
**Conclusion**
Pediatric CTD demonstrates higher aortic stiffness and altered ventricular–vascular coupling. The PWV–LVET inverse association is novel, reflecting earlier systolic load due to arterial stiffening. These findings support early cardiovascular monitoring and risk stratification to identify children at risk of developing overt cardiac dysfunction in adulthood, allowing timely preventive strategies or interventions.

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**Disclosure of interest**
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## P158

### Disease activity trajectories in childhood-onset SLE reveal distinct patterns of treat-to-target achievement

G. Olivieri[1,*], V. Messia[1], E. Marasco[1], C. Bracaglia[1], F. De Benedetti[1]
[1]Division of Rheumatology, ERN-RITA Centre, IRCCS Bambino Gesù Children's Hospital, Rome, Italy
**Correspondence:** G. Olivieri
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**Introduction**
Childhood-onset systemic lupus erythematosus (cSLE) is an autoimmune disease with heterogeneous clinical course. However, disease trajectories remain incompletely characterized.
**Objectives**
To define longitudinal disease activity trajectories in cSLE patients and assess their relationship with damage accrual and time to Treat-to-Target (T2T) achievement.
**Methods**
This combined retrospective and prospective analysis analyzed a cohort of cSLE patients followed in a single tertiary paediatric rheumatology centre in Italy. Data were collected at baseline and at each follow-up visit (with at least three follow-up visits per year). Disease activity and organ damage were assessed using the SLEDAI-2k and SDI, respectively. We evaluated disease activity trajectories over time using latent class growth analysis (LCGA) with the lcmm package in R.
**Results**
The cohort ($n$=40; 75% female; 88% Caucasian), had a median baseline age of 13.5 years (IQR 12.2 - 15.4). Median time from disease onset to the first specialist visit was 0.2 (IQR 0.1 - 0.4) years. Patients were followed for a median of 3.71 years (IQR 2 - 6.44), reaching a median age of 18.4 years (IQR: 16.1–20.7) at the last visit. At the final visit, 83% achieved Lupus Low Disease Activity State (LLDAS) 78% reached clinical remission (cCR), and 35% achieved glucocorticoid-free remission (cCR-0). Notably, 43% of the patients ($n$ = 17) had already accumulated significant organ damage, and the mean SDI score at the last visit was 0.80 ±1.26. LCGA identified two distinct patterns: Class 1 (*High-Baseline/Fast Responders*), characterized by high initial SLEDAI-2K scores followed by rapid clinical improvement, and Class 2 (*Persistent Low Activity*), with stable low disease activity from onset. Class 1, at univariate analysis, was associated with higher baseline neuropsychiatric (NP) (psychosis, $p$=0.011; cognitive impairment, $p$=0.011) and renal manifestations (proteinuria, $p$=0.009; hematuria, $p$=0.006). In the multivariate analysis, NP involvement emerged as the strongest independent predictor of Class 1 trajectory (OR: 0.04; $p$=0.033).Notably, Class 2 showed a significantly higher prevalence of the Serological Active Clinically Quiescent (SACQ) phenotype compared to Class 1 ($p$=0.0019). Although similar proportions achieved clinical targets by month 24, achievement kinetics differed significantly. Class 1 reached LLDAS earlier than Class 2 (7.0 vs. 16.0 months; $p$=0.036). The time to achieve cCR favored Class 1 ($p$ = 0.044). No significant difference in final SDI scores was observed between the two classes.
**Conclusion**
Severe baseline manifestations in cSLE do not always predict worse outcomes. 'Fast Responders' overcome high disease activity by achieving remission and LLDAS earlier, supporting a 'window of opportunity' for prompt intervention. In contrast, 'Slow Responders' experience ~9-month delays in T2T achievement due to persistent SACQ.
**Disclosure of interest**
G. Olivieri: None declared, V. Messia: None declared, E. Marasco: None declared, C. Bracaglia Consultant with: SOBI, Novartis, F. De Benedetti Consultant with: Abbvie, SOBI, Novimmune, Novartis, Roche, Pfizer.

## P160

### Paediatric antiphospholipid syndrome: experience from a tertiary referral center

D. Kiafzezi[1,*], V. Sgouropoulou[1], M. Economou[2], D. Zafeiriou[3], M. Trachana[1]
[1]Paediatric Immunology and Rheumatology Referral Centre, 1st Paediatric Department, Aristotle University of Thessaloniki, Hippokrateio General Hospital; [2]Paediatric Haematology Referral Centre, 1st Paediatric Department, Aristotle University of Thessaloniki, Hippokrateio General Hospital; [3]Paediatric Neurology Referral Centre, 1st Paediatric Department, Aristotle University of Thessaloniki, Hippokrateio General Hospital, Thessaloniki, Greece
**Correspondence:** D. Kiafzezi
*Pediatric Rheumatology 2026*, **24(S1):**P160

**Introduction**
Pediatric antiphospholipid syndrome (APS) is a rare autoimmune disorder characterized by thrombotic events, associated with persistent antiphospholipid antibodies [lupus anticoagulant (LA), anticardiolipin antibodies (aCL) IgG and/or IgM and/or anti-β2 glycoprotein-I antibod-

ies (anti-β2GPI) IgG and/or IgM]. The clinical expression differs from adults and remains an under-recognized condition in children.

**Objectives**

To present the clinical, laboratory and treatment characteristics of pediatric APS patients treated at a tertiary referral center and highlight diagnostic and therapeutic challenges.

**Methods**

A retrospective study was conducted including patients aged <16 years diagnosed with APS from 2020 to 2026 according to the 2023 ACR/EULAR classification criteria requiring clinical thrombosis and persistent antiphospholipid antibody positivity on ≥2 occasions ≥12 weeks apart. Clinical, laboratory, imaging, treatment and follow-up data were extracted from medical records. Descriptive statistical analysis was performed.

**Results**

Seven patients were included, 5 with primary and 2 with secondary APS associated with Systemic Lupus Erythematosus (SLE) and Primary Angiitis of Central Nervous System. Median age at diagnosis was 12.5 years and 57.1% were female. Arterial thrombosis was the predominant manifestation (85.7%), most frequently involving cerebral circulation (4/7), followed by pulmonary (2/7) and renal arteries (1/7). Venous thrombosis presenting as superior sagittal sinus thrombosis occurred in a patient with stroke. The patient with SLE-associated secondary APS experienced recurrent thromboses. Double positivity for aCL and anti-β2 GPI was present in all the patients presenting with low titers, while LA in 28.5%. Genetic thrombophilia screening was negative in all cases. All patients received anticoagulation therapy with low molecular weight heparin (57.1%), aspirin (42.9%), or combination therapy (14.3%). Corticosteroids were administered in 71.4%. During a median follow-up of 24 months, two patients remained on low-dose aspirin and no major treatment-related complications were observed. No cases of catastrophic APS or mortality were observed.

**Conclusion**

Pediatric APS poses substantial diagnostic challenges owing to the heterogeneity of clinical manifestations and antibody profiles. Early recognition and sustained multidisciplinary management are essential to reducing morbidity. These findings highlight the need for pediatric-specific classification criteria and prospective multicenter studies to optimize long-term outcomes.

**Disclosure of interest**

None declared.

## P161

### Rituximab as a steroid-sparing strategy in pediatric systemic lupus erythematosus: real-world evidence

M. Kapranova[*,1], S. Kurbanova[1], S. Valieva[1], E. Sholobova[1]
[1]Department of Rheumatology, Morozov Children's City Clinical Hospital, Moscow, Russian Federation
**Correspondence:** M. Kapranova
*Pediatric Rheumatology 2026*, **24(S1):**P161

**Introduction**

Childhood-onset systemic lupus erythematosus (SLE) is associated with high disease activity and substantial glucocorticoid (GC)-related toxicity. Evidence on predictors of response to rituximab in pediatric SLE remains limited.

**Objectives**

This study evaluated the effectiveness and safety of rituximab, with analysis of clinical response and steroid-sparing effects.

**Methods**

A retrospective single-center study included 21 pediatric patients with SLE (median age 14.0 [10.0–15.0] years, 81% female) who received at least one rituximab infusion between 2016 and 2025. Disease activity (SLEDAI), organ-specific response, GC dose, laboratory parameters, and adverse events were assessed at 6 and 12 months.

**Results**

Rituximab therapy led to a rapid and sustained reduction in SLEDAI from a median of 16.5 (IQR 11.0–18.8) at baseline to 2.0 (IQR 2.0–4.0) at 6 months and 2.0 (IQR 0.5–2.0) at 12 months ($p$<0.001). Low disease activity (SLEDAI ≤5) was achieved in 95% of patients at 12 months. The median daily prednisolone dose decreased from 40.0 mg (IQR 31.3–50.0) to 15.0 mg (IQR 10.0–20.0) ($p$=0.002), and the weight-adjusted dose fell from 1.00 mg/kg/day (IQR 1.00–1.00) to 0.26 mg/kg/day (IQR 0.21–0.32) ($p$<0.001). An organ-specific response was documented in 85.7% of patients: complete resolution of hematological and mucocutaneous manifestations was observed in all evaluable patients, and renal remission was achieved in 78.6% of those with lupus nephritis. Patients with hematological involvement showed rapid normalization of blood counts by 6 months. Immunoglobulin levels (IgG, IgM, IgA) decreased significantly ($p$<0.001), consistent with effective B-cell depletion. Adverse events occurred in 28.6% of patients, most commonly hypogammaglobulinemia requiring intravenous immunoglobulin. One fatal outcome (4.8%) was recorded in a patient with refractory, multiorgan disease.

**Conclusion**

Rituximab demonstrated robust clinical efficacy and a clinically meaningful steroid-sparing effect in pediatric SLE. These findings support rituximab as a viable therapeutic strategy in severe pediatric disease, while underscoring the need for careful monitoring of immunosuppression-related complications.

**Disclosure of interest**

None declared.

## P162

### Is there a difference in clinical phenotype between U1-RNP positive and negative juvenile systemic lupus erythematosus: results from a national cohort study

L. Gatti[1,*], G. Simonini[1,2], M. W. Beresford[3,4], C. Pain[3,4] on behalf of UK JSLE Study Group
[1]Rheumatology Unit, ERN-ReCONNET center, Meyer Children's Hospital IRCCS; [2]NEUROFARBA Department, University of Florence, Florence, Italy; [3]Department of Paediatric Rheumatology, Institute in the Park, Alder Hey Children's NHS Foundation Trust; [4]Institute of Life Course and Medical Sciences, University of Liverpool, Liverpool, UK
**Correspondence:** L. Gatti
*Pediatric Rheumatology 2026*, **24(S1):**P162

**Introduction**

Paediatric mixed connective tissue disease (MCTD) is a rare rheumatic disease characterized by overlapping features with other rheumatologic diseases and the presence of high-titre anti-U1-RNP antibodies. MCTD can also be present in other autoimmune diseases such as Systemic Lupus Erythematosus (SLE) (1,2).

**Objectives**

The aim of the study was to analyse the clinical presentation and outcomes of patients fulfilling the classification criteria for SLE who were positive for anti-U1-RNP antibodies and whether the various classification criteria for MCTD could be applied to these patients.

**Methods**

A multi-centre observational study, the UK JSLE Cohort Study, collects data between 1995 and 2020 from patients <18 years with a diagnosis of SLE fulfilling ≥4 criteria of the American College of Rheumatology (ACR) with ANA and ENA performed. Patients were divided in anti-u1-RNP positive and negative at baseline. Three MCTD classification were applied to determine which patients could fulfil them.

**Results**

We included 341 individuals (275 female, 80.1%) of which 124 patients (36.4%) were anti-u1-RNP positive. The majority were White-British/Caucasian (46%). Raynaud's phenomenon, sclerodactyly and myositis were significantly higher in the anti-u1-RNP cohort ($p$<0.05), while other clinical and laboratory features showed no statistical difference. Amongst the anti-u1-RNP positive cohort, no patient fulfilled the Sharp criteria, eight patients (6.5%) fulfilled the Alarçon-Segovia criteria, and eleven patients (8.9%) fulfilled the Kasukawa criteria. Globally, just 15 patients (12.1%) fulfilled one or more criteria. Amongst them, 12 patients (80%) presented with Raynaud's phenomenon, seven (46.7%) presented with swollen fingers, and ten (66.7%) and five (33.3%) patients presented with myalgia and myositis respectively. All fifteen of these patients (100%) presented with arthritis, while just one patient (6.7%) had renal involvement.

**Conclusion**

This study highlights the paucity of available about MCTD patients in paediatrics. In addition, it notes how the presence of anti-u1-RNP antibodies may potentially be linked to a clinical phenotype with spe-

cific manifestations such as Raynaud's phenomenon, sclerodactyly and muscular involvement.

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**Disclosure of interest**
None declared.

## P163

### Targeting IFNg-dependent IDO1 transcription to disrupt T-bet–IDO1-driven B-cell autoantibody production in pediatric systemic lupus erythematosus

M. Caforio[1,*], E. Piano Mortari[1], C. Bracaglia[1], F. De Benedetti[1], E. Marasco[1], V. Folgiero[1]
[1]Bambino Gesù Children's Hospital, Rome, Italy
**Correspondence:** M. Caforio
*Pediatric Rheumatology 2026*, **24(S1):**P163

**Introduction**
Childhood-onset systemic lupus erythematosus (cSLE) is a complex multiorgan systemic, inflammatory, and autoimmune chronic condition. The course of disease is variable, frequently associated with periods of flares and remission. The diagnosis typically occurs before the age of 18 with higher prevalence of nephritis, diffuse alveolar hemorrhage, neuropsychiatric and hematological involvements (1). The pathogenesis of cSLE is complex and not fully understood. It involves 3 key factors: genetic risk factors, epigenetic mechanisms, and environmental triggers. The precise causes of cSLE are complex and not yet fully understood but genetic factors play a role in the development of the disease including a pivotal role for IFNg in B cells dysregulation (2).
**Objectives**
Aim of this study is to find new molecular protagonists of cSLE B-cells autoantibody production in order to improve current treatment.
**Methods**
Peripheral blood derived from patients or healthy donors have been submitted to ficol to obtain mononuclear cells. To perform flow cytometry, cells have been marked for membrane markers and fixed and permed for intracellular staining. Supernatants were collected to perform Elisa assay. Primary cSLE cell lines were generated by Epstain Barr virus transduction.
**Results**
Among the genes regulated by IFNg we identified IDO1, an enzyme with a consolidated role in immunotolerance, significantly upregulated in the B-cell compartment of the blood of cSLE patients (3). IDO1 enzymatic inhibitor did not provide beneficial results directly reducing antibody production, leading to speculation that IDO1 could instead regulate the differentiation of autoreactive B cells into antibody-secreting plasma cells. Our data confirm high IDO-1 expression in B-cells and in pathogenic double-negative B-cells subpopulation overexpressing Tbet obtained by peripheral blood of cSLE patients. Additionally we observed that the inhibition of IDO1 protein expression by PROTAC instead of enzymatic inhibitors, resulted in a major reduction of Tbet expression. Furthermore, stimulation with CpG, INFg and anti-Ig revealed an increase in the number of plasmablasts when compared with not stimulated B cells, strongly reduced by the addition of IDO1 protein inhibitor. Upon 6 days of stimulation, differentiation into plasmablasts results in increase of autoantibodies release that are reduced when IDO1 inhibitor is added.
**Conclusion**
Further experiments will be necessary to understand Tbet-IDO1 interaction and to develop an experimental approach able to inhibit IFNg-dependent IDO1 activation by the delivery of a CRISPR/CAS9 system to sustain a potential use of it in the future in the cSLE patients.

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**Disclosure of interest**
None declared.

## P166

### Clinical phenotypes and damage accrual in childhood-onset lupus defined by organ involvement patterns

N. Almajed[1,*], R. Alhuthil[1], A. Alasmari[1], A. Alsaleem[1], A. Alsonbul[1], S. Alfraih[2], S. M. M. Al-Mayouf[1]
[1]Pediatric Rheumatology; [2]Rheumatology, King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia
**Correspondence:** N. Almajed
*Pediatric Rheumatology 2026*, **24(S1):**P166

**Introduction**
Childhood-onset systemic lupus erythematosus (cSLE) is associated with greater disease severity and cumulative organ damage than adult-onset SLE. Despite improved survival, irreversible damage remains a major determinant of long-term outcomes.
**Objectives**
To identify clinical phenotypes in cSLE based on organ involvement patterns and evaluate their associations with cumulative organ damage, sex, and age at disease onset.
**Methods**
We conducted a retrospective cohort study of patients with cSLE fulfilling the 2019 EULAR/ACR classification criteria at the Childhood Lupus Clinic, King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia (1997–2025). Clinical, laboratory, and treatment data were extracted from medical records. Damage accrual at last follow-up was assessed using the Paediatric SLICC/ACR Damage Index (pSDI). Descriptive statistics were used to characterize organ involvement and damage patterns, while univariate and multivariate analyses assessed associations with clinical variables.
**Results**
A total of 187 patients (152 female) were included. Median current age was 22 years (IQR: 15–28), while median age at disease onset and diagnosis was 10 years (IQR: 6–12 and 8–12, respectively). Median disease duration was 13 years (IQR: 5–17). Constitutional manifestations occurred in 89.8% of patients; mucocutaneous and musculoskeletal involvement each in 80.2%; renal involvement in 48.1%; and central nervous system (CNS) involvement in 38%. Among immunosuppressive therapies, mycophenolate mofetil was used in 71.1% of patients, rituximab in 39%, cyclophosphamide in 32.1%, belimumab in 19.3%) tacrolimus in 10.2%, and methotrexate in 4.3%. Median pSDI score was 1 (IQR: 0–3). Renal, musculoskeletal, neuropsychiatric, and ocular domains were the most frequently affected. Twelve deaths were recorded, predominantly infection-related. On multivariable analysis, lupus nephritis was the strongest independent predictor of higher pSDI ($p<0.001$). CNS involvement ($p=0.021$), mucocutaneous involvement ($p=0.048$), and anti-RNP positivity ($p=0.039$) were also independently associated with greater damage accrual. Sex and age at disease onset were not significantly associated with cumulative damage.
**Conclusion**
This cohort demonstrates substantial long-term organ damage and considerable mortality in cSLE. Distinct organ involvement patterns were associated with increased cumulative damage, supporting early risk stratification, routine pSDI monitoring, and treat-to-target strategies.
**Disclosure of interest**
None declared.

## P167

### Macrophage activation syndrome in pediatric patients with systemic lupus erythematosus: a monocentric retrospective observational study

E. Rizzo[1,2,*], C. Provini[1], A. G. Lanuto[1], M. Rossano[1], F. Di Stasio[1], G. Beretta[1], F. Minoia[1], G. Filocamo [1], F. Vianello[1]
[1]Pediatric Immunology and Rheumatology Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan; [2]Pediatrics and Neonatology, Ospedale Cardinal Massaia, Asti, Italy
**Correspondence:** E. Rizzo
*Pediatric Rheumatology 2026*, **24(S1):**P167

**Introduction**
Macrophage Activation Syndrome (MAS) is a rare but potentially life-threatening hyperinflammatory complication of pediatric Systemic Lupus Erythematosus (pSLE). Clinical overlap with lupus flares hinders early diagnosis. Currently available diagnostic criteria -Ravelli, Parodi, and Borgia- require further validation in pSLE cohorts[1-3].
**Objectives**
Our study aimed to describe the clinical and laboratory features of MAS in our cohort of pSLE patients and to identify any differences in clinical or laboratory presentation at SLE onset between patients who subsequently developed MAS and those who did not. A secondary objective was to evaluate the sensitivity and specificity of MAS diagnostic criteria in our cohort.
**Methods**
We conducted a monocentric retrospective observational study including all pediatric patients (age of SLE onset < 18) discharged between 2010 and 2025 with a diagnosis of SLE (ACR or EULAR/ACR 2019 criteria) from the Pediatric Immunology and Rheumatology Unit at Ospedale Maggiore Policlinico, Milan. Clinical, laboratory, and therapeutic data were collected at onset and during follow-up. MAS and non-MAS subgroups were compared using Wilcoxon, Fisher's exact, or Chi-square tests (p-value < 0.05). Sensitivity and specificity of Ravelli, Parodi, and Borgia criteria were calculated in our MAS population.
**Results**
Nine out of fifty-five patients (16%) developed MAS, largely near SLE diagnosis (median: 0.6 months). At SLE onset, MAS patients showed significantly higher serum ferritin (531 vs. 178 ng/mL), higher rates of serositis (37% vs. 9%), and hematuria (88% vs. 51%). MAS was associated with prolonged hospitalization (40 vs. 15 days) and increased pediatric ICU admissions (33% vs. 0%).The Ravelli and Parodi criteria both demonstrated a sensitivity of 88.9%, while the Borgia criteria showed markedly lower sensitivity (11.1%); all three sets achieved high specificity.
**Conclusion**
MAS in pSLE tends to develop early in the disease course and is associated with higher inflammatory burden at onset, particularly elevated ferritin, serositis, and hematuria. Among existing diagnostic criteria, Ravelli and Parodi remain the most clinically reliable tools. Multicenter studies with larger cohorts are needed to refine diagnostic criteria and validate these findings.

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**Disclosure of interest**
None declared.

## P168

### PROS (PIK3CA-related overgrowth spectrum) syndrome in a girl with systemic lupus erythematosus/overlap disease: a diagnostic challenge for rheumatologist

S. Arsenyeva[1,*], I. Nikishina[1], V. Matkava[1], G. Sagoyan[2], A. Firsa[1]
[1]Paediatric, V.A. Nasonova Scientific Research Institute of Rheumatology; [2]Paediatric, Russian Cancer Research Center NN Blokhin, Moscow, Russian Federation
**Correspondence:** S. Arsenyeva
*Pediatric Rheumatology 2026*, **24(S1):**P168

**Introduction**
PIK3CA-related overgrowth spectrum (PROS) is a heterogeneous group of rare mosaic disorders caused by activating somatic variants in the *PIK3CA* gene. Clinical manifestations include focal or asymmetric overgrowth of soft tissues and bones, vascular and lymphatic malformations and variable skeletal abnormalities.
**Objectives**
To describe a rare clinical case of genetically confirmed PROS in a girl with systemic lupus erythematosus (SLE)/overlap syndrome.
**Methods**
Case report. A 12-year-old girl was admitted to our paediatric rheumatology department in August 2025 because of persistent fever, Raynaud phenomenon, polyarthritis and elevated acute-phase reactants. Since birth, she had extensive cutaneous vascular malformations, lymphoedema/asymmetric left-sided overgrowth and congenital dactylitis-like abnormalities of toes.
**Results**
Initially she was diagnosed as juvenile arthritis and Raynaud syndrome. Methotrexate was stopped after the first injection because of transaminase elevation; NSAIDs gave only partial response. At our centre, SLE/overlap syndrome was established based on polyarthritis with imaging signs of synovitis/osteitis, proteinuria, lymphopenia, cheilitis, capillaritis ANA positivity, positivity of anti-dsDNA/anti-Sm antibodies, positive Coombs test and increased antiphospholipid antibodies. Overlap features of Juvenile Dermatomyositis (JDM) included Gottron-like rash, proximal muscle weakness, anti-Ku/anti-Ro52 positivity. High disease activity required treatment with prednisolone 1 mg/kg/day, mycophenolate mofetil, hydroxychloroquine and IVIG (0,5 g/kg). Persistent clinical and immunological activity led to initiation of anti-B-cell therapy with rituximab 375 mg/m$^2$. Some phenotype features (asymmetric overgrowth with hypertrophy of one leg, arm, cheek and toes macrodactyly, low extremities vascular malformations) was not typical for rheumatic disease and provoked genetic assessment. Targeted sequencing for segmental overgrowth/vascular malformations revealed a pathogenic PIK3CA variant in exon 21 (NM_006218.4:c.3129G> A; variant allele frequency 6%). PROS was confirmed and patient was referred to oncologists for PROS targeted therapy administration (alpelisib). After immunosuppressive treatment of SLE/overlap syndrome, fever and articular symptoms were improved. Further follow-up is required to assess activity of autoimmune disease and progression of PROS.
**Conclusion**
This case demonstrates that PROS may coexist with systemic autoimmunity and may complicate the interpretation of musculoskeletal symptoms in children with concomitant rheumatic disease. Vascular malformations and asymmetric overgrowth in a child with arthritis should suggest genetic assessment and multidisciplinary discussion.
**Disclosure of interest**
None declared.

## P169

### Damage accrual in juvenile- and adult-onset systemic lupus erythematosus: a multicenter cohort of 485 patients

O. A. Akin[1], M. O. Erkan[2], D. Isıyel[3], A. T. Cakıcı[4], S. Demir[1,*], N. S. Yaşar Bilge[3], Y. Bilginer[2], T. Kaşifoğlu[3], S. Özen[2]
[1]Pediatric Rheumatology, Eskisehir Osmangazi University, Eskisehir; [2]Pediatric Rheumatology, Hacettepe University, Ankara; [3]Rheumatology; [4]Internal Medicine, Eskisehir Osmangazi University, Eskisehir, Türkiye
**Correspondence:** S. Demir
*Pediatric Rheumatology 2026*, **24(S1):**P169

**Introduction**
Systemic lupus erythematosus (SLE) may show different clinical phenotypes and long-term damage patterns according to age at onset.
**Objectives**
To compare clinical involvement and permanent organ damage, assessed by the SLICC/ACR Damage Index (SDI), between juvenile-onset SLE (jSLE) and adult-onset SLE (aSLE).
**Methods**
This retrospective multicenter study included patients with SLE followed in the Pediatric and Adult Rheumatology clinics of Eskisehir Osmangazi University and the Pediatric Rheumatology clinic of Hacettepe University. Patients fulfilling SLICC or ACR criteria with at least 6 months of follow-up were included. Patients diagnosed before 18 years of age were classified as jSLE and those diagnosed at 18 years or older as aSLE. Demographic features, cumulative clinical involvement, and SDI damage domains at diagnosis and last visit were compared.
**Results**
Among 485 patients, 168 had jSLE and 317 had aSLE. Female sex was less frequent in jSLE (76.8% vs. 87.7%, $p$=0.002), and follow-up duration was shorter (5.4 vs. 9.2 years, $p$<0.001). Neurologic and hematologic involvement and lupus nephritis were more prominent in jSLE; lupus nephritis was observed in 45.8% of jSLE and 34.7% of aSLE patients ($p$=0.017). In contrast, permanent organ damage was significantly higher in aSLE. At diagnosis, SDI≥1 was present in 59.3% of aSLE and 17.9% of jSLE patients ($p$<0.001). This difference persisted at the last visit, with SDI≥1 in 74.1% of aSLE and 28.0% of jSLE patients ($p$<0.001). At diagnosis, renal, cardiovascular, musculoskeletal, and cutaneous damage were more frequent in aSLE. At the last visit, neuropsychiatric, renal, pulmonary, cardiovascular, peripheral vascular, musculoskeletal, cutaneous damage, and malignancy were significantly more common in aSLE.
**Conclusion**
Although jSLE was associated with more prominent renal, hematologic, and neurologic involvement, SDI-defined permanent organ damage was markedly higher in aSLE both at diagnosis and at last visit. Age at onset appears to be an important determinant of both clinical phenotype and long-term damage accrual in SLE.
**Disclosure of interest**
None declared.

## P170

### Adherence to and knowledge of non-pharmacological management in childhood-onset systemic lupus erythematosus: a real-world multicenter cross-sectional study aligned with EULAR/ACR recommendations (a PReS Lupus Working Party study)

S. D. Arik[1,*], A. Dudaklı[2], N. Z. Özaslan[3], Z. Hazıroğlu Ökmen[4], A. Alişan[5], D. Alkan[6], Z. Aydın[7], T. Kasap[8], Y. Uğur[9], M. Eksi[10], R. İşgüder[11], F. Çakmak[4], S. Şener[11], M. R. Kışla Ekinci[5], K. Öztürk[7], S. Özdel[6], S. Yüksel[8], N. Şahin[3], B. Çelikel Acar[9], S. Şahin[10], Ö. Kasapçopur[10], E. Marranı[12], D. Rigante[13], B. Aladaileh[14], N. Aktay Ayaz[2]
[1]Pediatric Rheumatology, Sakarya Research and Training Hospital, Sakarya; [2]Pediatric Rheumatology, Istanbul Unıversity Faculty of Medicine, İstanbul; [3]Pediatric Rheumatology, Kocaeli University Faculty of Medicine, Kocaeli; [4]Pediatric Rheumatology, University of Health Sciences Türkiye, Başakşehir Çam and Sakura City Hospital, İstanbul; [5]Pediatric Rheumatology, Çukurova University Faculty of Medicine, Adana; [6]Pediatric Rheumatology, University of Health Sciences Türkiye, Etlik City Hospital, Ankara; [7]Pediatric Rheumatology, Istanbul Medeniyet University Faculty of Medicine, İstanbul; [8]Pediatric Rheumatology, Çanakkale Onsekiz Mart University Faculty of Medicine, Çanakkale; [9]Pediatric Rheumatology, University of Health Sciences Türkiye, Ankara Bilkent City Hospital, Ankara; [10]Pediatric Rheumatology, Istanbul University-Cerrahpaşa, Cerrahpaşa Faculty of Medicine, İstanbul; [11]Pediatric Rheumatology, University of Health Sciences Türkiye, Adana City Training and Research Hospital, Adana, Türkiye; [12]Pediatric Rheumatology, Ospedale Pediatrico Meyer Firenze, Florance; [13]Pediatric Rheumatology, Associate professor at Università Cattolica del Sacro Cuore, Rome, Italy; [14]Pediatric Rheumatology, Queen Rania Children's Hospital, Jordan, Jordan
**Correspondence:** S. D. Arik
*Pediatric Rheumatology 2026*, **24(S1):**P170

**Introduction**
Non-pharmacological interventions are an essential component of comprehensive SLE care, yet real-world data on physician counseling, patient adherence, and perceived benefit in childhood-onset SLE remain limited.
**Objectives**
To investigate the real-world use and perceived value of non-pharmacological interventions in an international multicentre cohort with cSLE.
**Methods**
To evaluate physician advice, patient-reported adherence, and patient-perceived benefit regarding NPIs across eight domains in a multicentre international cSLE cohort, and to identify the largest advice–behaviour gaps. This cross-sectional multicentre study included patients with clinician-confirmed cSLE from Turkish, Jordanian, and Italian. Clinical and survey data were collected using an identical 97-variable case report form. Disease activity was assessed using SLEDAI-2K, PGA, PtGA, and fatigue VAS.
**Results**
A total of 163 patients were included (Türkiye, 77.3%; Jordan, 12.9%; Italy, 9.8%). The cohort was predominantly female (82.7%), with a median age at last visit of 15.0 years [13.6–17.3], a median age at diagnosis of 12.6 years [11.3–14.1], and a median disease duration of 1.8 years [1.3–3.8]. Median SLEDAI-2K decreased from 12 [7–18] at diagnosis to 2 [0–6] at last visit, and median PGA, PtGA, and fatigue VAS were each 1 [0–4]. Physician advice was most frequent for sun/UV protection (93.9%), supplements (77.9%), smoking avoidance (69.3%), healthy diet (62.2%; international cohort only), cold/Raynaud protection (58.7%), regular exercise (51.5%), psychological support (41.1%), and patient education (18.9%; international cohort only). Patient-reported adherence was highest for sun protection (88.3%), smoking avoidance (86.6%), and supplements (81%), but lower for psychological support (42.3%), exercise (33.1%), and educational activities (10.8%). Perceived benefit followed a similar pattern, peaking for sun protection (86.5%) and lowest for educational interventions (39.2%). Sunscreen use was common (90.8%), whereas physical barriers (long sleeves, hats, UV-protective clothing) were less frequent. Vitamin D was the predominant supplement (83.4%), and walking was the most reported activity (51.5%). Only 19.3% were currently receiving psychological support, and 11% had joined a lupus support group, yet 90.8% agreed that physician-provided information had a positive impact on disease management.
**Conclusion**
In this multicentre cSLE cohort, NPIs were commonly discussed and perceived as beneficial, particularly sun protection and supplement use. However, regular exercise and psychological support remained under-recommended and underused, highlighting modifiable targets for structured, culturally adapted NPI counseling.
**Disclosure of interest**
None declared.

## P171

### Validity of the 2023 ACR/EULAR antiphospholipid syndrome (APS) classification criteria to pediatric primary APS: inputs from a systematic literature review

S. Romano[1,*], E. Marrani[1], V. Maniscalco[1], G. Carreras[2], I. Maccora[1], I. Pagnini[1], M. V. Mastrolia[1], T. Giani[1], L. Gatti[1], G. Inguscio[1], G. Simonini[1]
[1]Rheumatology Unit, Meyer Children's Hospital IRCCS; [2]Meyer Children's Hospital IRCCS, Florence, Italy
**Correspondence:** S. Romano
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**Introduction**
Pediatric antiphospholipid syndrome (APS) is a rare autoimmune condition characterized by thrombotic and/or non-thrombotic manifestations associated with persistent antiphospholipid (aPL) antibodies. Al-

though the 2023 ACR/EULAR APS classification criteria were developed and validated in adults, their applicability and diagnostic performance in pediatric primary APS have not yet been established.
**Objectives**
To assess the sensitivity of the 2023 ACR/EULAR APS classification criteria in pediatric primary APS and to compare their performance with the 2006 revised Sapporo criteria.
**Methods**
A systematic literature review was conducted up to September 30, 2025, in accordance with PRISMA guidelines, to identify pediatric patients (<18 years) with primary APS. Individual patient data were extracted from eligible studies. Each case was assessed for fulfillment of both the 2006 Sapporo and 2023 ACR/EULAR classification criteria. Sensitivity was estimated using a two-stage individual participant data–style random-effects meta-analysis.
**Results**
A total of 118 studies were included, encompassing 148 pediatric patients with primary APS (median age 11.5 years; 56.5% male). Thrombotic events occurred in 93.2% of patients, while non thrombotic manifestations were reported in 56.1%, with thrombocytopenia being the most common (29%). Lupus anticoagulant was the most frequently detected aPL antibody (72.3%). The 2006 Sapporo criteria classified 135 patients corresponding to a sensitivity of 91.2% (95% CI 85.4–95.2). The 2023 ACR/EULAR criteria classified 123 patients with a sensitivity of 83.1% (95% CI 76.2–88.3). Among the 25 non-classified patients, most failed to meet laboratory entry criteria due to low-titer aCL and/or anti-β2GPI antibodies. Inclusion of non-thrombotic manifestations improved clinical capture, but did not fully compensate for laboratory restrictions.
**Conclusion**
The 2023 ACR/EULAR APS classification criteria show suboptimal sensitivity in pediatric primary APS compared with the 2006 Sapporo criteria. Differences in clinical presentation and aPL antibody profiles between pediatric and adult APS limit direct transposition of adult-derived criteria. These findings highlight the need for pediatric-specific APS classification criteria to enhance diagnostic accuracy, research consistency, and disease characterization in children.

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**Disclosure of interest**
None declared.

## P173

### Autoantibody profile and its relationship with clinical phenotypes in juvenile Sjögren's disease: a single-center experience

P. Zeytun Baloglu[1,*], S. Atamyıldız Uçar[1], N. Kara Çanlıoğlu[1], E. Tunce[1], H. Öztürk Aydın[1], G. Doğrusadık Pirim[1], S. Başaran Çoban[1], H. Kandemir Baloğlu[1], S. Türkmen[1,2], B. Sözeri[1]
[1]Department of Pediatric Rheumatology, Umraniye Training and Research Hospital, University of Health Sciences; [2]Department of Pediatric Rheumatology, Prof. Dr. İlhan Varank Sancaktepe Training and Research Hospital, Istanbul, Türkiye
**Correspondence:** P. Zeytun Baloglu
*Pediatric Rheumatology 2026*, **24(S1):**P173

**Introduction**
Juvenile Sjögren's disease (jSjD) is a rare and heterogeneous systemic autoimmune disorder. Although Sjögren-associated autoantibodies are important diagnostic markers, the clinical implications of seronegativity in jSjD remain limited.
**Objectives**
To compare the clinical and immunological characteristics of triple-seronegative jSjD patients with those positive for at least one Sjögren-associated autoantibody.
**Methods**
This retrospective cross-sectional study included 23 jSjD patients diagnosed and followed at a tertiary pediatric rheumatology center between June 2016 and May 2026. Demographic data, autoantibody profiles, clinical findings and treatment characteristics were analyzed. ANA-positive patients were stratified into two groups according to Sjögren-associated serology triple-seronegative (negative for anti-SSA, anti-SSB, and RF) and patients with positivity for at least one autoantibody.
**Results**
Among 23 patients, 21 (91.3%) were female. Median age at diagnosis was 13.0 years (IQR, 10.6-16.4), with a median follow-up duration of 42 months (IQR, 19.0-64.0). ANA positivity was present in 19 patients (82.6%). After excluding ANA-negative patients, 12 were classified as triple-seronegative and 7 had positivity for at least one Sjögren-associated autoantibody, including anti-SSA ($n$=6), RF ($n$=4), and anti-SSB ($n$=1). There was no statistical significance between groups regarding age at symptom onset or diagnosis ($p$=0.38 and $p$=0.43, respectively). Diagnostic delay was significantly longer in triple-seronegative patients ($p$=0.045). The most common symptom was xerophthalmia ($n$=15, 78.9%), followed by xerostomia ($n$=14, 73.7%) and arthritis ($n$=11, 57.9%). Only 1 (5.3%) patient was diagnosed with interstitial nephritis. Xerophthalmia was significantly more frequent in triple-seronegative patients (100% vs. 42.9%, $p$=0.009), whereas photosensitivity and hypergammaglobulinemia were more common in patients positive for at least one Sjögren-associated autoantibody (42.9% vs. 0% for both variables; $p$=0.036). ESSDAI scores at initial and final visits did not differ significantly between groups ($p$=0.536, $p$=0.967, respectively). Hydroxychloroquine was initiated in 21 patients (91.3%), while methotrexate in 19 (82.6%) and mycophenolate mofetil in 9 (39.1%). Rituximab therapy was required in 4 patients (17.4%), including 1 with triple-seronegative and interstitial nephritis.
**Conclusion**
Triple-seronegative jSjD patients demonstrated longer diagnostic delay and distinct clinical and immunological features compared with patients positive for at least one Sjögren-associated autoantibody. The absence of Sjögren-associated autoantibodies may not preclude the diagnosis of jSjD, and comprehensive clinical evaluation remains important, particularly in patients with suggestive sicca manifestations. Larger multicenter studies are needed to better define seronegative phenotypes in jSjD.
**Disclosure of interest**
None declared.

## P174

### Clinical phenotype and long-term follow-up of nephronophthisis type 1 complicated with Sjögren's syndrome

L. Zhong[1,*], J. Ding[1], S. Jian[1], M. Ma[1], H. Song[1]
[1]Peking Union Medical College Hospital, Beijing, China
**Correspondence:** L. Zhong
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**Introduction**
NA.
**Objectives**
To summarize the clinical characteristics and long-term follow-up outcomes of the first pediatric case of nephronophthisis type 1 (NPHP1) complicated with Sjögren's syndrome, and to improve clinicians' recognition of this rare comorbidity.
**Methods**
We retrospectively analyzed the clinical manifestations, whole-exome sequencing results, treatment strategies and follow-up data of a pediatric patient with genetically confirmed nephronophthisis type 1 caused by *NPHP1* gene mutation complicated with Sjögren's syndrome, who was hospitalized and managed in Peking Union Medical College Hospital.
**Results**
A 13-year-old female presented with fever and parotid swelling/pain in June 2023. Laboratory tests showed elevated serum creatinine (92.1

μmol/L) with normal urine routine, and persistent hypercreatininemia was confirmed 3 months later. Serological tests were positive for ANA (granular pattern, 1:1000), anti-SSA, anti-SSB, and elevated RF-IgA/IgM; salivary gland ultrasonography, decreased unstimulated salivary flow, reduced tear film break-up time, xerostomia, and labial gland biopsy confirmed Sjögren's syndrome (SS), and hydroxychloroquine (0.1 g twice daily) was initiated. Follow-up revealed mild-to-moderate normocytic normochromic anemia (Hb 85–92 g/L), fluctuating serum creatinine (91-106 μmol/L), and progressive elevation of blood urea nitrogen; mycophenolate mofetil was added in October 2023 without improvement. Further examinations showed Hb 77–80 g/L (normal hemoglobin electrophoresis, iron metabolism, folic acid, vitamin B12, erythropoietin, negative Coombs test), decreased hematopoietic tissue on bone marrow biopsy, elevated renal function indicators (eGFR 59 mL/min/1.732 $m^2$), slightly enhanced bilateral renal cortical echogenicity, and chronic tubulointerstitial injury on renal biopsy. Hereditary renal disease was suspected, and whole-exome sequencing identified a homozygous NPHP1 gene deletion (EX1-EX20 Del) inherited from both parents. Mycophenolate mofetil was discontinued, and symptomatic treatments were added. After over 2 years of follow-up, the patient was in good condition with stable Hb (110–120 g/L), relatively stable renal function (eGFR 40-51 mL/min/1.732 $m^2$), and persistent high-titer ANA/anti-SSA/anti-SSB antibodies. No similar cases have been reported.

**Conclusion**

Chronic renal insufficiency is rare in pediatric SS. Timely genetic testing for hereditary ciliopathy-related nephropathy is recommended in children with ineffective immunosuppressive therapy and atypical immune-mediated renal pathology. This is the first report of nephronophthisis combined with autoimmune rheumatic disease, suggesting genetic ciliary defects may increase autoimmune susceptibility. Pathological and genetic examinations are crucial for differential diagnosis and individualized treatment to avoid blind immunosuppression, and this case's long-term data provides valuable clinical evidence for such ultra-rare comorbidities.

**Disclosure of interest**

None declared.

## P175

### Clinical and immunological spectrum of pediatric overlap syndromes: a single-center cohort from Kazakhstan

A. Kappassov[1], Z. Mukusheva[1,*], M. Assylbekova[1]
[1]Pediatric Rheumatology, CF "University medical center", Astana, Kazakhstan
**Correspondence:** Z. Mukusheva
*Pediatric Rheumatology 2026*, **24(S1):**P175

**Introduction**

Pediatric overlap syndromes are rare immune-mediated rheumatic diseases characterized by the coexistence of clinical and serological features of two or more connective tissue diseases. Diagnosis is challenging because of significant phenotypic variability, multisystem involvement, and the absence of standardized pediatric management strategies. Data from Central Asia remain limited.

**Objectives**

To evaluate the clinical manifestations, immunological characteristics, and treatment approaches in children diagnosed with overlap syndromes at a tertiary pediatric rheumatology center in Kazakhstan.

**Methods**

A retrospective cohort study was conducted at the pediatric rheumatology department of the Corporate Fund "University Medical Center" (UMC), Kazakhstan. Medical records from January 2024 to December 2025 were reviewed. Among 50 screened patients, 12 children met inclusion criteria: age <18 years, newly diagnosed overlap syndrome, and complete clinical and immunological data. Diagnoses were established according to international classification criteria, including EULAR/ACR recommendations.

**Results**

The mean age at diagnosis was 11 years, with female predominance (75%). The estimated prevalence of pediatric overlap syndromes in Kazakhstan in 2025 was 1.01 per 100,000 children. Clinical manifestations were heterogeneous: juvenile idiopathic arthritis was present in 58% of patients, systemic lupus erythematosus in 42%, and juvenile dermatomyositis and juvenile systemic sclerosis each in 33%. Sjögren's syndrome occurred in 25% of cases. Autoimmune hepatitis and neurological involvement were identified in 8% each. Immunological evaluation demonstrated marked heterogeneity. Anti-U1-RNP/Sm antibodies were detected in 67% of patients, Ro52 in 58%, DFS70 in 42%, SSA in 33%, PM/Scl-75/100 in 25%, Ku in 17%, and PCNA and CENP antibodies in 8% each. Multiple autoantibody positivity was observed in 75% of the cohort. All patients received immunosuppressive therapy. Combination treatment was required in 54% of cases, while 36% received monotherapy. Systemic glucocorticoids were administered in 75% of patients, hydroxychloroquine in 67%, mycophenolate mofetil in 58%, and methotrexate in 33%. Biologic therapy, including rituximab or tocilizumab, was used in 17% of cases.

**Conclusion**

Pediatric overlap syndromes demonstrate substantial clinical and immunological heterogeneity, frequent multiple autoantibody positivity, and a high need for combination immunosuppressive therapy. Treatment strategies are mainly phenotype-driven rather than diagnosis-based. These findings emphasize the need for pediatric-specific registries and standardized diagnostic and therapeutic approaches.

**Disclosure of interest**

None declared.

## P176

### Middle aortic syndrome associated with a pathogenic PPP1R12A variant: differential diagnosis from Takayasu arteritis in a pediatric patient

C. Udaondo[1,*], N. C. Fuertes[2], R. Alcobendas Rueda[1], G. Moral Garcia[3], A. Hercberg Moreno[1], A. Remesal Camba[1], L. Martinez Marcelo[1], S. Barreiro Perez[1]
[1]Pediatric Rheumatology, Hospital La Paz, Madrid; [2]Pediatric, Hospital Puerta del Mar, Cadiz; [3]Rheumatology, Hospital Universitario Virgen Macarena, sevilla, Spain
**Correspondence:** C. Udaondo
*Pediatric Rheumatology 2026*, **24(S1):**PR176

**Introduction**

Middle aortic syndrome (MAS) is a rare vascular disorder characterized by segmental narrowing of the abdominal or distal thoracic aorta and its major branches, frequently resulting in severe renovascular hypertension. In pediatric patients, distinguishing congenital or genetic vasculopathies from inflammatory arteriopathies such as Takayasu arteritis is often challenging.

**Objectives**

To describe a case of severe renovascular hypertension secondary to MAS in a toddler initially evaluated for Takayasu arteritis, in whom a pathogenic PPP1R12A variant was identified.

**Methods**

Medical record review and a focused literature research.

**References**

1. A 2-year-old girl presented with recurrent morning vomiting, polyuria, polydipsia, and failure to thrive. Past history included neonatal erosive scalp dermatosis and mild motor delay. Initial laboratory evaluation revealed hyponatremia, hypokalemia, hypochloremia, hypophosphatemia, and metabolic alkalosis, together with marked proteinuria, hematuria, thrombocytopenia, and schistocytes, consistent with microangiopathic hemolytic anemia. Following fluid resuscitation, she developed severe hypertension with end-organ involvement, requiring admission to the pediatric intensive care unit and multiple antihypertensive agents. Echocardiography demonstrated severe left ventricular hypertrophy with right ventricular outflow tract obstruction. Computed tomography angiography revealed diffuse narrowing of the abdominal aorta involving both renal arteries, with critical proximal stenosis of the right renal artery, consistent with MAS. Renal scintigraphy confirmed functional exclusion of the right kidney. Given the initial suspicion of Takayasu arteritis, vascular PET-CT and cranial/cervical MR angiography were undertaken, showing no evidence of active vasculitis. Genetic testing identified a heterozygous de novo pathogenic PPP1R12A variant (c.658C> T; p.Gln220Ter), a gene recently linked to developmental malformation syndromes. With intensive blood pressure control, hemolysis resolved and cardiac function partially improved.

**Conclusion**
MAS should be considered in young children presenting with severe hypertension and electrolyte disturbances. Advanced vascular imaging is crucial to exclude inflammatory vasculitis, particularly Takayasu arteritis. This case broadens the phenotypic spectrum associated with PPP1R12A variants and suggests a potential link with complex vascular developmental anomalies, in a context of very limited and heterogeneous published cases.

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**Disclosure of interest**
None declared.

## P177

**Role of immunosuppression in desmosomal cardiomyopathy**

K. Rücklová[1,*], M. Prokopius[2], F. Blankenburg[1], A. Heinkele[1], A. Hospach[1]
[1]Pädiatrie 2; [2]Pädiatrie 3, Olgahospital, Stuttgart, Germany
**Correspondence:** K. Rücklová
*Pediatric Rheumatology 2026*, **24(S1):**P177

**Introduction**
A 16-year-old boy first presented in June 2025 with back and chest pain, tachydyspnea, presyncope, and palpitations. Concomitantly, cough, intermittent fever, and increased sweating were present. Laboratory results showed a significant inflammatory response (CRP 220 mg/l) as well as markedly elevated cardiac markers (hs-troponin I 11,000 ng/l, normal < 45; NT-proBNP 9,000 pg/ml, normal < 400). Chest radiography showed bilateral pulmonary infiltrates. The ECG showed T-wave inversions in II, III, aVL, aVF, and V5 leads and he had an impaired left ventricular function on echocardiography. Cardiac MRI confirmed florid myocarditis of the left ventricle. No pathogen could be detected in blood cultures and serologic investigations.
**Therapy**
Inotropy-enhancing therapy with milrinone and levosimendan was administered, later supplemented with lisinopril, bisoprolol, and spironolactone with a good effect on the left ventricular function. In accordance with guidelines, intravenous immunoglobulins were administered.
**Further course**
The inflammatory markers normalized. However, troponin remained chronically mildly elevated. Six months later, the symptoms with chest pain, massive rise in troponin to 20,000 ng/l and transiently decreased pump function recurred. Endomyocardial biopsy yielded no pathogen. A renewed administration of immunoglobulins led to a further rise in troponin. At this point markedly dry feet as well as woolly hair were noted.
**Objectives: Discussion**
The combination of recurrent myocarditis-like episodes, chronically elevated troponin, and cutaneous abnormalities suggest an arrhythmogenic cardiomyopathy due to pathogenic variants in desmoplakin or other desmosomal genes. The desmosomal complex plays a central role in cell adhesion of cardiac and skin tissue. Hence, pathogenic desmosomal variants lead to structural instability, secondary inflammation and fibrotic remodeling with arrhythmogenic potential. The myocarditis-like episodes or the so-called "hot phases" are characterised by acute troponin release and inflammatory infiltration of the myocardium. Studies show a reduction of arrhythmia and heart failure risk through early, short-term immunosuppression in these phases. Therefore, the patient received methylprednisolone pulse therapy, which led to a rapid decline in troponin. Results of the genetic analysis are pending.
**Methods: Conclusion**
The case illustrates a relatively new disease entity, which requires a prompt immunosuppressive treatment and therefore a high level of suspicion even before genetic results are available. It is an example of an interplay between a genetic structural abnormality with secondary inflammation. And finally, the case underscores the importance of interdisciplinary collaboration between cardiology and rheumatology/immunology experts.

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**Disclosure of interest**
None declared.

## P178

**Unexplained rash in a toddler with familial autoimmunity: a diagnostic dilemma**

D. Mondal[1,*], V. Arunath[1,2], G. Varnier[1], A. Chieng[1]
[1]Paediatric Rheumatology, Royal Manchester Children's Hospital, Manchester, UK; [2]Paediatrics, Postgraduate Institute of Medicine, University of Colombo, Colombo, Sri Lanka
**Correspondence:** D. Mondal
*Pediatric Rheumatology 2026*, **24(S1):**P178

**Introduction**
Monogenic systemic lupus erythematosus (SLE) is rare, severe, and usually presents early in life. Deficiencies of early classical complement pathway components, particularly C1q, are strongly associated with lupus-like disease due to impaired clearance of apoptotic cells and immune complexes. Patients often present with severe cutaneous disease, recurrent infections, vasculitis, and early-onset lupus manifestations.
**Objectives**
Objective of the case report was to report a toddler with a complex clinical presentation and significant family history of autoimmunity, highlighting the diagnostic challenges and the role of genetic testing in identifying rare form of monogenic lupus.
**Methods**
A 2-year-6-month-old boy born to consanguineous parents was referred for evaluation of unexplained rash and family history of SLE. At 2 months of age, he developed annular facial and truncal rashes during sun exposure, which resolved spontaneously. At 28 months, he developed erythematous lesions on the ears and chest during a febrile illness, progressing to blistering vasculitic lesions involving the face, trunk, palms, soles, and purpuric toe lesions. He also had recurrent chest infections and speech delay. Investigations showed positive ANA and SSA/Ro antibodies with negative dsDNA. Complement studies demonstrated persistently low CH50 (<12.75) and profoundly low C1q (<13), while C3 and C4 were normal/slightly elevated. Skin biopsy showed interface dermatitis, and direct immunofluorescence demonstrated weak granular IgM deposition along the basement membrane with absent C1q staining, supporting lupus. Given the early onset, consanguinity, recurrent infections, and family history, monogenic lupus and interferonopathies were considered. Genetic testing identified a homozygous C1qA mutation (c.622C> T; p.Gln208Ter), confirming autosomal recessive C1q deficiency causing monogenic SLE. The patient was treated with prednisolone and mycophenolate mofetil with partial improvement. Due to the aggressive nature of monogenic lupus and limited response to standard immunosuppression, he was referred for bone marrow transplant evaluation.
**Conclusion**
Monogenic lupus should be considered in young children with lupus-like features, consanguinity, recurrent infections, family history of autoimmunity, and low C1q levels. Early genetic diagnosis is important for prognosis, targeted therapy, and consideration of definitive treatment options such as haematopoietic stem cell transplantation.
**Disclosure of interest**
None declared.

## P179

### Clinical case: severe paediatric GPA with myopericarditis — from diagnostic pitfall to remission with biologic maintenance

A. M. Laterza[1,2,*], N. Cabrera[2,3], L. Spinzi[4], C. Montiel[5], M. Martinez-Pico[6], M. Ramos[3], J. Segovia[4]
[1]Pediatric, IPS; [2]Universidad Catolica Nuestra Señora de la Asuncion; [3]Rheumatology Pediatric; [4]Radiology; [5]Cardiology Pediatric; [6]Nephrology, IPS, Asuncion, Paraguay
**Correspondence:** A. M. Laterza
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**Introduction**
Granulomatosis with polyangiitis (GPA) is a rare ANCA-associated vasculitis with an estimated incidence of 0.1/100,000 children/year. Cardiac involvement occurs in up to 30% of adult cases but is rarely documented in paediatric GPA.
**Objectives**
To describe a paediatric case of GPA with MRI-confirmed myopericarditis, severe crescentic nephritis, and elevated inflammatory markers including procalcitonin — mimicking severe bacterial infection — and to report the 9-month outcome of combined RTX-MMF maintenance in a resource-limited tertiary centre.
**Methods**
Case report of a 10-year-old girl with 2 months of fever, fatigue, epistaxis, and generalised pain, initially treated as bacterial infection without response. Evaluation included serial haematology, CRP, PCT, cardiac biomarkers, urinalysis, blood cultures, chest/sinus/abdominal CT, cardiac and renal MRI, and renal biopsy. PR3/MPO-ANCA serology performed by ELISA. Treatment followed EULAR/PReS guidelines.
**Results**
PR3-ANCA positivity with four-system involvement was confirmed. CRP peaked at 192 mg/L and procalcitonin at 9.16 ng/mL — overlapping with severe bacterial infection, complicating early diagnostic. Blood cultures were sterile. Cardiac MRI showed non-ischemic mesoepicardial fibrosis with segmental hypokinesia. Renal biopsy confirmed pauci-immune crescentic glomerulonephritis (21.4% cellular crescents); renal deterioration was rapid (micro- to macrohematuria within 72 h). Induction comprised IV methylprednisolone, IV cyclophosphamide, and rituximab. Maintenance was established with RTX and rapidly combined with MMF 1 g/day because persistance of biological inflammation. At 9 months, CRP normalised, proteinuria fell from 700 to 162 mg/24 h with preserved renal function (creatinine 0.61 mg/dL), cardiac MRI showed resolution of inflammatory activity, and gonadal function was preserved despite cyclophosphamide exposure. Chest CT confirmed pulmonary nodule resolution.
**Conclusion**
This is the first comprehensively documented paediatric GPA case from Paraguay. Markedly elevated procalcitonin mimicked severe bacterial infection, highlighting the diagnostic challenge of GPA in resource-limited settings where advanced serology may be delayed. MRI-confirmed cardiac involvement and rapid crescentic nephritis progression underscore the severity of paediatric GPA onset. Combined RTX-MMF maintenance achieved clinical, laboratory, and imaging remission at 9 months with preserved gonadal function — a critical safety outcome in a prepubertal girl exposed to cyclophosphamide.

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**Disclosure of interest**
None declared.

## P181

### Under the mask of vasculitis: a rare pediatric case of MacDuffie syndrome

A. A. Abushhaiwia[*,1]
[1]Pediatric Rheumatology, Tripoli Children's Teaching Hospital, Faculty of Medicine, University of Tripoli, Tripoli, Libya
**Correspondence:** A. A. Abushhaiwia
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**Introduction**
Hypocomplementemic urticarial vasculitis syndrome (HUVS), also known as MacDuffie syndrome, is a rare immune-complex mediated small-vessel vasculitis characterized by urticarial or vasculitic skin lesions, persistent hypocomplementemia, and systemic involvement.
**Objectives**
To report a rare pediatric case of HUVS presenting with severe cutaneous vasculitic lesions and renal involvement.
**Methods A 4-year-old** Libyan girl, born to consanguineous parents, presented with a one-month history of progressive bullous hemorrhagic, ulcerative, and necrotic skin lesions involving the limbs, trunk, and abdomen, with predominant involvement of the lower extremities including toes and fingers. Associated symptoms included abdominal pain and arthralgia without fever, preceded by an upper respiratory tract infection. She had a prior history of recurrent urticarial rash previously attributed to cow's milk protein allergy. She had been sequentially misdiagnosed as hand-foot-mouth disease and then IgA vasculitis.On examination, she had bullous hemorrhagic lesions, crusted ulcers, and ischemic changes affecting the fingers. Initial laboratory investigations showed WBC $10.6\times10^3$/µL, HGB 8.8 g/dL, PLT $568\times10^3$/µL, ESR 27 mm/hr, CRP 19 mg/L. Renal and LFT were normal. Urinalysis revealed significant microscopic hematuria (RBC60–65/HPF) with normal protein/creatinine ratio. low C3 (0.79 g/L) with low C4 (0.17 g/L). ANA was borderline/doubtful, anti-dsDNA was negative. ANCA, p-ANCA with elevated (MPO 21 IU/mL), while PR3 was negative.Skin biopsy demonstrated dermal neutrophilic infiltrates without definite leukocytoclastic vasculitis or granulomatous inflammation, consistent with neutrophilic dermatosis.The patient received (IVIG 2 g/kg once) and intravenous methylprednisolone pulses (30 mg/kg/day for 3 days), followed by oral prednisolone with partial response. Due to renal involvement, renal biopsy was performed and confirmed immune-complex mediated small-vessel vasculitis consistent with hypocomplementemic urticarial vasculitis (MacDuffie syndrome).
**Treatment and Outcome**
She was maintained on oral prednisolone tapered to 7.5 mg/day and hydroxychloroquine 100 mg daily. Methotrexate was discontinued after stabilization.Thepatient showed marked clinical improvement with resolution of systemic symptoms, although mild residual cutaneous lesions persisted over the lower limbs. During follow-up, no progression of renal impairment was observed.
**Conclusion**
MacDuffie syndrome is a rare pediatric hypocomplementemic vasculitis that may mimic IgA vasculitis, neutrophilic dermatoses, and infectious skin diseases.This case highlights the importance of sequential biopsies, particularly renal biopsy, in establishing diagnosis. Early recognition is essential to prevent irreversible renal damage and improve long-term outcomes.
**Disclosure of interest**
None declared.

## PR182

### Urinary microalbumin and neutrophil gelatinase–associated lipocalin in children with IgA vasculitis: a long-term follow-up study

B. Buddhpriya[1,*], D. Desai[2], P. Suku[2], V. Joshi[2], M. Dhaliwal[2], S. Sharma[2], R. K. Pilania[2], V. Pandiarajan[2], A. Rawat[2], A. Gupta[2], D. Suri[2]
[1]Department of Pediatrics; [2]Pediatric Allergy Immunology Unit, Advanced Pediatrics Center, Postgraduate Institute of Medical Education and Research, Chandigarh, India
**Correspondence:** B. Buddhpriya
*Pediatric Rheumatology 2026*, **24(S1):**PR182

**Introduction**
Renal involvement is the principal determinant of long-term outcome in IgA vasculitis (IgAV). Current recommendations emphasize urine examination and blood pressure monitoring for six months after disease onset; however, subtle renal injury may be underrecognized and result in sequalae. Data on long-term urinary biomarkers in children with IgAV from the Indian subcontinent are limited.
**Objectives**
To evaluate urinary microalbumin and urinary neutrophil gelatinase-associated lipocalin (NGAL) in children with IgAV at ≥2 years after disease onset, and to compare findings between patients with and without nephritis at diagnosis.

**Methods**
This prospective cohort study was conducted at the Pediatric Allergy and Immunology Unit, PGIMER, Chandigarh. Children with IgA vasculitis diagnosed using EULAR/PReS/PRINTO criteria and with ≥2 years of follow-up from disease onset were enrolled ($n$=50). Patients on active immunosuppression or unclear diagnosis were excluded. Follow-up assessment included growth, blood pressure, serum urea and creatinine, urinalysis, and spot urine protein–creatinine ratio. First-void urine samples were analysed for microalbumin via nephelometry (albumin–creatinine ratio) and NGAL using standardized assays. Non-parametric statistical tests were applied.
**Results**
At a median follow-up of approximately 6.5 years after disease onset, renal function was preserved in all 50 children, with normal serum urea and creatinine values across the cohort. Spot urine protein–creatinine ratio (UPCR) was <0.2 in 47/50 (94%) children, while 3/50 (6%) had persistent sub-nephrotic proteinuria; all three had IgAV nephritis at diagnosis. No child had nephrotic-range proteinuria. Microscopic haematuria was detected in 4/50 (8%) children, including two with IgAV nephritis and two without nephritis at onset. Microalbuminuria (urinary albumin–creatinine ratio 30–300 µg/g) was present in 4/50 (8%) children. Two had prior biopsy-proven IgAV nephritis with persistent sub-nephrotic proteinuria despite completion of immunosuppressive therapy, while two had no nephritis at diagnosis or during initial 6-month follow-up; one of these had stage 1 hypertension with otherwise normal renal parameters. Urinary NGAL and NGAL/creatinine levels did not differ significantly between controls, IgAV without nephritis, and IgAV nephritis (median NGAL 6.08, 9.41, and 5.70 ng/mL, respectively; $p$=0.217; NGAL/creatinine $p$=0.234). All children with residual urinary abnormalities had received prior immunomodulatory therapy, with longer and more intensive regimens in those with higher ISKDC grades.
**Conclusion**
At long-term follow-up, most children with IgA vasculitis had preserved renal function; however, **microalbuminuria revealed persistent or subclinical renal involvement**, including in children without nephritis at onset. In contrast, **urinary NGAL did not discriminate chronic renal sequelae**. These findings suggest that **short-term follow-up is insufficient** and support incorporating **long-term blood pressure and microalbuminuria surveillance** to better identify delayed renal risk in IgA vasculitis.
**Disclosure of interest**
None declared.

## P185

### Clinical phenotypes, diagnosis, and interventional outcomes in childhood-onset Takayasu arteritis: a longitudinal cohort study

D. Desai[1,*], R. K. Pilania[1], V. Pandiarajan[1], D. Suri[1], R. M. Kumar[2], R. Vijayvergiya[2], A. K. Saxena[3], A. Gupta[1]
[1]Pediatric Allergy Immunology Unit, Department of Pediatrics; [2]Cardiology; [3]Radiodiagnosis and Imaging, PGIMER, Chandigarh, India
**Correspondence:** D. Desai
*Pediatric Rheumatology 2026*, **24(S1):**P185

**Introduction**
Childhood-onset Takayasu Arteritis (c-TA) is a rare, life-threatening large-vessel vasculitis that frequently presents with hypertension. The diagnosis is often delayed due to non-specific early systemic symptoms.
**Objectives**
This study aimed to delineate the clinical spectrum, gender-specific phenotypes, Numano anatomical classifications, and the long-term outcomes of immunosuppressive and endovascular interventions in a longitudinal paediatric cohort.
**Methods**
Data was collected by retrospective review of clinical records of 77 patients diagnosed with c-TA at a tertiary referral centre in North Western India between 2001 and 2026. Demographic variables, clinical presentations, laboratory markers, and serial CT/MR angiographic findings were entered in a predesignated proforma. Disease distribution was categorized using the Numano classification. Treatment regimen, including corticosteroid duration and biologic usage, along with details of vascular interventions (timing, complications, and outcomes), were evaluated.
**Results**
The mean age at diagnosis of 9.8 ± 3.4 years. Females constituted 55.8% ($n$=43) of the patients. Median diagnostic delay was 3.0 months (range 2 days–6 years). Male patients demonstrated more severe renovascular disease, with renal artery involvement observed in 73.3% of males compared with 46.5% of females ($p$ = 0.03). Hypertension was similarly more frequent among males (80.0% vs. 69.8%). Hypertension was the predominant presenting feature overall, affecting 70.1% ($n$=54), followed by pulse deficits in 63.6% ($n$=49), headache in 50.6% ($n$=39), and congestive cardiac failure in 40.3% ($n$=31). Patients presenting with heart failure demonstrated severe myocardial dysfunction, with a mean echocardiographic ejection fraction of 26.8%. Among 76 patients with classifiable angiographic imaging, Numano Type IV disease was the dominant phenotype, observed in 53.9% ($n$=41). 59.3% ($n$=32/54) of tested patients tested positive on Tuberculin skin testing, and 65.4% ($n$=50) received concurrent anti-tubercular therapy. Immunosuppressive therapy included methotrexate in 82.7% ($n$=64) and cyclophosphamide in 9.8% ($n$=8), along with prolonged corticosteroids in most. 44.2% ($n$=34) required vascular intervention, including angioplasty, stenting, or nephrectomy. At last follow-up, 71.4% ($n$=24/34) of patients undergoing intervention achieved clinical stabilization or remission. Despite treatment, persistent disease activity or relapse remained common, and overall mortality was 3.9% ($n$=3).
**Conclusion**
c-TA presents as a predominantly Type IV, renal disease with a phenotype that is more severe in males. The rapid development of hypertensive cardiomyopathy and high rates of multi-vessel endovascular interventions highlight the critical need for early diagnosis.
**Disclosure of interest**
None declared.

## P186

### Aortoarteritis in tuberculosis: a diagnostic and therapeutic enigma

P. Kaur[1,*], S. Chandola[2], Y. Dharmashaktu[3], S. A. Shamim[3], M. Jana[2], N. Bagri[1]
[1]Pediatrics; [2]Radiodiagnosis & Interventional Radiology; [3]Nuclear medicine, All India Institute of Medical Sciences, New Delhi, New Delhi, India
**Correspondence:** P. Kaur
*Pediatric Rheumatology 2026*, **24(S1):**P186

**Introduction**
Aortitis is rare in children (0.01 cases/100,000 children per year 1), with rheumatological illnesses as the most common causative pathology, apart from infectious and idiopathic fibrotic, each bearing a distinct management implication. Although exceedingly uncommon, tuberculous aortitis is difficult to clinically or radiologically distinguish from Takayasu arteritis, a matter further complicated by their reported coexistence. This case elucidates the challenges of diagnosing and treating aortitis in the context of tuberculosis in an immunocompromised child.
**Objectives**
A 13-year-old girl, completing six months of antitubercular treatment (ATT) for pulmonary tuberculosis, was referred for a pediatric rheumatology opinion due to new-onset fever and left limb pulselessness, detected during examination in her recent routine visit. A review of history revealed that the child had been symptomatic since the age of 7 years, with four instances of disseminated tuberculosis in a period of five years, with microbiological evidence of a sensitive organism in two, and a recent onset of pulselessness. Evaluation aimed to discern tubercular infective aortoarteritis from immune-mediated arteritis and identify the underlying error of immunity. Inflammatory markers were elevated, suggestive of active disease. Thus, a PET-CT was undertaken to map the extent of involvement. There was an increase in the size of cavitation with multiple nodules with high metabolic activity and fibrosing mediastinitis with vascular constriction, compared to previous imaging. A subclavian artery biopsy was undertaken once the inflammatory markers normalised. It did not reveal any granuloma nor acid-fast bacilli. The initial Indian Takayasu Clinical Activity Score (ITAS) of 6. Hence, the plan was to treat with anti-tubercular therapy (ATT), oral steroids and methotrexate in this case. Whole-exome sequencing revealed a heterozygous IL12RB1 defect (autosomal recessive), report-

edly pathogenic, considering the disease phenotype. As she had leucopenia and low-normal platelet count, a bone marrow examination was performed, which was normal. She was found to have worsening ITAS (12) on steroid tapering, and hence treatment was escalated to Tocilizumab, with concomitant ATT prophylaxis. The disease activity improved thereafter, 3 months into therapy (ITAS 8).

**Methods: Conclusion**

Therapy in the index case consisted of a three-pronged approach, i.e., management of active tuberculosis, immune-mediated vasculits, and prevention of opportunistic infections. The context of repeated, disseminated infections in precarious sites or with atypical organisms should alert a physician towards a possible inborn error of immunity. Moreover, molecular diagnosis of such cases might provide an opportunity for precision therapy and prognostication. Takayasu arteritis may be an epiphenomenon of IL12RB1 defect-associated autoimmunity in this therapeutically challenging case.

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**Disclosure of interest**

None declared.

## P188

**Comparison of clinical characteristics and treatment outcomes of Kawasaki disease in two countries: a propensity score-matched multicentre study**

B. Başer Taşkın[1,*], A. Doğru Kılınç[1], A. Dudaklı[1], D. Bartkus[2], J. A. John[3], K. S. E. Marucoikal[3], S. B. Smith[3], F. Çakmak[4], Ö. Akgün[1], A. Šnipaitienė[2], N. Aktay Ayaz[1]

[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine, İstanbul, Türkiye; [2]Pediatric Department; [3]Medical Academy of Lithuanian University of Health Sciences, Kaunas, Lithuania; [4]Pediatric Rheumatology, Basaksehir Cam and Sakura City Hospital, İstanbul, Türkiye

**Correspondence:** B. Başer Taşkın

*Pediatric Rheumatology 2026*, **24(S1):**P188

**Introduction**

Kawasaki disease (KD) is an acute pediatric medium-vessel vasculitis associated with heterogeneous clinical manifestations and variable cardiovascular outcomes across different populations. Despite increasing recognition of geographic and ethnic differences in disease presentation and outcomes, comparative data between different geographic regions remain limited.

**Objectives**

This study aimed to compare the clinical characteristics, cardiac involvement, and treatment outcomes of pediatric patients with KD in Türkiye and Lithuania using a propensity score-matched analysis.

**Methods**

This multicentre retrospective study included 125 patients from Türkiye and 10 patients from Lithuania diagnosed with Kawasaki disease. To reduce the potential impact of unequal sample sizes between cohorts, propensity score matching was performed using age and sex as matching covariates. Patients from the Lithuanian cohort were matched with patients from the Turkish cohort at a 1:3 ratio using nearest-neighbor matching. Following matching, 10 Lithuanian patients were compared with 30 matched Turkish patients.

**Results**

Fever duration at diagnosis was comparable between the groups ($p$=0.254). Irritability ($p$=0.010) and respiratory symptoms ($p$=0.027) were significantly more frequent in the Lithuanian cohort. Intensive care unit admission ($p$=0.011) and antibiotic use during hospitalization ($p$=0.006) were also significantly higher among Lithuanian patients. Although cardiac involvement was more common in the Lithuanian cohort, the difference did not reach statistical significance ($p$=0.140). No significant differences were observed regarding intravenous immunoglobulin resistance ($p$=1.000), timing of initial IVIG administration ($p$=0.067), or length of hospital stay ($p$=0.943) (see Table 1).

**Table 1 (Abstract P188)** Clinical phenotypes, coronary involvement, and risk scores in propensity score-matched Kawasaki disease cohorts from Türkiye and Lithuania

| | Türkiye ($n$=30) | Lithuania ($n$=10) | $p$-value |
|---|---|---|---|
| Atypical KD, *n* (%) | 4 (13.3) | 0 | 0.556* |
| Incomplete KD, n (%) | 18 (60.0) | 3 (30.0) | 0.148* |
| Increased coronary echogenicity | 3 (10.0) | 0 | **0.008*** |
| Coronary dilatation | 7 (23.3) | 2 (20.0) | |
| Coronary aneurysm | 0 | 2 (20.0) | |
| Coronary dilatation + aneurysm | 0 | 2 (20.0) | |
| Harada score ≥4, n (%) | 22 (73.3) | 8 (80.0) | 1.000* |
| Egami score ≥3, n (%) | 7 (23.3) | 3 (30.0) | 0.689* |
| Kawanet score ≥2, n (%) | 4 (13.3) | 3 (30.0) | 0.338* |

*Fisher's exact test

**Conclusion**

In this relatively small propensity score-matched cohort, certain clinical manifestations and cardiac involvement patterns appeared to differ between the Lithuanian and Turkish cohorts. These findings support the presence of geographic differences in the clinical spectrum of Kawasaki disease.

**Disclosure of interest**

None declared.

## P189

**Predicting gastrointestinal involvement at initial presentation: machine learning–based risk stratification in IgA vasculitis**

Ç. Yildiz[1,*], B. Küçükali[1], M. Bayar[2], K. A. Aksu[3], S. A. Bakkaloğlu[1], D. Gezgin Yıldırım[1]

[1]Pediatric Rheumatology; [2]Department of Pediatrics, Gazi University Faculty of Medicine, Ankara; [3]Boğaziçi University, İstanbul, Türkiye

**Correspondence:** Ç. Yildiz

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**Introduction**

IgA vasculitis (IgAV) is the most common childhood vasculitis, and gastrointestinal (GI) involvement is a major contributor to disease morbidity (1). Early identification of patients at risk for GI involvement at diagnosis is important for monitoring and treatment planning (2).

**Objectives**

This study aimed to evaluate the performance of machine learning–based models in predicting IgAV-associated GI involvement using clinical and laboratory parameters obtained at initial presentation.

**Methods**

A total of 618 patients under 18 years diagnosed with IgAV and followed between 2016 and 2025 were included in this retrospective study. Demographic, clinical, laboratory data, and MEFV mutation status at diagnosis were extracted as baseline variables. GI involvement was defined as abdominal pain and/or other gastrointestinal symptoms. Machine learning models (Random Forest, LightGBM, XGBoost, and CatBoost) were developed to predict GI involvement. Performance was evaluated using stratified 5-fold cross-validation with AUC for discrimination, and model interpretability was assessed using SHapley Additive Explanations (SHAP).

**Results**

The median age at diagnosis was 7.08 years (IQR: 5.47–9.60), and 54.7% of patients were male. Cutaneous involvement was universal, while joint involvement was present in 77.3%, gastrointestinal involvement in 52.9%, renal involvement in 17.8%, and testicular involvement in 8.4% of male patients. Among the machine learning models, CatBoost showed the best performance for predicting GI involvement (mean AUC: 0.657 ± 0.029; accuracy: 0.618 ± 0.037) and the most stable cross-validation results. ROC analysis confirmed its superior discriminative ability. SHAP analysis indicated that lower serum albumin, no preceding infection, higher neutrophil count, and elevated inflammatory markers were the strongest predictors of GI involvement, with MEFV mutation status also contributing to the model.

**Conclusion**
Machine learning models showed moderate discriminative performance for predicting GI involvement in IgAV using baseline clinical and laboratory data. Among them, CatBoost performed most consistently. SHAP analysis identified hypoalbuminemia and inflammatory markers as key predictors. These results suggest that machine learning may support early risk stratification in clinical practice, although prospective multicenter studies are needed to improve performance and generalizability.

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**Disclosure of interest**
None declared.

## P190

**Association of systemic inflammatory indices with renal and gastrointestinal involvement in childhood IgA vasculitis**

O. Altug Gucenmez[*,1], B. E. Kosali[1]
[1]Department of Pediatric Rheumatology, University of Health Sciences, Dr. Behçet Uz Pediatric Diseases and Surgery Training and Research Hospital, Izmir, Türkiye
**Correspondence:** O. Altug Gucenmez
*Pediatric Rheumatology 2026*, **24(S1):**P190

**Introduction**
Henoch–Schönlein purpura (HSP), currently termed IgA vasculitis, is the most common small-vessel vasculitis in childhood (1). Hematological inflammatory indices, including neutrophil-to-lymphocyte ratio (NLR), platelet-to-lymphocyte ratio (PLR), monocyte-to-lymphocyte ratio (MLR), systemic immune-inflammation index (SII), and systemic inflammation response index (SIRI), have been increasingly investigated as markers of systemic inflammation (2). In HSP, these markers have mainly been studied for their potential role in predicting renal involvement; however, the relationship between newer indices such as SII and SIRI and organ involvement remains unclear (3).
**Objectives**
To compare the distribution of SII, SIRI, NLR, PLR, and MLR parameters according to renal involvement, renal + gastrointestinal (GI) involvement, and other clinical involvement groups in patients diagnosed with HSP, and to determine whether these parameters have clinical significance.
**Methods**
This retrospective study included HSP patients with complete laboratory data. Patients were grouped according to organ involvement as renal involvement only, renal plus gastrointestinal involvement, or other non-renal involvement patterns. Inflammatory indices were calculated from complete blood count parameters. Between-group comparisons were performed using one-way ANOVA or the Kruskal–Wallis test, as appropriate, and $p<0.05$ was considered statistically significant.
**Results**
A total of 48 patients were included in the study. Of the patients, 50% were male and 50% were female, with a mean age of 6.6 years. The distribution of patients according to groups was as follows: 17 patients in the renal involvement group, 7 patients in the renal + GI involvement group, and 24 patients in the other involvement group. Statistical analysis revealed no significant differences between the groups in terms of SII, SIRI, NLR, PLR, and MLR values ($p > 0.05$) .
**Conclusion**
SII, SIRI, NLR, PLR, and MLR do not show a statistically significant difference in patients with HSP, particularly in those with renal involvement. However, SII, SIRI, and NLR values tended to be higher in the renal + GI involvement group. Due to the small sample size, the statistical power of the study is limited. These markers should be evaluated in larger cohorts with longer follow-up periods .

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**Disclosure of interest**
None declared.

## P191

**Severe childhood ANCA associated vasculitis complicated by recurrent PRES: a case report**

O. Altug Gucenmez[1,*], B. E. Kosali[1]
[1]Department of Pediatric Rheumatology, University of Health Sciences, Dr. Behçet Uz Pediatric Diseases and Surgery Training and Research Hospital, Izmir, Türkiye
**Correspondence:** O. Altug Gucenmez
*Pediatric Rheumatology 2026*, **24(S1):**P191

**Introduction**
ANCA associated vasculitis (AAV) is a rare but severe systemic autoimmune disease in childhood and often presents with nonspecific symptoms that may mimic common pediatric conditions (1).
**Objectives**
The aim of this study is to highlight the diagnostic and therapeutic challenges of a pediatric AAV case initially presenting with gastrointestinal symptoms, as well as the importance of multidisciplinary management.
**Methods**
We present an 11-year-old girl with AAV who initially presented with gastrointestinal and systemic symptoms suggestive of common pediatric conditions and subsequently developed multi-organ involvement. An 11-year-old girl presented with complaints of persistent vomiting, diarrhea, epistaxis and oliguria. On admission, she was hypertensive, pale, and dehydrated. Laboratory evaluation revealed severe acute kidney injury (creatinine 25 mg/dL, urea 196 mg/dL), metabolic acidosis, hyperkalemia, anemia (Hb 7.7 g/dL) and elevated inflammatory markers (CRP 95 mg/L, ESR 72 mm/h). Urinalysis revealed significant hematuria and proteinuria. Complement levels were within normal limits. Immunological testing demonstrated positivity for c-ANCA and PR3-ANCA. Imaging studies revealed pleural and pericardial effusions. Due to progressive renal failure and the development of anuria, urgent hemodialysis was initiated, followed by continuous renal replacement therapy. Renal biopsy demonstrated crescentic glomerulonephritis characterized by approximately 90% fibrocellular and fibrous crescents, consistent with AAV. The patient was treated with pulse corticosteroids, plasmapheresis, cyclophosphamide and subsequently rituximab. During follow-up, she developed status epilepticus secondary to posterior reversible encephalopathy syndrome (PRES) and was managed in the intensive care unit. Despite aggressive immunosuppressive therapy, the patient progressed to chronic kidney disease and was started on maintenance hemodialysis. Due to recurrent PRES episodes and ongoing seizures despite immunosuppressive treatment, she required multiple admissions to the intensive care unit. Based on the persistent systemic inflammatory findings, treatment with the IL-1 inhibitor anakinra was initiated. Following initiation of anakinra therapy, a marked reduction in seizure frequency was observed, and no further PRES episodes occurred during follow-up. The patient continues to be monitored under the current treatment regimen. This case demonstrates that AAV may present with conditions such as dehydration, gastrointestinal, or other systemic symptoms. Early recognition of systemic features such as hypertension, epistaxis and nephritic urinary findings is critical for timely diagnosis and management. Furthermore, in cases complicated by recurrent PRES episodes despite standard immunosuppressive therapy, IL-1 inhibitor therapy targeting persistent systemic inflammation may be considered a potential rescue treatment option (2).

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**Disclosure of interest**
None declared.

## P193

### Clinical profile, management and outcomes in paediatric ANCA-associated vasculitis managed in a UK tertiary service

E. Lawrence[1,*], K. Warrier[1]
[1]Paediatric Rheumatology, Nottingham University Hospitals NHS Trust, Nottingham, UK
**Correspondence:** E.Lawrence
*Pediatric Rheumatology 2026*, **24(S1):**P193

**Introduction**
Paediatric ANCA-associated vasculitis (AAV) is a rare multisystem disease with significant morbidity and risk of end organ damage requiring specialised multidisciplinary input.
**Objectives**
To describe clinically relevant phenotype-specific outcomes in children and young people with AAV managed in a tertiary paediatric rheumatology, renal and ENT service.
**Methods**
Digital health records of all children and young people with AAV managed within the service over the last 10 years were retrospectively reviewed and analysed.
**Results**
Sixteen patients were identified (9 granulomatosis with polyangiitis [GPA], 7 microscopic polyangiitis [MPA]), with a female predominance (69%). Pulmonary involvement was seen in around three quarters of the patients, which was similar across both subtypes. Renal involvement occurred in 69% overall and was more frequent and severe in MPA than GPA (86% vs. 56%), with 71% of patients with MPA requiring / awaiting renal transplantation. In contrast, ENT involvement occurred exclusively in GPA (67%), with subglottic or tracheal disease requiring airway intervention in 33% of GPA patients. Similarly, vascular complications (deep vein thrombosis, pulmonary embolism and digital ischaemia) occurred in 3 patients (19%), all with GPA. Cyclophosphamide was used for induction in the majority of patients at diagnosis (13/16), reflecting high disease severity across subtypes. We observed a surge in the diagnosis of AAV with 13 new patients between 2020 and 2026 compared to just 3 diagnosed in the same time frame between 2013 and 2019 (see Table 1).

**Table 1 (Abstract P193)** Disease burden, treatment exposure and outcomes by AAV subtype

| Feature | Overall | GPA (*n*=9) | MPA (*n*=7) |
|---|---|---|---|
| Female sex | 9 (56%) | 4 (44%) | 5 (71%) |
| Renal involvement | 11 (69%) | 5 (56%) | 6 (86%) |
| Renal transplantation / awaiting | 5 (31%) | 1 (11%) | 5 (71%) |
| Pulmonary involvement | 12 (75%) | 7 (78%) | 5 (71%) |
| ENT involvement | 6 (38%) | 6 (67%) | 0 (0%) |
| Airway procedures | 3 (19%) | 3 (33%) | 0 (0%) |
| Vascular events (DVT/PE/digital ischaemia) | 3 (19%) | 3 (33%) | 0 (0%) |
| Cyclophosphamide (anytime) | 14 (88%) | 7 (78%) | 7 (100%) |
| Rituximab induction (<12 weeks from diagnosis) | 10 (63%) | 7 (78%) | 3 (43%) |

**Conclusion**
Paediatric AAV in this tertiary cohort demonstrates clear phenotype-specific morbidity, with severe renal disease requiring transplantation predominating in MPO-associated MPA, and GPA characterised by a distinct airway and vascular complication profile. The exclusive occurrence of vascular events in GPA represents a clinically significant but under-recognised pattern. In addition, the increase in diagnoses after 2020 suggests evolving disease epidemiology, warranting further multicentre investigation and prospective study.
**Disclosure of interest**
None declared.

## P194

### Case series of children with Kawasaki disease and giant coronary artery aneurysms in Scotland, 2014-2024

E. Twynam-Perkins[1,*], M. Ilina[2], F. Pinto[3], P. Gupta[2], C. Anderson[4], N. Martin[1]
[1]Paediatric Rheumatology; [2]Paediatric Cardiology; [3]Paediatric Haematology, Royal Hospital for Children, Glasgow; [4]Paediatric Rheumatology, Royal Hospital for Children and Young People, Edinburgh, UK
**Correspondence:** E. Twynam-Perkins
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**Introduction**
Coronary artery aneurysms (CAA) are the most significant complication of Kawasaki Disease (KD). Those with giant CAA (z-score ≥ 10 or internal diameter ≥ 8 mm) (GCAA) have the highest risk of subsequent coronary events. For those with CAA at diagnosis, corticosteroids and infliximab may reduce CAA progression or promote resolution.
**Objectives**
To review the presentation, treatment and outcomes of patients with KD and GCAA across Scotland over the past decade.
**Methods**
Retrospective review of medical records of all patients diagnosed with GCAA across Scotland between 2014 and 2024.
**Results**
8 children developed GCAA, aged 2 months to 5 years at diagnosis (median 27 months). 5 were male. 4 were white, 2 Asian, 1 African and 1 mixed ethnicity.Time to diagnosis from start of symptoms ranged from 5 to 28 days (median 9 days). 4 patients had incomplete KD, of them 3 infants ≤6 months old had delayed diagnosis 20-28 days after first fever. The first echocardiogram performed 7-23 days after onset of fever showed GCAA in 3 patients, smaller CAA in 2, and abnormal features without CAA in 2. All GCAA were present on the second echocardiogram 12-31 (median 23) days from symptom onset. All patients received aspirin, intravenous immunoglobulin (IVIG) and corticosteroids. 3 were IVIG resistant. 7 received infliximab. 7 were anticoagulated, initially receiving Low Molecular Weight Heparin (LMWH) and subsequently switched to warfarin. One 4-month-old patient diagnosed in 2014 was not anticoagulated. All continue daily aspirin, 2 remain on warfarin, 1 remains on clopidogrel. Duration of follow up was 33 months to 12 years (median 54 months). One IVIG resistant boy, diagnosed aged 4, developed asymptomatic coronary artery thrombus and circumflex artery stenosis 6 months after diagnosis, whilst on LMWH. He had received infliximab. Reduction in size or resolution of CAA was documented in all patients.
**Conclusion**
3 young infants in our cohort with incomplete KD had a late diagnosis and giant CAA. All patients received standard of care and majority infliximab and anticoagulation, and in most, regression of CAA occurred. Only one patient had a significant coronary event. A recent national KD guideline will help improve time to diagnosis for infants with incomplete KD.

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**Disclosure of interest**
None declared.

P195

**Novel hematological inflammatory indices as emerging markers of disease severity and organ involvement in pediatric Henoch–Schönlein purpura**

E. Sapountzi[1,*], M. Loutsou[1], E. P. Kotanidou[1], V. R. Tsinopoulou[1], M. Stamou[1], A. Galli-Tsinopoulou[1]
[1]2nd Department of Pediatrics, School of Medicine, Faculty of Health Sciences, AHEPA University General Hospital, Aristotle University of Thessaloniki, Thessaloniki, Greece
**Correspondence:** E. Sapountzi
*Pediatric Rheumatology 2026*, **24(S1):**P195

**Introduction**
Henoch–Schönlein purpura (HSP) constitutes the most common systemic vasculitis of childhood, characterized by IgA-mediated small-vessel inflammation. The disease typically manifests with purpura, arthritis and variable gastrointestinal (GI) and/or renal involvement. Conventional inflammatory markers often demonstrate limited sensitivity, highlighting the need for novel hematological indices such as neutrophil-to-lymphocyte ratio (NLR) and systemic immune-inflammation index (SII).
**Objectives**
To assess the diagnostic and clinical utility of hematological inflammatory indices in pediatric HSP and investigate their association with GI/renal involvement.
**Methods**
This retrospective cross-sectional study included 80 participants: 40 children with HSP [boys/girls:10/30, median age 76.5 months (range:57.5–96.0)] and 40 healthy controls [boys/girls:16/24, median age 68.0 months (range:40.0–79.0)]. Baseline clinical and laboratory parameters including white blood cells (WBC), neutrophils, lymphocytes, platelets, mean platelet volume (MPV), C-reactive protein (CRP) and erythrocyte sedimentation rate (ESR) were recorded. Indices NLR, platelet-to-lymphocyte ratio (PLR), mean platelet volume-to-lymphocyte ratio (MPVLR) and platelet×neutrophil/lymphocyte count (SII) were calculated. Comparative analyses between the groups and ROC analysis for diagnostic performance were performed.
**Results**
All HSP patients presented with purpura, while GI bleeding and renal involvement were observed in 37.5% and 32.5%, respectively. Hospitalization was required in 87.5% of patients (median duration: 5 days). CRP and ESR levels did not differ significantly between groups. Compared with controls, HSP patients demonstrated statistically significant higher WBC, neutrophils ($p<0.001$), platelets ($p<0.05$), NLR and SII ($p<0.001$) levels. Lymphocytes, PLR, and MPVLR levels did not differ between patients and controls. Stratification according to GI or renal involvement revealed significant differences across all investigated indices. NLR, PLR, MPVLP and SII values were markedly increased ($p<0.001$) in patients with GI or renal involvement compared with those without organ involvement. Nevertheless, SII presented poor diagnostic performance on ROC analysis (AUC=0.5, $p=0.284$).
**Conclusion**
NLR and SII may serve as promising novel biomarkers of disease severity in pediatric HSP, particularly in relation to GI and renal involvement.
**Disclosure of interest**
None declared.

P196

**Clinical presentation of IgA vasculitis in a Mexican center**

C. E. Lopez Cardona[1], Z. I. Garcia Murillo[1], F. A. Vasquez Triminio[1,*], F. Garcia Rodriguez[1], A. V. Villarreal Treviño[1], R. A. Falcon Zuñiga[1], N. E. Rubio Pérez[1]
[1]Pediatrics, Hospital Universitario "Dr. José Eleuterio González", Monterrey, Mexico
**Correspondence:** F. A. Vasquez Triminio
*Pediatric Rheumatology 2026*, **24(S1):**P196

**Introduction**
IgA vasculitis (IgAV) is a small-vessel vasculitis marked by IgA-containing immune complexes in vessel walls, most common in children but often more severe in adults.
**Objectives**
To describe the clinical manifestations of IgAV in a Mexican tertiary medical center.

**Table 1 (Abstract P196)** Temporal distribution of clinical manifestation events

| Time from symptom onset | Cutaneous manifestations *n* (%) | Gastrointestinal manifestations *n* (%) | Arthritis *n* (%) | Renal manifestations *n* (%) |
|---|---|---|---|---|
| **Day 1** | 19 (70.4%) | 6 (33.3%) | 10 (62.5%) | 0 (0%) |
| **Days 2–7** | 6 (22.2%) | 3 (16.7%) | 4 (25%) | 0 (0%) |
| **Days 8–14** | 1 (3.7%) | 4 (22.2%) | 0 (0%) | 0 (0%) |
| **Days 15–30** | 1 (3.7%) | 2 (11.1%) | 2 (12.5%) | 1 (5.3%) |
| **Days 31–90** | 0 (0%) | 3 (16.7%) | 0 (0%) | 2 (10.5%) |
| **>90 days** | 0 (0%) | 0 (0%) | 0 (0%) | 16 (84.2%) |
| **Total Events** | 27(100%) | 18 (100%) | 16 (100%) | 19 (100%) |

*Some events occurred more than once

**Conclusion**: The lack of understanding of pediatric rheumatology diseases within the medical community may contribute to delays in diagnosis and inappropriate management, consequently obstructing the clinical resolution of these medical conditions. This report describes the clinical presentation of these patients in our country to improve the understanding of this disease

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**Methods**
A retrospective longitudinal descriptive study was conducted at a Mexican tertiary medical center. Medical records of patients with IgAV diagnosed between 2019 and 2026 were reviewed.
**Results**
A total of 25 children were evaluated, with a mean age of 7.8 ± 3.84 years. 14 (56%) of them were female. The prevalence of the symptoms was cutaneous (100%), arthritis (64%), abdominal involvement (52%), and renal manifestations (28%). As for the cutaneous clinical manifestations, 14 (56%) had an atypical presentation with a greater presence in both lower (25, 100%) and upper limbs (12, 48%). Facial and genital involvement were the least frequent manifestations, with only 3 cases (12%). All children presented cutaneous symptoms within the first 10 days of disease onset. Only 5 (20%) children required a skin biopsy, revealing granular IgA deposition within vessel walls. Arthritis developed in 16 (64%) children at some point, predominantly affecting the lower limbs in 11 cases (44%). Articular manifestations mainly occurred during the initial weeks of disease evolution, being the first symptom in 10 (40%) of the cases. As for the renal affection, the most common symptom was microscopic hematuria (7, 28%). Most of the manifestations appeared after several months of disease onset, with the earliest renal involvement documented on day 26 of follow-up. The temporal distribution of clinical manifestations is summarized in Table 1. The mean delay from symptom onset to diagnosis was 20.89 ± 16.5 days. Before diagnosis, patients attended between 1 and 5 medical consultations and frequently received treatment for alternative diagnoses, including viral exanthema, insect bite reactions, and infectious processes associated with thrombocytopenia. 5 children (20%) required hospitalization.
**Disclosure of interest**
None declared.

P197

**Beyond histology: outcome predictors in pediatric IgAV nephropathy with nephrotic-range proteinuria**

G. Inguscio[1,2,*], C. S. Barberis[1], C. Errichiello[3], I. Pagnini[2], I. Maccora[2], M. V. Mastrolia[2], L. Gatti[2], E. Marrani[2], S. Romano[1,2], L. Oni[4,5], A. Vaglio[3,6], G. Simonini[1,2], T. Giani[2]
[1]Paediatric Department, University of Florence; [2]Rheumatology Unit, AOU Meyer Children's Hospital IRCCS; [3]Nephrology and Dialysis Unit, AOU Meyer Children's Hospital IRCCS, Florence, Italy; [4]Department of Paediatric Nephrology, Alder Hey Children's NIHR Foundation Trust; [5]Department of Women's and Children's Health, University of Liverpool, Liverpool, UK;

[6]Department of Biomedical, Experimental and Clinical Sciences "Mario Serio", Florence, Italy
**Correspondence:** G. Inguscio
*Pediatric Rheumatology 2026*, **24(S1):**P197

**Introduction**
Nephrotic-range proteinuria (NRP) identifies patients at increased risk of severe renal disease and long-term renal damage in IgA vasculitis (IgAV), the most common pediatric systemic vasculitis. However, paediatric data remain limited.
**Objectives**
To investigate clinical features, histopathology, treatment, and outcomes of IgA vasculitis nephropathy (IgAVN) with NRP in two tertiary centers.
**Methods**
A retrospective study (2015–2026) including paediatric patients with IgAVN-associated NRP. IgAV was defined according to EULAR/PRINTO/PRES criteria; NRP as urine protein/creatinine ratio>2 mg/mg, albumin/creatinine ratio>200 mg/mmol, or proteinuria≥50 mg/kg/24 h. Renal outcomes at last follow-up were classified as: A, normal; B, minor abnormalities; C, active disease; D, chronic kidney disease (CKD).
**Results**
In 399 IgAV patients, 48 (12%) developed NRP (median age 7 years; 58% male). Renal biopsies ($n$=34) showed mesangial hypercellularity (76%), endocapillary proliferation (74%), and crescents (50%). In 18 patients with available ISKDC classification, class III predominated (72%). Histopathological findings did not correlate with proteinuria resolution or renal outcome. Treatments included corticosteroids (89%), ACE inhibitors (64%), and second-line immunosuppressive therapies. Over a median follow-up of 30 months, renal outcomes were A in 55% of patients, B in 27%, and C in 16%, one patient progressed to CKD (4 patients lost to follow-up). Complete proteinuria resolution occurred in 60% of cases. Persistent active renal disease was significantly associated with prolonged purpura (>1 month) and joint involvement during follow-up. Significant differences in functional renal markers were observed across outcome groups and proteinuria resolution status at different timepoints (Table 1). Persistent proteinuria was associated with higher ESR and C4, and lower IgG at baseline ($p$<0.05).
**Conclusion**
NRP identifies a severe and heterogeneous subset of paediatric IgAVN characterized by frequent persistent renal activity, prolonged proteinuria, and substantial need for immunosuppressive therapy. Conversely, histopathological findings did not correlate with renal outcome or proteinuria resolution. Our findings suggest that longitudinal clinical and laboratory evolution better predict renal prognosis than baseline histology and support prolonged monitoring in pediatric IgAVN with NRP.

**Disclosure of interest**
None declared.

## P198

### BCG scar changes are associated with a Kawasaki-like phenotype in children with MIS-C: a multicenter Latin American study of the REKAMLATINA network

H. Menchaca-Aguayo[1,*], A. V. Villarreal-Treviño[2], F. García-Rodríguez[2], J. García-Silva[3], G. Ivankovich-Escoto[4], A. H. Tremoulet[5], R. Ulloa-Gutiérrez[6], E. Faugier Fuentes[7], T. REKAMLATINA-3 MIS-C Study Group Investigators[8]
[1]Reumatología Pediátrica, Hospital General Agustín O'Horán, Mérida; [2]Reumatología Pediátrica, Hospital Universitario Dr. José Eleuterio Gonzalez, Monterrey; [3]Pediatría, Insituto Nacional de Pediatría, Ciudad de México, Mexico; [4]Pediatría, Hospital Nacional de Niños "Dr. Carlos Sáenz Herrera", Caja Costarricense de Seguro Social (CCSS), San José, Costa Rica; [5]Pediatrics, University of California San Diego, San Diego, USA; [6]Pediatría, Hospital Nacional de Niños "Dr. Carlos Sáenz Herrera", San José, Costa Rica; [7]Reumatología Pediátrica, Hospital Infantil de México Federico Gómez; [8]Pediatrics, REKAMLATINA-3, Ciudad de México, Mexico
**Correspondence:** H. Menchaca-Aguayo
*Pediatric Rheumatology 2026*, **24(S1):**P198

**Introduction**
Inflammatory changes at the Bacillus Calmette-Guérin (BCG) scar site are well recognized in Kawasaki disease, but their significance in Multisystem Inflammatory Syndrome in Children (MIS-C) remains unclear.
**Objectives**
We aimed to evaluate the frequency of inflammatory BCG scar changes and their association with clinical phenotype, inflammatory profile, and cardiac involvement in MIS-C.
**Methods**
A multicenter observational study with retrospective and prospective data collection was conducted using the REKAMLATINA Network registry. Patients fulfilling MIS-C criteria with visible BCG scars were included and classified according to the presence or absence of inflammatory BCG scar changes. Demographic, clinical, laboratory, echocardiographic, treatment, and outcome variables were compared. Multivariable logistic regression analyses were performed to identify factors independently associated with Kawasaki-like phenotype classification and coronary abnormalities at admission.
**Results**
Among 697 MIS-C patients with visible BCG scars, 61 (8.7%) presented inflammatory scar changes. These patients were younger and more frequently exhibited Kawasaki-like mucocutaneous manifestations, including conjunctivitis, mucositis, extremity changes, and perianal rash,

**Table 1 (Abstract P197)** Clinical features and renal markers by outcome/proteinuria resolution.

| Variable | A *N*=24 | B *N*=12 | C *N*=7 | D *N*=1 | *p* | Proteinuria Persistence *N*=19 | Proteinuria Resolution *N*=29 | *p* |
|---|---|---|---|---|---|---|---|---|
| Joint involvement | 20 (83%) | 7 (58%) | 7 (100%) | 0 (0%) | **0.033** | | | |
| Purpura > 1 m | 8 (62%) | 0 (0%) | 4 (80%) | 0 (0%) | **0.022** | | | |
| Cr 3 m | 39 (35-48) | 35 (30-38) | 69 (60-74) | 73 (73-73) | **0.006** | | | |
| Alb 3 m | 4.00 (3.60-4.30) | 3.40 (3.20-3.92) | 3.20 (3.20-3.60) | 2.88 (2.88-2.88) | **0.012** | 3.40 (3.20-3.90) | 3.95 (3.60-4.30) | **0.035** |
| eGFR 3 m | 102 (92-116) | 115 (80-119) | 77 (64-82) | 71 (71-71) | **0.015** | 81 (69-105) | 102 (89-116) | **0.033** |
| Alb 6 m | 4.20 (4.10-4.50) | 4.04 (3.80-4.50) | 3.55 (3.25-3.85) | 3.45 (3.45-3.45) | **0.029** | 3.80 (3.30-3.90) | 4.20 (4.10-4.50) | **0.014** |
| eGFR 6 m | 104 (99-122) | 94 (85-126) | 80 (80-89) | 72 (72-72) | **0.012** | 80 (79-93) | 103 (95-123) | **0.002** |
| Alb 12 m | 4.40 (4.10-4.50) | 3.98 (3.35-4.28) | 3.60 (3.40-3.90) | 3.58 (3.58-3.58) | **0.014** | 3.60 (3.40-3.90) | 4.40 (4.10-4.50) | **<0.001** |
| Cr 12 m | | | | | | 46 (40-67) | 39 (31-43) | **0.029** |

Median (Q1–Q3); Creatinine (Cr) µmol/L; Albumin (Alb) g/dL; eGFR mL/min/1.73 m$^2$. p values: Kruskal–Wallis/Wilcoxon and $\chi^2$/Fisher's tests

whereas shock and gastrointestinal involvement were less frequent. They also showed lower inflammatory markers, including C-reactive protein, procalcitonin, and ferritin, together with higher platelet and lymphocyte counts, suggesting a less severe systemic inflammatory profile. In multivariable analysis, younger age and mucocutaneous manifestations were independently associated with Kawasaki-like phenotype classification, while BCG scar changes showed a marginal association (OR 1.97; 95% CI 0.99–3.35; $p = 0.053$). Coronary dilation at admission was more frequent in patients with BCG scar changes (23% vs. 12.1%; $p = 0.026$), although this association was not maintained after adjustment.

**Conclusion**

Inflammatory BCG scar changes identify a distinct Kawasaki-like phenotype within MIS-C characterized by younger age, predominant mucocutaneous involvement, lower systemic inflammatory burden, and increased frequency of coronary dilation at presentation. BCG scar evaluation may represent a useful clinical marker for early recognition of Kawasaki-like MIS-C phenotypes.

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**Disclosure of interest**

None declared.

## P199

**ANCA-negative destructive sinonasal granulomatosis with polyangiitis mimicking invasive fungal rhinosinusitis in a child**

H. Menchaca-Aguayo[1,*], A. González Reynoso[2], N. Negroe Ocampo[3], C. Valerio Carballo[4]
[1]Pediatric Rheumatology; [2]Pediatric Infectious Diseases; [3]Pediatric Oncology; [4]Pediatric Cardiology, Hospital General Agustín O'Horán, Mérida, Mexico
**Correspondence:** H. Menchaca-Aguayo
*Pediatric Rheumatology 2026*, **24(S1):**P199

**Introduction**

Granulomatosis with polyangiitis (GPA) is a rare pediatric small-vessel vasculitis that may present with localized upper airway involvement. Destructive sinonasal disease can mimic invasive fungal infection or malignancy, particularly in ANCA-negative patients, leading to diagnostic delay.

**Objectives**

To describe the clinical, radiologic, and histopathologic features of a pediatric patient with ANCA-negative destructive sinonasal GPA initially suspected as invasive fungal rhinosinusitis.

**Methods: Results**

The patient developed progressive right periorbital and hemifacial edema associated with dacryocystitis and a painful palatal ulcer. Initial computed tomography demonstrated heterogeneous occupation of the right maxillary sinus with bone lysis and extension to the ipsilateral nasolacrimal duct, raising suspicion for invasive fungal rhinosinusitis or neoplastic disease. Despite broad-spectrum antimicrobial and antifungal therapy, disease progression persisted. Magnetic resonance imaging later demonstrated diffuse inflammatory occupation of the right maxillary, ethmoidal, and sphenoidal sinuses with focal bone erosion, mucosal necrosis, and extension into adjacent soft tissues. High-resolution chest computed tomography additionally demonstrated focal areas of air trapping suggestive of small airway involvement despite the absence of significant respiratory symptoms. Histopathologic examination of maxillary sinus and palatal biopsies revealed necrotizing granulomatous small-vessel vasculitis with fibrinoid necrosis, prominent leukocytoclasia, eosinophil-rich inflammation, and granuloma formation, without evidence of pathogenic microorganisms on special stains. ANCA testing was negative. Based on the clinical, radiologic, and histopathologic findings, a diagnosis of ANCA-negative granulomatosis with polyangiitis was established.

**Conclusion**

Pediatric GPA may present as destructive unilateral sinonasal disease closely mimicking invasive fungal rhinosinusitis. In children with progressive sinonasal destruction, palatal ulceration, and negative microbiologic studies, GPA should be considered even in the absence of ANCA positivity. Early biopsy and multidisciplinary evaluation are critical to establish the diagnosis and avoid delays in immunosuppressive treatment.

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**Disclosure of interest**

None declared.

## P200

**Beyond traditional clinical scores: 3D markerless kinematics as a digital biomarker in juvenile idiopathic arthritis**

A. Yekdaneh[1,*], S. Karamollaoğlu[2], B. Yumak[3,4], I. Donmez[2], A. Albayrak[3,5], N. Aktay Ayaz[6], N. Arman[7]
[1]Vocational School of Health Services Physiotherapy English Program, Fenerbahçe University; [2]Institute of Graduate Studies Physiotherapy and Rehabilitation Master Program; [3]Institute of Graduate Studies Physiotherapy and Rehabilitation Doctorate Program, Istanbul University-Cerrahpaşa; [4]Faculty of Health Sciences, Physiotherapy and Rehabilitation (English) Program, Istanbul Atlas University; [5]Faculty of Health Sciences Department of Physiotherapy and Rehabilitation English, Istanbul Kent University; [6]Istanbul Faculty of Medicine, Department of Pediatric Rheumatology, Istanbul University; [7]Faculty of Health Sciences Department of Physiotherapy and Rehabilitation, Istanbul University-Cerrahpaşa, Istanbul, Türkiye
**Correspondence:** A. Yekdaneh
*Pediatric Rheumatology 2026*, **24(S1):**P200

**Introduction**

In Juvenile Idiopathic Arthritis (JIA), monitoring functional mobility is essential for understanding disease-related limitations and guiding rehabilitation. Standard clinical assessments, such as the 30-Second Sit-to-Stand Test (30STS) and 6-Minute Walk Test (6MWT), are widely used indicators of functional capacity. However, these tests may not fully capture the underlying movement strategies and joint-level compensations contributing to functional limitations.

**Objectives**

This study aimed to use smartphone-based three-dimensional markerless motion capture via OpenCap to evaluate lower-extremity kinematics during squat and sit-to-stand tasks in children and adolescents with JIA. The objective was to identify task-related biomechanical asymmetries and examine their associations with clinical joint involvement and functional capacity.

**Methods**

Eighteen pediatric patients with JIA, with a mean age of 14.28±2.27 years, were prospectively assessed. Three-dimensional kinematics during squat and sit-to-stand transitions were captured using the OpenCap platform. Clinical and functional data included lower-limb joint count, 30STS repetitions, and 6MWT distance. Pearson correlation analysis was used to examine associations between clinical joint involvement, lower-extremity symmetry indices, and functional capacity outcomes.

**Results**

Kinematic analysis showed that the sit-to-stand task produced a significantly higher Ankle Symmetry Index than the squat task, with values of 17.24% and 10.07%, respectively ($p$=0.015). This suggests that the sit-to-stand transition may be more sensitive for detecting ankle-related asymmetry in this sample. A higher lower-limb joint count was positively correlated with increased sit-to-stand Ankle Symmetry Index ($r$=0.49). Ankle asymmetry during sit-to-stand also showed moderate

negative correlations with both 6MWT distance ($r$=-0.54) and 30STS performance ($r$=-0.51), which were stronger than the associations observed for clinical joint count.

**Conclusion**

Smartphone-based three-dimensional kinematic analysis using OpenCap may provide clinically relevant insights into lower-extremity movement asymmetries in pediatric patients with JIA. The sit-to-stand transition appeared to reveal greater ankle asymmetry than the squat task, suggesting that it may be a useful functional movement for identifying distal biomechanical deficits. The associations between ankle asymmetry and both walking capacity and sit-to-stand performance indicate that ankle-related movement asymmetry may contribute to functional limitations in JIA. These findings support the potential value of integrating accessible markerless motion capture with standard clinical tests to guide individualized rehabilitation strategies targeting biomechanical symmetry and functional mobility.

**Disclosure of interest**

None declared.

## P201

### Gait adaptations and movement constraints in adolescents with familial Mediterranean fever: a wearable motion capture study

A. Albayrak[1,2,*], S. Ozbal[3], A. Yekdaneh[4], I. Donmez[5], N. Aktay Ayaz[6], H. Argunsah[7], N. Arman[8]

[1]Istanbul University-Cerrahpasa, Institute of Graduate Studies, Physiotherapy and Rehabilitation Doctorate Program; [2]Istanbul Kent University, Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation; [3]Acıbadem Mehmet Ali Aydınlar University, Graduate School of Natural and Applied Sciences, Biomedical Engineering Doctorate Program; [4]Fenerbahçe University, Vocational School of Health Services, Physiotherapy English Program; [5]Private Health Services Unit, Physiotherapist Irem Donmez; [6]Istanbul University, Istanbul Faculty of Medicine, Department of Pediatric Rheumatology; [7]Acıbadem Mehmet Ali Aydınlar University, Faculty of Engineering and Natural Sciences, Department of Biomedical Engineering; [8]Istanbul University-Cerrahpasa, Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Istanbul, Türkiye

**Correspondence:** A. Albayrak

*Pediatric Rheumatology 2026*, **24(S1):**P201

**Introduction**

In adolescents with Familial Mediterranean Fever (AwFMF), symptoms like joint pain, swelling, fatigue and reduced physical activity can negatively impact gait mechanics and overall locomotor function.

**Objectives**

This study aimed to compare spatiotemporal and mean kinematic gait parameters between aFMF and healthy controls using a wearable motion capture system.

**Methods**

Fifteen AwFMF, aged 12–18 years and 20 healthy controls were included in this cross-sectional study. Three-dimensional gait analysis was performed using the Xsens MVN Awinda system during self-selected overground walking. The healthy control group was obtained from the publicly available COMPWALK-ACL dataset, which includes biomechanical gait data from typically developing children without reported neurological, musculoskeletal or orthopedic conditions affecting gait. Spatiotemporal gait parameters, including stride time, cadence, stance and swing characteristics, step time and asymmetry were analyzed. Mean kinematic parameters included lower extremity joint motion, pelvic and trunk movements, center of mass displacement and shoulder motion during walking.

**Results**

The mean ages of AwFMF and healthy controls were 13.1±1.3 and 15.7±0.7 years, respectively.AwFMF showed significantly longer temporal gait parameters and lower cadence compared with healthy controls, including longer stride time (1.15±0.12; 0.96±0.05 s), prolonged stance time (0.69±0.09; 0.56±0.06 s), increased step time (0.59±0.07; 0.49±0.03 s) and lower cadence (105.2±12.41; 125.26±7.1 steps/min) ($p$<0.001).Kinematic analysis revealed significantly reduced movement amplitudes in lower extremity joints, including lower hip flexion–extension range (38.44±6.18°; 51.67±4.07°), knee flexion–extension range (59.27±6.15°; 68.85±3.64°) and ankle dorsiflexion–plantarflexion range (29.67±6.77°; 40.34±2.92°) ($p$<0.001). AwFMF also demonstrated significantly reduced pelvic tilt, pelvic rotation, trunk tilt, and shoulder flexion–extension amplitudes ($p$<0.001). In addition, anterior–posterior and vertical center of mass displacement values were significantly lower in the AwFMF group than in controls ($p$<0.001).

**Conclusion**

AwFMF exhibited distinct spatiotemporal and kinematic gait alterations, characterized by reduced lower extremity and trunk movement, decreased center of mass displacement and a slower walking pattern. These findings may suggest a movement-constrained or energy-conserving gait strategy in AwFMF.Wearable motion capture systems may provide physiotherapists with clinically meaningful information to identify subtle functional adaptations and guide individualized exercise, gait training, and physical activity interventions.However, due to the significant age difference between groups, age was included as a covariate and the observed gait differences should be interpreted with caution.

**Disclosure of interest**

None declared.

## P202

### Determinants of functional capacity in children and adolescents with Familial Mediterranean Fever: potential indicators of neuromuscular control

A. Albayrak[1,2,*], I. Donmez[3], A. Namli Seker[1,4], O. Akgun[5], N. Aktay Ayaz[5], N. Arman[6]

[1]Istanbul University-Cerrahpasa, Institute of Graduate Studies, Physiotherapy and Rehabilitation Doctorate Program; [2]Istanbul Kent University, Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation; [3]Private Health Services Unit, Physiotherapist Irem Donmez; [4]Bahcesehir University, Vocational School of Health Services, Physiotherapy Program; [5]Istanbul University, Istanbul Faculty of Medicine, Department of Pediatric Rheumatology; [6]Istanbul University-Cerrahpasa, Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Istanbul, Türkiye

**Correspondence:** A. Albayrak

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**Introduction**

Symptoms experienced by children with Familial Mediterranean Fever (cFMF) may limit participation in activities of daily living and reduce functional capacity.Therefore, identifying modifiable factors associated with functional capacity is clinically important.

**Objectives**

This study aimed to identify the determinants of functional capacity in cFMF and to examine its relationship with physical fitness, balance, muscle strength, and gait.

**Methods**

Twenty-six cFMF, comprising 11 females and 15 males, aged 10–17 years, were included in the study.Functional capacity was assessed using the 6-Minute Walk Test (6MWT), 30-Second Sit-to-Stand Test (30SSTS), and 10-Step Stair Climb Test (10SCT).Physical fitness was evaluated using the Curl-up, Push-up, Trunk Lift and Sit-and-Reach Tests. Balance was assessed using landing tests on a Kinvent KForce Plate.Gait was evaluated using the Digitsole Pro smart insole system and lower-extremity muscle strength was measured isometrically at 30°, 60° and 90° knee extension angles using a KPull dynamometer.Pearson correlation analysis was used to examine bivariate associations and multiple linear regression was used to identify independent determinants of functional capacity outcomes.

**Results**

The mean age of cFMF was 13.69±1.56 years.Correlation analysis revealed significant positive correlations between 6MWT, which was 624.81±59.35 m, and Curl-up Test ($r$=0.565, $p$=0.003), 30SSTS ($r$=0.445, $p$=0.023), and gait speed ($r$=0.473, $p$=0.015).The 30SSTS score, which was 14.92±3.15 repetitions, showed significant positive correlations with the Curl-up Test ($r$=0.410, $p$=0.034) and the 60° knee extension fatigue index ($r$=0.498, $p$=0.011), and a significant negative correlation with 10SCT duration ($r$=-0.561, $p$=0.004). In regression analyses, gait speed was the strongest determinant of 6MWT distance, explaining 49.9% of the variance ($p$=0.047).Curl-up Test explained 36.7% of the variance in 30SSTS ($p$=0.035), indicating core strength as an important determinant of sit-to-stand performance. In the 10SCT model ($p$=0.0063), 60° knee extensor fatigue was the strongest determinant

($p$=0.003), with greater fatigue linked to longer stair-climbing duration, while better 30SSTS performance was associated with shorter duration ($p$=0.041).

**Conclusion**

Functional capacity in cFMF was significantly associated with physical fitness, gait speed, muscle strength and endurance.Gait speed, abdominal muscle strength, and 60° knee extensor fatigue emerged as the primary determinants of 6MWT, 30SSTS and 10SCT respectively.These findings suggest that gait efficiency, core strength, and lower-extremity muscle endurance may play important roles in functional capacity in cFMF. Rehabilitation programs for cFMF may therefore benefit from including exercises targeting core stabilization, lower-extremity strength and endurance, balance, proprioception, agility and speed.

**Disclosure of interest**

None declared.

## P203

### Comparison of agility, balance, reaction time and attention between children with juvenile idiopathic arthritis and healthy peer

B. Yaşar[1,*], S. Aktay[1], A. Meral[1], A. Altun[1], A. Sarı[1], F. İlhan[1], A. Yekdaneh[2], A. Albayrak[3], I. Dönmez[4], F. Cakmak[5], N. Aktay Ayaz[6], N. Arman[1]
[1]Department of Physiotherapy and Rehabilitation, Istanbul University-Cerrahpasa; [2]Vocational School of Health Services, Fenerbahçe University; [3]Department of Physiotherapy and Rehabilitation, Istanbul Kent University; [4]Physiotherapist Irem Donmez, Private Health Services Unit; [5]Pediatric Rheumatology Department, Basaksehir Cam and Sakura City Hospital; [6]Department of Pediatric Rheumatology, Istanbul University, Istanbul, Türkiye
**Correspondence:** B. Yaşar
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**Introduction**

Juvenile Idiopathic Arthritis (JIA) may impair physical performance, neuromuscular control, balance and coordination in children.

**Objectives**

This study aimed to investigate agility, dynamic balance, reaction time and attention in children with JIA compared with healthy peers.

**Methods**

Children with JIA and age- and sex-matched healthy controls were included in this study. Agility was assessed using the Agility Ladder Test with single-leg, double-leg and lateral jumping tasks. Reaction time and attention were evaluated using the CatchPad Interactive Platform. Dynamic balance was assessed with the Y-Balance Test. Group comparisons were performed using independent samples analyses, and associations were examined using Pearson Correlation Analysis.

**Results**

The JIA and healthy control groups were comparable for age, body weight, height and BMI($p$>0.05). Compared with healthy controls, children with JIA showed significantly poorer performance in selected agility and dynamic balance parameters. Single-leg and lateral Agility Ladder performances were significantly different between groups ($p$<0.05). Total Y-Balance and posterolateral balance scores were lower in the JIA group than in healthy controls ($p$<0.05). Reaction time outcomes also differed between groups, with significantly poorer worst and total reaction time performances in children with JIA ($p$<0.05). Attention-related CatchPad scores were generally lower in the JIA group, although most attention parameters did not reach statistical significance. Within the JIA group, lateral Agility Ladder performance was significantly correlated with CatchPad attention scores ($r$=-0.694, $p$<0.001). Single-leg agility was also significantly associated with posterolateral Y-Balance performance ($r$=-0.614, $p$<0.001).

**Conclusion**

Children with JIA demonstrated impairments in agility, dynamic balance and reaction time compared with healthy peers. Agility performance was associated with attention, reaction time and balance outcomes, suggesting that functional limitations in JIA may involve both motor and cognitive-motor components. Physiotherapy and Rehabilitation programs for JIA should include agility, dynamic balance, reaction-time and attention-demanding motor exercises.

**Disclosure of interest**

None declared.

## P205

### Comparison of the effects of synchronous and interactive asynchronous telerehabilitation on fatigue and functional capacity in adolescents with Familial Mediterranean Fever: a pilot study

A. Namli Seker[1,2,*], N. Aktay Ayaz[3], N. Arman[4]
[1]Istanbul University-Cerrahpasa, Institute of Graduate Studies, Physiotherapy and Rehabilitation Doctorate Program; [2]Bahçeşehir University, Vocational School of Health Services, Physiotherapy Program; [3]Istanbul University, Istanbul Faculty of Medicine, Department of Pediatric Rheumatology; [4]Istanbul University-Cerrahpasa, Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Istanbul, Türkiye
**Correspondence:** A. Namli Seker
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**Introduction**

Familial Mediterranean Fever (FMF) symptoms often lead to fatigue and impaired functional capacity in pediatric patients.Although telerehabilitation approaches, including supervised online and asynchronous exercise programs, have shown beneficial effects in other pediatric rheumatic diseases, evidence regarding different telerehabilitation delivery models in adolescents with FMF remains limited.

**Objectives**

This study aimed to compare the effects of synchronous and asynchronous telerehabilitation-based exercise delivery models on fatigue, pain, functional capacity, physical fitness, gait parameters and balance in adolescents with FMF.

**Methods**

Sixty-eight adolescents with FMF (12–18 years) were randomized to either a physiotherapist-supervised synchronous telerehabilitation group ($n$=42) or an asynchronous telerehabilitation group delivered via the Edpuzzle platform ($n$=26).Both groups completed exercise training twice weekly for 8 weeks.In the asynchronous group, Edpuzzle provided structured exercise videos with interactive features to support engagement and follow-up.Functional capacity was assessed using the 6-Minute Walk Test(6MWT), Stair Climb Test, Half-Squat Test, and Sit-to-Stand Test.Pain and fatigue were evaluated with the Visual Analog Scale(VAS) and physical fitness with the FitnessGram Test Battery.Balance was assessed using the Single-Leg Stance Test and Kinvent system, and gait parameters using DigitsolePro smart insoles.Assessments were conducted at baseline and post-intervention.

**Results**

The synchronous and asynchronous groups had comparable mean ages (14.19±1.77 vs. 14.30±2.20 years).Due to dropouts in both the synchronous (19/42) and asynchronous (15/26) groups, an intention-to-treat analysis was conducted using five multiple imputations.Both groups showed significant within-group improvements in nearly all outcomes ($p$<0.001), except gait symmetry and double-leg stance postural sway with eyes closed.Between-group comparisons significantly favored the synchronous group for VASpain, VASfatigue, 6MWT, and double-leg stance stability under both eyes-open and eyes-closed conditions, with p-values ranging from<0.001 to 0.02.

**Conclusion**

Both synchronous and interactive asynchronous telerehabilitation delivery models improved functional capacity, fatigue, pain, physical fitness, gait parameters, and balance in adolescents with FMF.However, supervised synchronous telerehabilitation yielded superior improvements in functional outcomes compared to asynchronous methods.Pilot findings suggest that telerehabilitation is feasible and beneficial for adolescents with FMF, with synchronous delivery resulting in greater improvements.

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**Trial registration identifying number**

NCT06743152.

**Disclosure of interest**

None declared.

## P206

### Feasibility and effects of a smart exergame balance board-based exercise program in children with juvenile idiopathic arthritis

I. Dönmez[1,*], A. Albayrak[2,3], A. Yekdaneh[4], F. Cakmak[5], N. Aktay Ayaz[6], N. Arman[7]

[1]Physiotherapist Irem Donmez, Private Health Services Unit,; [2]Institute of Graduate Studies, Physiotherapy and Rehabilitation Doctorate Program, Istanbul University-Cerrahpasa; [3]Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Istanbul Kent University; [4]Vocational School of Health Services Physiotherapy English Program, Fenerbahce University; [5]Pediatric Rheumatology Department, Basaksehir Cam and Sakura City Hospital; [6]Department of Pediatric Rheumatology, Istanbul University, Istanbul Faculty of Medicine; [7]Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Istanbul University-Cerrahpasa, Istanbul, Türkiye

**Correspondence:** I. Dönmez

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**Introduction**

Juvenile idiopathic arthritis (JIA) severely impacts children's physical fitness and balance. Despite the known benefits of exercise, evidence regarding technology-supported, game-based balance interventions in this population is still limited.

**Objectives**

This study aimed to investigate the feasibility and effects of a novel smart exergame balance board program on physical fitness, muscle strength, flexibility, functional capacity, postural stability, and patient-reported experiences in children with JIA.

**Methods**

Twenty-four children with JIA completed a 12-week pilot program (twice weekly) using a novel, team-developed smart exergame balance board (SEG-Board) featuring real-time biofeedback and gamification. Assessments evaluated physical fitness and functional performance via the 30-Second Sit-to-Stand Test (30STS), 6-Minute Walk Test (6MWT), muscle strength, flexibility, and stair performance. Postural stability was measured using Kinvent Physio. Post-intervention satisfaction was explored via a semi-structured survey and thematic analysis.

**Results**

Significant improvements were observed in 30STS performance ($p<0.001$) and 6MWT distance ($p<0.05$). Muscle strength, flexibility, and stair performance also improved significantly after the intervention ($p<0.05$). Postural stability improved, with total sway surface area decreasing from 590.57±654.70 to 287.08±131.30 mm² ($p<0.05$). The program showed high feasibility and acceptability, with a mean satisfaction score of 9.5±0.5/10. Thematic analysis identified five main themes: functional muscle strength, posture and mobility, endurance, pain control, and psychosocial well-being. Participants reported being able to kick a ball more effectively, climb stairs or slopes more easily, maintain better posture, walk without dragging their feet, feel less tired, experience reduced localized pain, and feel more confident and willing to participate in play with peers.

**Conclusion**

The SEG-Board-based exercise program was feasible, highly acceptable, and associated with improvements in functional capacity, muscle strength, flexibility, stair performance, and postural stability in children with JIA. By integrating balance-based movement tasks with real-time biofeedback and gamification, this technology-supported approach may enhance engagement while supporting functional and balance-related gains. Qualitative findings further suggest potential benefits for motivation, confidence, pain perception, and social participation. Although these preliminary findings require confirmation in larger randomized controlled trials, the SEG-Board-based smart exergame approach appears to be a promising, safe, and motivating tool for pediatric rheumatology rehabilitation.

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**Disclosure of interest**

None declared.

## P207

### OpenCap-based markerless gait analysis in juvenile idiopathic arthritis: associations between pelvic motion and functional capacity

S. Karamollaoğlu[1,*], A. Yekdaneh[2], B. N. Yumak[3,4], I. Donmez[5], N. A. Ayaz[6], N. Arman[7]

[1]Institute of Graduate Studies Physiotherapy and Rehabilitation Master Program, Istanbul University-Cerrahpasa; [2]Vocational School of Health Services Physiotherapy English Program, Fenerbahçe University; [3]Institute of Graduate Studies Physiotherapy and Rehabilitation Doctorate Program, Istanbul University-Cerrahpasa; [4]Faculty of Health Sciences, Physiotherapy and Rehabilitation (English) Program, Istanbul Atlas University; [5]Physiotherapist Irem Donmez, Private Health Services Unit; [6]Istanbul Faculty of Medicine, Department of Pediatric Rheumatology, Istanbul University; [7]Faculty of Health Sciences Department of Physiotherapy and Rehabilitation, Istanbul University-Cerrahpasa, İstanbul, Türkiye

**Correspondence:** S. Karamollaoğlu

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**Introduction**

Gait abnormalities may contribute to functional limitations in juvenile idiopathic arthritis (JIA), while the 6-minute walk test (6MWT) provides a valid estimate of functional capacity. OpenCap is a smartphone-based markerless motion capture platform that enables accessible three-dimensional movement analysis; however, its potential clinical application in pediatric rheumatology remains insufficiently explored.

**Objectives**

This study aimed to explore the feasibility and clinical relevance of OpenCap-based gait assessment in children and adolescents with JIA and to examine the associations between OpenCap-derived gait parameters and functional capacity measured by the 6MWT.

**Methods**

Twenty-one patients with JIA, with a mean age of 13.67 ± 2.76 years, were included. Functional capacity was assessed using the 6MWT, and gait analysis was conducted using OpenCap. The markerless motion capture setup used two smartphone cameras positioned at 30°, with an inter-camera distance of 2 m. Participants walked at a self-selected comfortable speed. OpenCap-derived spatiotemporal and lower-extremity kinematic parameters were extracted. Associations between gait parameters and 6MWT distance were analyzed using Spearman correlation coefficients.

**Results**

Participants had a mean BMI of 20.1 ± 4.3 kg/m² and a mean of 1.9 ± 1.0 involved lower-extremity joints. The mean 6MWT distance was 673.7 ± 69.6 m, and the mean OpenCap-derived walking speed was 1.47 ± 0.32 m/s. Pelvic list range of motion showed a strong negative correlation with 6MWT distance (rho = -0.793, $p = 0.0036$). Similarly, pelvic list maximum was negatively correlated with 6MWT distance (rho = -0.738, $p = 0.0095$). The number of involved lower-extremity joints was also negatively correlated with 6MWT distance (rho = -0.499, $p = 0.021$). Walking speed showed a weak, non-significant correlation with 6MWT distance (rho = 0.197, $p = 0.391$).

**Conclusion**

OpenCap-based markerless gait analysis appears to be a feasible approach for biomechanical gait assessment in children and adolescents with JIA. Pelvic motion parameters, particularly pelvic list range of motion and maximum pelvic list, were more strongly associated with 6MWT distance than walking speed, suggesting that frontal-plane pelvic control may be clinically relevant to functional mobility in JIA. Given its scalable, cost-effective, and smartphone-based structure, OpenCap may support accessible three-dimensional gait analysis and objective functional monitoring in pediatric rheumatology. Further studies with larger samples are needed to confirm its clinical utility and responsiveness to rehabilitation interventions.

**Disclosure of interest**

None declared.

## P208

### Comparing internal and external focus strategies in telerehabilitation for children and adolescents with pediatric rheumatic diseases

A. Yilmaz[1,*], N. Aktay Ayaz[2], N.Arman[3]
[1]Physiotherapy and Rehabilitation Master of Science Program, Istanbul University-Cerrahpasa, Institute of Graduate Studies; [2] Department of Pediatric Rheumatology, Istanbul University, Istanbul Faculty of Medicine; [3]Department of Physiotherapy and Rehabilitation, Istanbul University-Cerrahpasa, Faculty of Health Sciences, Istanbul, Türkiye
**Correspondence:** A.Yilmaz
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**Introduction**
Pain and fatigue are common in Juvenile Idiopathic Arthritis (JIA), Familial Mediterranean Fever (FMF), and Juvenile Dermatomyositis (JDM), negatively affecting physical fitness and functional capacity. Exercise may help interrupt the pain-fatigue-inactivity cycle.Attentional focus strategies may further influence exercise performance. Internal focus (IF) directs attention to body movements or specific body parts, supporting body awareness, alignment, and movement control.External focus (EF) directs attention toward an external cue or target, promoting more automatic movement and reducing symptom-focused attention.
**Objectives**
This study aimed to compare the effects of IF and EF based telerehabilitation in patients with pediatric rheumatic diseases.
**Methods**
Twenty-eight participants diagnosed with FMF, JIA or JDM were randomized into EF ($n$=14) or IF ($n$=14) groups. Both groups completed an 8-week supervised telerehabilitation, delivered twice weekly. EF cues directed attention toward external visual targets, such as stickers, whereas IF cues focused on the moving body part. Outcomes included pain and fatigue using VAS and FSS; physical fitness using the FitnessGram Test Battery; functional capacity using the 6MWT, 30SST, and SCT; muscle strength using K-Push dynamometry/K-Force plates; and gait parameters using Digitsole Pro insoles. Participant experiences were explored qualitatively.
**Results**
Both approaches significantly improved physical fitness and functional capacity ($p$<0.05). The EF group showed significant improvements in upper extremity muscle strength and hamstring flexibility, whereas the IF group demonstrated improvements in body composition. Clinically meaningful improvements in 6MWT distance were observed in both groups. The EF group demonstrated significant reductions in pain, fatigue, and quadriceps asymmetry, while the IF group showed significant improvements in walking speed and cadence ($p$<0.05). Disease-specific analyses suggested that EF-based training was more favorable for pain and fatigue outcomes in FMF and JDM, whereas IF-based training was more effective for functional capacity and muscle strength outcomes in JIA ($p$<0.05). Participant feedback indicated that EF cues made exercises easier to understand, more engaging, and more enjoyable.
**Conclusion**
Both IF-and EF-based telerehabilitation programs appear beneficial for improving physical fitness and functional capacity in children and adolescents with pediatric rheumatic diseases. EF-based exercise may offer advantages for pain and fatigue management by shifting attention away from symptom perception. IF-based exercise may be useful when the therapeutic aim is to improve postural awareness, movement quality, gait parameters, and joint-related control.These findings suggest that attentional focus strategies may be tailored according to disease-specific clinical needs and rehabilitation goals.
**Trial registration identifying number**
NCT07138157.
**Disclosure of interest**
None declared.

## P209

### Clinical case of destructive arthritis of the first metatarsophalangeal joint as a very rare form of the onset of enthesitis–related arthritis in an adolescent female

A. Kozhevnikov[1,2,*], E. Chikulaeva[1], S. Semenov[1]
[1]H. Turner National Medical Research Center for Children's Orthopedics and Trauma Surgery; [2]Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation
**Correspondence:** A. Kozhevnikov
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**Introduction**
Enthesitis-related arthritis (ERA) is one of the subtypes of juvenile idiopathic arthritis (JIA), which is characterized by enthesitis and axial manifestations (sacroiliits). For ERA with childhood onset sacroiliitis may be delayed. According to literature ERA most commonly affected boys. Clinical manifestations of ERA are variable and similar to orthopedic pathology which determine diagnostic delay.
**Objectives**
This article presents a clinical case describing the successful treatment of a 15-year-old girl with atypical onset of ERA in form of destructive arthritis of the first metatarsophalangeal joint.
**Methods: Clinical case**
A 15-year-old girl was referred to our clinic with local pain of the first metatarsophalangeal joint (1st MTPJ) for a long year. Past medical history was uneventful. She complained of the pain every day with morning stiffness. The girl received treatment for hallus valgus and irregularly NSAIDs. Physical examination showed joint tenderness, swelling and painful mobility of the 1st MTPJ. X-ray and MRI revealed elevated destructive arthritis of the 1st MTPJ. There are no clinical and instrumental manifestations of tarsitis, enthesitis or dactylitis. Laboratory blood tests including CRP and ESR were normally; ANF titer of 1/1280, gene HLA B27 – negative. Antibody M и G to Borrelia burgdorferi were negative. JIA was diagnosed by biopsy and classified as a pauciarticular subtype. Bone tumor, tuberculosis lesion and osteomyelitis were excluded. The combination of methotrexate and anti-TNF etanercept was efficacy. ERA was revealed after 1.5 years treatment due to appear of axial lesion (low back pain, MRI sacroiliitis). The clinical manifestations of sacroiliitis reduced after switching to the anti-TNF golimumab treatment.
**Discussion**
The clinical manifestation of ERA can be highly variable. A feature of the juvenile onset is the prevalence of peripheral enthesitis and delayed appearance of axial lesion. Moreover, there are clinical differences at the onset and course of the disease between male and female patients. Foot lesion in children typically present as tarsitis or combined of the forefoot and mid-foot. Long-term of monoarthritis of the 1st MTPJ is not considered a typical onset of ERA in children. Destructive monoarthritis without enthesitis requires exclusion of wide range tumors-like disease or specific infections. In our case was diagnosis of oligoarticular subtype JIA. We decided that methotrexate and etanercept therapy efficacy for atypical oligoarthritis. But appear signs of axial involvement despite anti-TNF therapy was surprised. Switch to golimumab anti-TNF therapy was optimally.
**Conclusion**
ERA is a special subtype of JIA with a variety of heterogeneous forms. ERA can occur in both genders in forms of atypically onset and late axial involvement.
**Disclosure of interest**
None declared.

## P210

### Exergaming-assisted exercise in a child with coexisting juvenile idiopathic arthritis and Type 1 Diabetes Mellitus: a case report

I. Dönmez[1,*], A. Yekdaneh[2], A. Albayrak[3,4], S. Dogantan[5], N. Arman[6]
[1]Physiotherapist Irem Donmez, Private Health Services Unit; [2]Vocational School of Health Services, Physiotherapy English Program, Fenerbahce University; [3]Institute of Graduate Studies, Physiotherapy and Rehabilitation Doctorate Program, Istanbul University-Cerrahpasa; [4]Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Istanbul Kent University; [5]Pediatric Rheumatology Department, Basaksehir Cam and Sakura City Hospital; [6]Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Istanbul University-Cerrahpasa, Istanbul, Türkiye
**Correspondence:** I. Dönmez
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**Introduction**
Juvenile Idiopathic Arthritis (JIA) is the most common chronic inflammatory rheumatic disease in childhood. Type 1 Diabetes Mellitus (T1DM) is a chronic metabolic disorder caused by autoimmune destruction of insulin producing pancreatic beta cells. The coexistence

of these two autoimmune conditions is rare and may create additional challenges for physical activity, functional performance, exercise tolerance, and safety monitoring. Although traditional exercise programs can improve physical function, maintaining long term adherence may be difficult in children with complex chronic conditions. Exergaming assisted exercise may provide a motivating approach to support functional outcomes and patient engagement.

**Objectives**

This case report aimed to evaluate the effects and safety of a 6 week exergaming assisted exercise program on functional capacity, muscle strength, neuromuscular activation, flexibility, and lower-extremity performance in a child diagnosed with both JIA and T1DM.

**Methods**

A 9-year-old female with JIA-related knee restriction and T1DM completed a 6-week, twice-weekly combined program of traditional and exergaming-assisted exercises (45 min/session). Sessions targeted core stability, neuromuscular control, lower-extremity strength, and functional movement. Digital sensors (Kinvent Physio) assessed quadriceps sEMG (KMYO), handgrip strength (KGrip), and isometric hamstring/quadriceps strength (KPush). Functional capacity was evaluated via the 6-Minute Walk Test (6MWT) and 30-Second Sit-to-Stand Test (30STS). Pre-/post-session blood glucose and Numerical Rating Scale (NRS) scores for pain/fatigue monitored safety.

**Results**

Post-intervention, 6MWT distance increased by 16.7% (425 to 496 m) and 30STS repetitions by 42.8% (14 to 20). Left quadriceps sEMG activity rose by 619% (2.42 to 17.4). Right hamstring flexibility improved by 250% (6 to 21 cm). Isometric strength increased up to 86.9% (hamstrings) and 24.3% (quadriceps). Blood glucose remained within target (70–180 mg/dL) in 81% of sessions. The program was well-tolerated (mean pain/fatigue: 2/10).

**Conclusion**

This case report suggests that a combined traditional and exergaming-assisted exercise program may be feasible, safe, and beneficial for improving neuromuscular activation, lower-extremity strength, flexibility, and functional performance in a child with coexisting JIA and T1DM. The marked increase in quadriceps sEMG activity may indicate improved activation of the affected lower limb. Improvements in 30STS and 6MWT suggest potential gains in functional power, local muscular endurance, and walking capacity. Blood glucose monitoring showed that the program was generally safe within a supervised exercise setting. Further studies are needed to explore the safety and effectiveness of exergaming-assisted rehabilitation in pediatric patients with multiple chronic conditions.

**Disclosure of interest**

None declared.

## P212

### Disease duration influences the value of C-reactive protein and erythrocyte sedimentation rate at diagnosis in treatment-naive children with juvenile idiopathic arthritis

J. B. De Jonge[1,2,3,*], S. de Roock[1,2], D. A. Marshall[4,5], S. J. Vastert[1,2,3], R. S. Yeung[6], S. M. Benseler[5,7,8], J. F. Swart[1,2]

[1]Department of Pediatric Rheumatology, Division of Paediatrics, Wilhelmina Children's Hospital, University Medical Center Utrecht; [2]Faculty of Medicine, Utrecht University; [3]Center for Translational Immunology, University Medical Center Utrecht, Utrecht University, Utrecht, Netherlands; [4]Departments of Community Health Services and Medicine, McCaig Institute for Bone and Joint Health, O'Brien Institute for Public Health, Cumming School of Medicine, University of Calgary; [5]Alberta Children's Hospital Research Institute, Cumming School of Medicine, University of Calgary, Calgary, Alberta; [6]Division of Rheumatology, Department of Paediatrics, Immunology and Institute of Medical Science, The Hospital for Sick Children, University of Toronto, Toronto, Ontario; [7]Division of Rheumatology, Department of Pediatrics, Alberta Children's Hospital, Cumming School of Medicine, University of Calgary, Calgary, Alberta, Canada; [8]Children's Health Ireland, University College Dublin, Trinity College Dublin, Dublin, Ireland

**Correspondence:** J. B. De Jonge

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**Introduction**

The traditional inflammatory markers C-reactive protein (CRP) and erythrocyte sedimentation rate (ESR) are widely measured in children with Juvenile Idiopathic Arthritis (JIA), without knowing the exact value of these markers at diagnosis.

**Objectives**

This study aimed to describe the association of CRP and ESR with disease parameters at diagnosis and to investigate which variables best explain elevated CRP and ESR levels within treatment-naïve JIA patients.

**Methods**

The international UCAN CAN-DU study prospectively enrolled JIA patients across Canada and the Netherlands. This nested cohort study included treatment-naive (NSAIDs excluded) JIA patients with active arthritis and CRP and/or ESR levels available between April 2019 and December 2025. Children with systemic JIA were excluded. CRP and ESR values were considered elevated when CRP was ≥1 mg/dL and ESR ≥20 mm/h. Patient- and disease-related parameters were compared using (non)parametric tests among patients with normal and elevated CRP/ESR. Variables with a statistically significant ($p < 0.05$) effect were included in a logistic regression model to assess adjusted odds ratios between individual variables and elevated CRP and ESR levels.

**Results**

A total of 527 patients with treatment-naïve JIA were included; 62% were female, and the median age at symptom onset was 9.1 years. CRP and ESR levels were within normal range in 53% of the patients. The regression model for elevated CRP revealed an adjusted odds ratio of 1.38 (interquartile range (IQR): 1.23-1.55) for physician global assessment (PhGA) and 0.94 (IQR: 0.91-0.97) for time since symptom onset in months. The regression model for elevated ESR showed an adjusted odds ratio of 0.96 (IQR: 0.92-1.00) for age at symptom onset, 1.30 (IQR: 1.17-1.46) for PhGA, and 0.94 (0.92-0.97) for time since symptom onset in months.

**Conclusion**

An increase in PhGA and a decrease in time since symptom onset were associated with elevated CRP and ESR. The association between time since symptom onset and laboratory markers highlights the importance of biomarker measurement timing in their interpretation.

**Disclosure of interest**

J. De Jonge: None declared, S. de Roock: None declared, D. Marshall: None declared, S. Vastert Consultant with: Consulting, funding grants and speaking fees from Sobi; Consulting and speaking fees from Novartis, R. Yeung Consultant with: Consulting fees from Sobi, S. Benseler: None declared, J. Swart Consultant with: Consulting fees, funding grants, and lecture fees from Pfizer; advisory fees from Bristol-Myers Squibb.

## P215

### Serum biomarkers as predictors of remission and flare in juvenile idiopathic arthritis: results from the DAISY cohort

D. Lazarevic [1,*], D. Föll[2], F. Bovis[3], B. Sozeri[4], L. Rossi-Semerano[5], A. Snipaitiene[6], M. Burrone[7], D. Windschall[8], M. Tsinti[9], C. Høst[10], S. Lanni[11], C. Kessel[2], S. Fuehner[2], M. Senjug Perica[12], S. Scala[13], C. Malattia[14,15], V. Jelena[16]

[1]Clinic of Pediatrics, University of Nis, Nis, Serbia; [2]Depth of Pediatric Rheumatology and Immunology, University Hospital Muenster, Muenster, Germany; [3]Department of Health Sciences (DISSAL), University of Genova, Genova, Italy; [4]Division of Pediatric Rheumatology, Istanbul Umraniye Training and Research Hospital, Istanbul, Türkiye; [5]Department of Pediatric Rheumatology, Bicêtre Hospital, Le Kremlin Bicêtre, France; [6]Hospital of Lithuanian, University of Health Sciences Kauno Klinikos, Kaunas, Lithuania; [7]UOC Reumatologia e Malattie Autoinfiammatorie, IRCCS Istituto Giannina Gaslini, Genova, Italy; [8]University of Halle-Wittenberg and St. Josef-Stift, Sendenhorst, Germany; [9]Children Hospital Srebrnjak, Zagreb, Croatia; [10]Department of Paediatrics and Adolescent Medicine, Aarhus University Hospital, Aarhus, Denmark; [11]Fondazione IRCCS Cà Granda Ospedale Maggiore Policlinico, Milano, Italy; [12]Aghia Sophia Children's Hospital, Athens, Greece; [13]UOC Servizio Sperimentazioni Cliniche Pediatriche, IRCCS Istituto Giannina Gaslini; [14]Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genova; [157]UOC Reumatologia e Malattie Autoinfiammatorie, IRCCS Istituto Giannina Gaslini, Genova, Italy; [16]Clinic of Pediatrics, University Clinical Center, University of Nis, Nis, Serbia

**Correspondence:** D. Lazarevic

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**Introduction**
Juvenile idiopathic arthritis (JIA) comprises heterogeneous chronic inflammatory diseases characterized by a relapsing–remitting course and variable long-term outcomes. Despite treat-to-target strategies, predicting treatment response and relapse remains challenging.
**Objectives**
To evaluate the role of serum biomarkers in assessing disease activity and predicting remission and flare in JIA.
**Methods**
Patients with active JIA requiring treatment escalation were evaluated at baseline and after 3, 6, and 12 months. Disease activity was assessed using JADAS10. Serum levels of S100A12, S100A8/A9, IL-6, IL-17A, soluble IL-2 receptor alpha (sIL-2Rα/CD25), CXCL9, CXCL10, VEGF-A, G-CSF, TNF-α, and IL-13 were measured at each visit. Biomarker distribution across JIA subtypes and correlations with JADAS10 were analysed. Associations between baseline biomarkers and time to inactive disease were assessed using Cox regression adjusted for JIA subtype, ultrasound score, and ongoing treatment. Landmark analysis evaluated the association between biomarker levels at remission and subsequent flare risk.
**Results**
A total of 101 patients were included: 33 oligoJIA, 55 polyJIA, and 13 ERA; median disease duration was 37 months. S100 proteins, CXCL9, and CXCL10 were significantly higher in polyJIA, whereas IL-17A and VEGF-A were increased in ERA. TNF-α levels were higher in oligoJIA. S100A12, S100A8/A9, and IL-6 showed the strongest correlations with JADAS10 across subtypes. Median time to inactive disease was 6.4 months, and 68% of patients achieved remission within 12 months. Higher baseline S100A12 levels were associated with a lower probability of remission (HR 0.60, 95%CI 0.36–0.99; $p$=0.045). No significant associations were observed for the other biomarkers. During follow-up, 48 patients experienced disease flare after remission. Higher S100A12 (HR 7.63, 95%CI 2.01–33.85; $p$=0.002) and S100A8/A9 levels (HR 6.85, 95%CI 1.65–32.86; $p$=0.008) at remission were associated with increased flare risk, whereas IL-17A was inversely associated with flare (HR 0.18, 95%CI 0.05–0.59; $p$=0.004).
**Conclusion**
S100A12, S100A8/A9 may represent valuable biomarkers for integration into routine clinical practice to identify patients with JIA at increased risk of an unfavourable disease course, including delayed achievement of inactive disease and relapse following remission.
**Funding**
This study is supported by the Foundation for Research in Rheumatology (FOREUM).

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**Disclosure of interest**
None declared.

## P217

### Baricitinib for interstitial lung involvement in juvenile idiopathic arthritis: a two-case series

O. Lomakina[1,*], S. Valieva[1,2], S. Kurbanova[1], I. Samolygo[3]
[1]Morozov City Children's Clinical Hospital; [2]Federal State Autonomous Educational Institution of Higher Education «N.I. Pirogov Russian National Research Medical University of the Ministry of Health of the Russian Federation; [3] Department of Propaedeutics of Children's Diseases N.F. Filatov Clinical Institute of Child Health Sechenov University, Moscow, Russian Federation
**Correspondence:** O. Lomakina
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**Introduction**
Pulmonary involvement in juvenile idiopathic arthritis (JIA) is uncommon and may be difficult to distinguish from infection, particularly in young children receiving biologics. Evidence on treatment of JIA-associated interstitial lung disease (ILD) is limited.
**Objectives**
To describe clinical and radiological outcomes of two young children with JIA and CT-confirmed ILD treated with baricitinib without systemic glucocorticoids.
**Methods**
Retrospective case series from a tertiary paediatric rheumatology centre. Data included arthritis activity, ESR/CRP, chest CT findings, and infectious work-up (including bronchoscopy with bronchoalveolar lavage (BAL) when indicated). Baricitinib was prescribed using standard-paediatric/weight-baseddosing.
**Results**
**Case 1:** Female (born 2023) developed severe polyarthritis after a viral infection in infancy with high inflammatory activity; polyarticular JIA (high activity) diagnosed after malignancy exclusion. Tocilizumab plus methotrexate induced remission. In Oct 2024, despite no respiratory symptoms, CT revealed multifocal ground-glass opacities and fibrotic changes, in connection with this, therapy with tocilizumab and methotrexate was cancelled. BAL/CT findings were interpreted as invasive pulmonary aspergillosis; voriconazole led to initial CT improvement. By Jul 2025, CT abnormalities persisted and arthritis flared with elevated ESR/CRP; treatment was switched to baricitinib while antifungal therapy continued. In Oct 2025 (age 3y) arthritis was inactive (CRP 1 mg/L, ESR 10 mm/h) and CT improved, with only a single residual subpleural opacity; antifungal therapy was stopped.
**Case 2:** Female (born 2021) with Down syndrome developed oligoarticular JIA in Jun 2025 after recent pneumonia. Inflammatory markers were mildly elevated (ESR 20 mm/h, CRP 3 mg/L). Baseline CT showed reticular changes and ground-glass consolidations in posterior lower lobes consistent with ILD. Baricitinib was started; by Oct 2025 clinical symptoms resolved and follow-up CT demonstrated improvement.
**Conclusion**
In two young children with JIA and CT-confirmed ILD, baricitinib (without systemic glucocorticoids) was associated with control of arthritis and improvement/stabilisation of lung abnormalities. Careful infectious evaluation and close monitoring are essential, particularly in patients with suspected opportunistic infection.
**Disclosure of interest**
None declared.

## P218

### Body composition in children with juvenile idiopathic arthritis (pilot study)

L. Firsova[1,*], I. Avrusin[2], O. Kalashnikova[2], A. Zavyalova[1], V. Novikova[1], M. Kostik [2]
[1]Propaedeutics of Childhood Diseases named after V.Yuriev; [2]Hospital Pediatrics, Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation
**Correspondence:** L. Firsova
*Pediatric Rheumatology 2026*, **24(S1):**P218

**Introduction**
Bioimpedance analysis of the body in children with juvenile idiopathic arthritis (JIA) is an additional tool for the early diagnosis of changes in body composition.
**Objectives**
To analyze physical development and body composition in children with JIA.
**Methods**
The pilot study included 27 patients (13 boys, 14 girls) with JIA aged 13 [10; 16] years. There were 13 children with oligoarthritis (JIA-o), 5 – with polyarthritis (JIA-p), and 9 – with spondyloarthritis (JIA-s). All of them underwent anthropometric measurements and bioimpedance analysis of the body using MEDASS ABC-02 device. Anthropometric parameters were assessed using WHO AnthroPlus program. Waist circumference was assessed using centile tables developed for Russian children (2014). Body components were assessed using centile graphs, presented in MEDASS ABC-02 computer program.
**Results**
Median height, body weight, and BMI Z-scores were average in all studied groups. The distribution of *waist circumference* indicators in JIA-o group according to centile corridors (CCs) was as follows: 3$^{rd}$ CC-15.38%, 4$^{th}$ CC -61.54%, 7$^{th}$ CC -23.08%; JIA-p group: 2$^{nd}$, 5$^{th}$ and 7$^{th}$ CCs – 20% each, 4$^{th}$ CC-40%; JIA-s group: 1$^{st}$, 3$^{rd}$, and 6$^{th}$ CCs – 22.2% each, in 4$^{th}$, 5$^{th}$, and 7$^{th}$ CCs – 11.1% each.
*Phase angle* Z-score in JIA-o group was -0.52 [-1.6; 0.32], in JIA-p group, Z-score was -1.9 [-2.8; -1.6], and in JIA-s group, it was -0.28 [-0.98; 0.45].

**Table 1 (Abstract P218)** Body components in children of study groups

| Centile corridor<br>Body component | 1st (%) | 2nd (%) | 3rd (%) | 4th (%) | 5th (%) | 6th (%) | 7th (%) |
|---|---|---|---|---|---|---|---|
| **Fatty mass**, abs./% | 0/ 0 | 0/ 0 | 15.4/ 15.4 | 53.9/ 38.5 | 0/ 23.1 | 7,7/ 23.1 | 23.1/ 0 |
| | 0/ 0 | 0/ 0 | 20/ 20 | 40/ 40 | 0/ 0 | 20/ 40 | 20/ 0 |
| | 0/ 0 | 22.2/ 22.2 | 0/ 11.1 | 44.4/ 22.2 | 11.1/ 22.2 | 11.1/ 22.2 | 11.1/ 0 |
| **Active cell mass**, abs./% | 0/ 23.1 | 30.8/ 7.7 | 7.7/ 15.4 | 46.2/ 46.2 | 0/ 0 | 15.3/ 7.7 | 0/ 0 |
| | 20/ 20 | 20/ 60 | 20/ 20 | 40/ 0 | 0/ 0 | 0/ 0 | 0/ 0 |
| | 0/ 11.1 | 11.1/ 0 | 33.3/ 22.2 | 33.3/ 44.4 | 22.2/ 0 | 0/ 0 | 0/ 0 |
| **Musculoskeletal mass**, abs./% | 0/ 0 | 0/ 0 | 15.4/ 0 | 38.5/ 30.7 | 15.4/ 23.1 | 15.4/ 7.7 | 15.4/38.5 |
| | 0/ 0 | 0/ 0 | 20/ 20 | 60/ 0 | 20/ 20 | 0/ 60 | 0/ 0 |
| | 0/ 0 | 11.1/ 0 | 0/0 | 66.7/ 44.4 | 22.2/ 11.1 | 0/ 11.1 | 0/ 33.3 |

Data in columns are presented in the order (from top to bottom): JIA-o, JIA-p, JIA-s

*Abbreviations*: abs – absolute value

*Total body fluid* was mostly within the 4th CC (46.2% – JIA-o, 60% – JIA-p, 66.7% – JIA-s), and only in JIA-o group, 15.4% of cases were in the 7th CC. The distribution of main body components is presented in Table 1.

**Conclusion**

Body composition was different in children with different JIA cathegories, while medians of basic anthropometric parameters were average. It is necessary to conduct additional study to investigate the role of therapy, physical activity and nutrition.

**Disclosure of interest**

None declared.

## P219

### Are we missing active disease? MRI-detected subclinical synovitis predicts flare in juvenile idiopathic arthritis

M. Natal[1,2], R. Nicolau[3], F. Gomes[4], J. P. Moreira[5], P. J. Pereira[4], I. Cardoso[6], B. Fernandes Esteves[1,2], D. Milkowska Mikiel [7], S. Ganhão[8], F. Aguiar[2,8], M. Rodrigues[2,9,*], S. Dias Costa[2,10], I. Brito[2,9]

[1]Rheumatology Department, ULS São João; [2]Department of Medicine, Faculty of Medicine, University of Porto, Porto; [3]Rheumatology Department, ULS Trás-os-Montes e Alto Douro, Vila Real; [4]Rheumatology Department, ULS Braga, Braga; [5]Department of Pediatric Cardiology, ULS São João, Porto; [6]Pediatrics Department, ULS Gaia/Espinho, Gaia/Espinho; [7]Pediatrics Department, ULS Matosinhos, Matosinhos; [8]Pediatric and Young Adult Rheumatology Unit; [9]Pediatrics Department; [10]Radiology Department, ULS São João, Porto, Portugal

**Correspondence:** M. Rodrigues

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**Introduction**

Clinical remission in juvenile idiopathic arthritis (JIA) does not exclude ongoing synovial inflammation on imaging, the prognostic significance of which remains uncertain.

**Objectives**

To evaluate the association between knee MRI findings and clinical disease activity in patients with JIA, and to assess whether subclinical synovial inflammation predicts disease flare during follow-up.

**Methods**

Single-center prospective cohort of children aged 1–18 years with JIA with current or previous knee involvement who underwent knee MRI. MRI findings were categorised as showing substantial synovial inflammation (DSI) or absent/low level findings (non-DSI), based on the degree of synovial abnormality. Clinical remission was defined as JADAS-27-ESR ≤1. Patients without available follow-up data were excluded. Flares were identified by the attending rheumatologist. Survival analysis included Kaplan-Meier estimation, log-rank testing, and univariable Cox regression.

**Results**

Forty-nine children were included (mean age 11.3 ± 4.2 years; 30.6% male; mean disease duration 6.1 ± 4.2 years; 86% ANA-positive). Patients were followed for a median of 19.7 months (range 5.0–24.8). DSI and non-DSI groups did not differ in age, sex, JIA subtype, or treatment (all $p>0.05$). Patients with DSI had higher JADAS-27-ESR ($p=0.021$), more swollen joints ($p=0.022$), higher VAS Physician ($p=0.030$), higher CRP ($p=0.046$), and more frequent clinical knee activity (36.4% vs. 7.4%, $p=0.029$). Agreement of MRI with JADAS-ESR (κ=0.270) and clinical knee assessment (κ=0.305) was only fair. Subclinical inflammation (DSI in the absence of clinical disease activity) was present in 7/23 (30.4%, 95% CI 15.6–50.9%) patients in JADAS-ESR remission and in 14/39 (35.9%, 95% CI 22.7–51.6%) patients in clinical knee remission. During follow-up, 14/49 patients (28.6%) experienced flare. DSI predicted flare in the overall cohort (HR 3.38, 95% CI 1.01–11.35, $p=0.049$). Among those in clinical remission by JADAS, subclinical inflammation was associated with markedly increased flare risk (HR 11.42, 95% CI 1.12–116.15, $p=0.040$), with a consistent finding when remission was defined by clinical knee assessment (HR 5.14, 95% CI 1.27–20.73, $p=0.022$).

**Conclusion**

Knee MRI findings reflected clinical disease activity but showed only fair agreement with clinical assessment. Subclinical synovial inflammation was identified in a clinically meaningful proportion of JIA patients otherwise considered to be in remission, whether defined by systemic disease activity (JADAS-ESR) or by clinical knee assessment. The substantial association with subsequent flare suggests that knee MRI may add prognostic information to clinical assessment in identifying patients at higher risk of relapse, with potential implications for treatment de-escalation decisions.

**Disclosure of interest**

None declared.

## P220

### Interim results of "RIANA" – registry for intra-articular corticosteroid injection (IACI): follow-up protocols for gonarthritis in children and adolescents

S. Meister[1,*], M. Georgi[1], T. Strauss[2], L. Buchtala[3], F. Weller-Heinemann[3], J. P. Haas[1]

[1]German Center for Pediatric and Adolescent Rheumatology, Center for Pain Therapy in Young People, Garmisch-Partenkirchen, Garmisch-Partenkirchen; [2]Department of Pediatrics and Adolescent Medicine, Faculty of Medicine and Carl Gustav Carus University Hospital, Dresden University of Technology, Dresden; [3]Prof. Hess Parent-Child Center, Bremen-Mitte Hospital, Bremen, Germany

**Correspondence:** S. Meister

*Pediatric Rheumatology 2026*, **24(S1):**P220

**Introduction**

The lack of evidence-based recommendations for postoperative management after knee IACI, along with practice variability within the Society of child and adolescence rheumatology (GKJR)[1], prompted the establishment of a prospective registry to assess joint loading vs. unloading after knee IACI and its impact on time to onset of clinical and/or laboratory signs of inflammation. Data are collected from pediatric rheumatology centers providing follow-up care with/without knee unloading after IACI.

**Objectives**

To present available T0(pre-IACI) and T1(12–20 weeks post-IACI) data from centers with and without post-IACI unloading on pain, active/passive joint mobility, physical/sports activity.

**Table 1 (Abstract P220)** Descriptive statistics of each group; Min, Max, mean, standard deviation(SD), (afROM/pfROM)

| | unloading T0 | | T1 | | non-unloading T0 | | T1 | |
|---|---|---|---|---|---|---|---|---|
| | (*n*=22) f=68% | | | | (*n*=4) f=75% | | | |
| | (*n*=22)Min-Max | Mean(SD) | (*n*)Min-Max | Mean(SD) | (*n*)Min-Max | Mean(SD) | (*n*)Min-Max | Mean(SD) |
| **pain(NAS)** | 0-7 | 3,6(2,2) | (20)0-6 | 0,7(1,6) | (4)1,5-2 | 1,9(0,2) | (4) 0 | 0 |
| **afROM(°)** | 110-155 | 127(11) | (22)120-160 | 144(11) | (1)155 | | (2)125-155 | 148(13) |
| **pfROM(°)** | 120-165 | 135(11) | (22)138-170 | 153(9) | (3)140-155 | 152(9) | (4)125-165 | 153(16) |
| **physical activity(h)** | 2-30 | 10,3(7,8) | (21)2-35 | 12,8(9,9) | (4)10-35 | 18(13) | (4)2-20 | 7,6(7,6) |
| **sports activity(h)** | 0-8 | 3,3(2,7) | (21)0-10 | 3,6(2,9) | (4)3-6 | 3,8(1,3) | (4)3-5,5 | 3,9(1) |

**Methods**
Knee joint mobility was assessed by goniometry at T0 and T1 in unloading and non-unloading groups. Pain and weekly physical/sports activity were recorded. Data were analyzed descriptively. Normality was assessed using Shapiro–Wilk test, paired t-tests/Wilcoxon signed-rank tests were used as appropriate to compare T0 and T1.
**Results**
26 datasets from three centers completing T1 were analyzed. (Table 1). Due to the small control group, inferential statistics were limited to the unloading group. Passive knee range of motion (pfROM: sum of passive flexion and extension minus extension deficit) increased from T0(mean=131, SD=17.84) to T1(mean=153, SD=9.08), t(21)=6.49, $p$<0.001, d=1.38. Total active range of motion (afROM) increased from T0(mean=122, SD=18.17) to T1(mean=144, SD=11.48), t(21)=6.13, $p$<0.001, d=1.31. Pain decreased from T0(Md=3.5) to T1(Md=0), $p$<0.001, $r$=0.75. Physical and sports activities remained unchanged.
**Conclusion**
The unloading group showed significant improvements in pain and joint mobility over time. Whether similar changes occur in the non-unloading group is currently under investigation. Currently, neither approach can be considered superior.

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**Trial registration identifying number**
DRKS00036083.
**Disclosure of interest**
None declared.

## P221

### Evaluation of outcomes of intra-articular steroid injections into temporomandibular joint in patients with juvenile idiopathic arthritis

N. Gürbüz[1,*], V. Shivamurthy[2]
[1]Department of Pediatric Rheumatology, Ege University, İzmir, Türkiye; [2]Department of Pediatric Rheumatology, Evelina Children's Hospital, London, UK
**Correspondence:** N. Gürbüz
*Pediatric Rheumatology 2026*, **24(S1):**P221

**Introduction**
Temporomandibular joint (TMJ) involvement is a common yet often underrecognized manifestation of Juvenile Idiopathic Arthritis (JIA). Delayed recognition may lead to functional impairment, facial asymmetry, mandibular growth disturbances. Early detection and appropriate treatment are therefore essential.
**Objectives**
This study aims to evaluate the outcomes of intra-articular steroid (IAS) injections in a cohort of pediatric patients with JIA.
**Methods**
This retrospective study included patients with JIA who were treated with IAS injection into TMJ at a tertiary pediatric rheumatology center between January 2021 and October 2025. Demographic data, clinical diagnoses, TMJ symptoms, JIA subtype, serological markers (ANA, RF, HLA-B27, CCP), systemic treatments, magnetic resonance imaging (MRI) findings and clinical outcomes of IAS injection therapy were extracted from the medical records. MRI findings were evaluated for active inflammatory changes, including synovitis, joint effusion, bone marrow edema, as well as chronic structural abnormalities such as condylar flattening, erosions, disc displacement, mandibular ramus shortening.
**Results**
The cohort comprised 15 patients (12 females, 3 males) with a median age of 16 years (range 10–19 years). TMJ involvement presented with jaw pain, limited mouth opening, jaw deviation, clicking. MRI commonly demonstrated active synovitis, condylar flattening, erosions, bone marrow edema, disc displacement. The JIA subtypes included oligoarticular ($n$=5, %33.3), polyarticular ($n$=6, %40), psoriatic ($n$=3, %20), IBD-associated arthritis ($n$=1, %6.7). IAS injections were administered to 15 patients, involving a total of 18 TMJ joints. Methylprednisolone was used in 4 patients (%26.7), whereas triamcinolone acetonide/hexaacetonide (2–4 mg) was administered in 11 patients (%73.3). All patients were receiving systemic therapy, including methotrexate monotherapy, biologic monotherapy (adalimumab, etanercept, infliximab, tocilizumab) or combination therapy with conventional synthetic and biologic DMARDs. 5 patients (%33.3) had associated extra-articular manifestations, including uveitis in 3 patients and psoriasis in 2 patients. 12 patients (%80) demonstrated improvement in pain, chewing function, mouth opening, whereas 3 patients (%20) showed no improvement in inflammatory symptoms.
**Conclusion**
TMJ involvement is a significant manifestation of JIA and may occur across multiple disease subtypes. MRI remains the gold standard for diagnosis. In conclusion, early intervention with IAS injections and systemic therapy may improve clinical inflammatory symptoms, particularly by significantly reducing pain.

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**Disclosure of interest**
None declared.

## P222

### Relapse factors and seasonal influences in juvenile idiopathic arthritis following DMARD withdrawal: the impact of vitamin D and anatomical patterns

Ö. Özenli Yağcı[1,*], Ç. Yıldız[1], D. Gezgin Yıldırım[2], S. A. Bakkaloğlu[1]
[1]Department of Rheumatology; [2]Gazi University Faculty of Medicine, Ankara, Türkiye
**Correspondence:** Ö. Özenli Yağcı
*Pediatric Rheumatology 2026*, **24(S1):**P222

**Introduction**
Optimal timing of DMARD discontinuation and identification of relapse-associated factors remain major uncertainties in JIA management.
**Objectives**
This study aimed to evaluate withdrawal success of conventional synthetic (cs) and biologic (b) DMARDs in oligoarticular (oJIA) and polyarticular (pJIA) JIA, and to identify clinical, laboratory, metabolic and seasonal factors associated with relapse.

**Methods**
We retrospectively analyzed 132 DMARD withdrawal attempts in 58 patients (49 oJIA, 9 pJIA) diagnosed according to ILAR criteria. Clinical and laboratory parameters, 25-OH vitamin D levels, seasonal distribution of relapses, and anatomical joint concordance at relapse were analyzed.
**Results**
bDMARD discontinuation success was significantly higher compared to csDMARD ($p$=0.026). A remission duration of ≥18 months prior to bDMARD discontinuation was associated with longer relapse-free survival ($p$=0.001). In oJIA, younger age at symptom onset, higher baseline ESR, and ANA positivity were significantly associated with relapse ($p$<0.05 for all). The combination of ANA positivity and failure to achieve remission within the first three months predicted relapse in 100% of cases. 25-OH vitamin D levels at relapse were significantly lower than at withdrawal (median 9.4 ng/mL, $p$<0.001), with a threshold of 10.5 ng/mL predicting relapse with 100% sensitivity and 66.7% specificity (AUC: 0.868). Relapses peaked in autumn (33.3%). Anatomically, all oJIA patients with initially only large joint involvement significantly transitioned to small joint involvement during relapse ($p$=0.003).
**Conclusion**
bDMARD discontinuation appears safer when remission exceeds 18 months. ANA positivity, early-onset disease, and low vitamin D levels are important factors associated with relapse. The seasonal pattern and vitamin D threshold identified in this study may inform individualized monitoring protocols in JIA.

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**Disclosure of interest**
None declared.

## P223

### Baseline clinical, histologic and immunologic features associated with clinically inactive disease off-medications at 6-months in children with oligoarticular JIA

P. Vega-Fernandez[1,2,*], S. Thornton[3,4], D. Lovell[2,3], S. Angeles-Han[2,3], M. Vater[2,3], J. Rodriguez-Smith[2,3], E. Ogbu[2,3], K. Banschbach[2,3], A. Grom[2,3], H. Brunner[2,3], J. Huggins[2,3], M. Matt[2,3], M. Altaye[2,3], A. Cassedy[5], S. Szabo[3,6], T. V. Ting[1,2]
[1]Pediatrics, University of Cincinnati, Cincinnat; [2]Rheumatology, Cincinnati Children's Hospital Medical Center; [3]Pediatrics, University of Cincinnati; [4]Pediatrics; [5]Biostatistics; [6]Pathology, Cincinnati Children's Hospital Medical Center, Cincinnati, USA
**Correspondence:** P. Vega-Fernandez
*Pediatric Rheumatology 2026*, **24(S1):**P223

**Introduction**
Current treatment strategies for juvenile idiopathic arthritis (JIA) often follow a step-up approach progressing from intra-articular corticosteroids to conventional DMARDs and biologic DMARDs. However, standard-of-care practices cannot predict need for systemic therapy, disease course, or treatment response.
**Objectives**
Investigate the baseline clinical characteristics, histologic findings, and immunologic features associated with attainment of clinically inactive disease (CID) at 6 months following an intra-articular corticosteroid injection in treatment-naive children with oligoarticular JIA.
**Methods**
Treatment-naïve patients with oligoarticular JIA followed at Cincinnati Children's Rheumatology clinic were enrolled at diagnosis. At baseline, patients underwent standardized clinical assessment, serum and synovial fluid (SF) collection, ultrasound-guided synovial biopsy and intra-articular corticosteroid injection of the affected joint(s). Synovial biopsy severity was assessed using the Krenn's score (KS). Biomarkers measured in serum and SF included: TNFR1, IL-6, IL-10, MCP-1, ANG-1, MMP-2, MMP-8, CCL18, CCL20, IL-17a, IL-2Ra, IL-23, IL-18, CXCL9, VEGF, by Luminex, and S100A12, S100A8/A9 by ELISA. CID was defined as active joint count = 0 and physician global assessment = 0 at 6 months. Children who achieve CID following a single intra-articular corticosteroid injection without subsequent systemic therapy (CID off-medications group) were compared with those who required additional medications or did not achieve CID (Comparison group). Descriptive statistics were calculated, and group differences were assessed using chi Square, T-tests, and Wilcoxon rank sum tests as appropriate.
**Results**
This preliminary analysis included 27 patients (33% males, 85% White) with a median age of 10 (5-12) years. Children in the CID off-medications group tended to be older ($p$=0.08) and had lower ESR ($p$=0.01), SF Ang-1 ($p$=0.02) and serum IL2Ra ($p$=0.03) levels. In addition, lower baseline total KS, and lower levels of SF IL-6, IL-2Ra, CXCL-9, and IL-18 were observed in the CID off-medications group ($p$<0.10).

| | Total | CID off-medications group | Comparison group | *p*-Value |
|---|---|---|---|---|
| **High ESR, n (%)** | 11 (42.3) | 1 (10.0) | 10 (62.5) | 0.010 |
| **Total KS, median (IQR)** | 5 (2-6) | 3 (2-4.5) | 5.5 (4-7) | 0.042 |
| **SF ANG-1 >0.20, n (%)** | 7 (41.2) | 0 (0.0) | 7 (63.6) | 0.017 |
| **Serum IL2Ra** | 1.06 (0.69-1.33) | 0.69 (0.52-0.88) | 1.2 (1.06-1.31) | 0.027 |

**Conclusion**
Pilot data suggest that baseline clinical, histological and inflammatory features may distinguish children with oligoarticular JIA who respond to local therapy alone from those who require treatment escalation or develop persistent disease. Early identification of high-risk patients could enable more timely therapeutic optimization. Larger studies are needed to validate predictive markers of disease course and treatment response.
**Disclosure of interest**
None declared.

## P224

### 12 months follow up of the PROKIND cohort: Treat-to-target (T2T) approaches provide comparable outcomes in polyarticular and oligoarticular JIA

P. Berger[1,*], K. Tenbrock[1,2], J. Klotsche[3]
[1]Translational Rheumatology, RWTH Aachen, Aachen, Germany; [2]Pediatric Rheumatology, Inselspital Bern, Bern, Switzerland; [3]Deutsches Rheumaforschungszentrum, Berlin, Germany
**Correspondence:** P. Berger
*Pediatric Rheumatology 2026*, **24(S1):**P224

**Introduction**
Oligoarticular (oJIA) and polyarticular juvenile idiopathic arthritis (pJIA) are the most common JIA subtypes, affecting approximately 1–3 per 1000 children in highincome countries. OJIA typically presents with gonarthritis and involvement of ankles and elbows, often leading to pain, reduced mobility, and impaired quality of life even during remission. Despite major therapeutic advances, no validated treatment algorithms exist, and clinical decisions still rely heavily on individual experience and centerspecific standards. Standardization has been shown to benefit both patient care and research. In 2020, Klein et al. introduced a treattotarget (T2T) strategy for pJIA, demonstrating feasibility and clinical benefit over six months. Building on this, an adapted T2T algorithm for both oJIA and pJIA was developed, allowing free choice of first-line therapy but requiring mandatory 3monthly assessments over 12 months. A total of 248 pediatric patients were enrolled in the German multicenter ProKind initiative to validate this approach, with JADAS improvement or remission as the primary endpoint. Comparative longitudinal analyses of oJIA and pJIA cohorts provide important insights into

disease activity, treatment patterns, and remaining challenges in routine care. This study represents the largest T2Ttreated cohort to date and the first published dataset including oJIA patients.

**Objectives**

Analysis of feasibility and clinical benefit of T2T in oligoarticular and poliarticular JIA over the course of 12 months.

**Methods**

Patients with early and active polyarticular JIA (pJIA) and oligoarticular JIA (oJIA) were recruited between 2019 and 2023 and treated according to published protocols. Outcome measures after 12 months were Juvenile Arthritis Disease Activity Score (JADAS) and PEDsQL.

**Results**

530 patients (248 oJIA, 282 pJIA) were enrolled of which 417 (191 oJIA, 226 pJIA) were suitable for 12 months follow up analysis. 50.1% of oJIA and 63.7% of pJIA reached JADAS remission and 80.2% of oJIA and 80.6% of pJIA reached JADAS minimal disease activity (JADAS-MDA). PedsQL improved to 85/88% respectively for oJIA and pJIA. A bDMARD was used in oJIA in 10% and in pJIA in 41% of patients.

**Conclusion**

The T2T concept is feasible in oJIA and pJIA. High rates of patients reached JADAS-MDA and JADAS remission after 12 months. pJIA is slightly better with respect to remission which might be related to the higher bDMARD usage in this cohort.

**Trial registration identifying number**

DRKS-ID 00032928.

**Disclosure of interest**

None declared.

## P225

### The characteristics of patients with juvenile idiopathic arthritis depending on age of the disease onset

R. Raupov[1,*], S.Vissarionov[1], M. Orlova[1,2]
[1]H. Turner National Medical Research Center for Children's Orthopedics and Trauma Surgery; [2]Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation
**Correspondence:** R. Raupov
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**Introduction**

Age at Juvenile idiopathic arthritis (JIA) onset represents an important dimension of this heterogeneity, being associated with specific patterns of joint involvement, extraarticular features, and laboratory characteristics, and may thus inform risk stratification and treatment planning.

**Objectives**

to compare the patients with JIA according the age of disease onset.

**Methods**

198 patients with JIA were involved in the single-center prospective study. They were divided into 3 groups depending the disease onset: less 6 years (110 patients)-gr, 7-11 years (64 patients) and older than 12 years (23 patients). Clinical and laboratory data on disease onset, treatment and outcome were compared in 3 groups.

**Results**

The girls are predominated in the <6 years group, than in 7-11 and in the >12 years (70.9% vs. 56.2% vs. 56.5%, $p$=0.105). Median time to JIA diagnosis was 2.03 months in children <6 years, 5.07 months in those 7–11 years, and 4.1 months) in those ≥12 years ($p$<0.001). Oligoarthritis was most frequent in the youngest group, occurring in 68.2% of patients <6 years, compared with 50.0% of patients 7–11 years and 17.4% of those ≥12 years ($p$<0.001). Enthesitis-related arthritis was diagnosed in 7.3% of patients <6 years, 18.8% of those 7–11 years, and 47.8% of those ≥12 years. RF-negative polyarthritis was similar in groups – 19.1%, 26.6%, and 17.4%, as psoriatic – 3.6%, 0% and 4.3%, as undifferentiated arthritis for 1.8%, 4.7%, and 8.7%. Uveitis was documented in 13.6% of children with onset <6 years, 3.1% of those with onset 7–11 years, and was absent in the ≥12 years group ($p$=0.017). Elevated ESR at onset was found in 31.5%, 16.4%, and 40.9% of patients in the three groups, respectively ($p$=0.037), whereas elevated CRP was present in 17.3%, 15.9%, and 39.1% ($p$=0.038). ANA positivity was seen in 62.4% of patients <6 years, 60.3% of those 7–11 years, and 39.1% of those ≥12 years ($p$=0.116). HLA-B27 positivity increased with age, from 9.3% in the youngest group to 22.2% in the 7–11 years group and 47.8% in the ≥12 years group ($p$<0.001). IAGCS were used in 53.6% of patients with onset <6 years, 52.4% with onset 7–11 years, and 57.1% with onset ≥12 years ($p$=0.931). Methotrexate was prescribed in 89.0%, 64.1%, and 69.6% of patients in the three age groups, respectively ($p$<0.001). Biologic DMARDs was used in 21.1% of patients <6 years, 31.2% of those 7–11 years, and 30.4% of those ≥12 years ($p$=0.284). Median time to biologic was 41 months in the <6 years group, 13.25 months in the 7–11 years group, and 6.8 months in the ≥12 years group ($p$=0.020). JIA flares occurred in 55.7% of patients <6 years, 50.0% of those 7–11 years, and 45.5% of those ≥12 years ($p$=0.601). Median follow-up duration were 50.2 month in the <6 years group, 30.4 months in the 7–11 years group, and 14.2 months in the ≥12 years group ($p$<0.001).

**Conclusion**

the age-related differences in JIA phenotype and laboratory profile in this cohort are consistent with previously published data. A new and clinically relevant observation is that patients with onset after 12 years required biologic therapy much earlier than younger children, emphasizing the need for more aggressive early management in this subgroup.

**Disclosure of interest**

None declared.

## P226

### The elbow involvement in juvenile idiopathic arthritis

R. Raupov[1,*], S. Vissarionov[1], M. Orlova[1,2], M. Kostik[2]
[1]H. Turner National Medical Research Center for Children's Orthopedics and Trauma Surgery; [2]Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation
**Correspondence:** R. Raupov
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**Introduction**

Hip, cervical spine, ankle and wrist joint involvement in juvenile idiopathic arthritis (JIA) patients are predictors of more severe disease course and poor outcomes. The elbow involvement is less common in JIA and its prognostic role is understudied.

**Objectives**

to analyze patients with JIA with and without elbow involvement.

**Methods**

198 patients with non-systemic JIA categories were included in the study and 20 (10%) patients had the elbow involvement. We compared patients without and without elbow involvement. The median follow-up duration was 35 (15–62) months.

**Results**

Elbow joint involvement was most frequently observed in the polyarticular RF-negative category, less often in the oligoarticular and enthesitis-related subtypes (65% vs. 30% vs. 5%, $p$<0.001). Elbow arthritis was commonly accompanied by involvement of the knee (90%), ankle (70%), small joints of the hands (50%), and wrists (30%). It was also associated with cervical spine (25%, $p$=0.005) and small foot joint involvement (30%, $p$=0.025). Elevated inflammatory markers (ESR and CRP) were present in 40% of patients with elbow arthritis compared with 26% of those without it ($p$=0.074). Methotrexate was administered to 85% of patients; disease flare was noted in 6 of 7 patients after dose reduction or discontinuation of methotrexate. Biologic agents were used more frequently in patients with elbow involvement, than without it (45% vs. 23%, $p$=0.054).

**Conclusion**

Although elbow joint involvement is relatively uncommon, it may be represented as a "high-risk joint". Larger studies are warranted to confirm these findings.

**Disclosure of interest**

None declared.

## P227

### Methotrexate discontinuation in patients with non-systemic juvenile idiopathic arthritis

R. Raupov[*,1,2], S. Vissarionov[1], M. Orlova[1,3], M. Dubko[1,3], M. Kostik[3]
[1]H. Turner National Medical Research Center for Children's Orthopedics and Trauma Surgery; [2]BELOOSTROV Clinic of High Technologies; [3]Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation

**Correspondence:** R. Raupov
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**Introduction**
Remission or inactive disease is the main target of the treatment in patients with juvenile idiopathic arthritis (JIA). Methotrexate is the most commonly used basic medication in JIA, and issues of dosing regimen modification in patients in prolonged remission remain relevant.
**Objectives**
To study the outcomes of juvenile idiopathic arthritis in patients with non-systemic JIA categories treated with methotrexate and to identify predictors of clinical remission off-medication (CROM).
**Methods**
The retrospective study included 155 patients with non-systemic JIA categories who received methotrexate with observation duration of at least 12 months. Baseline clinical-laboratory characteristics, reasons for methotrexate discontinuation and predictors of CROM were analyzed.
**Results**
Clinical remission on medication was achieved in 69/155 (44.5%) patients. 71 of 155 patients discontinued methotrexate: 45 (62.5%) due to remission, 13 (18.1%) due to intolerance, 3 (4.2%) due to elevated liver enzyme levels, and in 9 cases (12.5%), parents discontinued therapy due to their own decision. In patients who discontinued methotrexate due to remission, the comparative analysis of baseline characteristics was done compared patients who subsequently developed flare (25/35; 71.4%) or not (10/35; 28.6%) after methotrexate withdrawal at last follow-up, excluding patients on biologic DMARDs at discontinuation ($n$=4) and those with <18 months follow-up post-discontinuation ($n$=6). Median time to flare was 9 (4;20) months. Both groups were comparable by treatment profile, post-discontinuation follow-up duration, and baseline clinical-laboratory characteristics. Patients who flared more often received intra-articular corticosteroids at disease onset (0 vs. 60%, $p < 0.001$) and had shorter methotrexate duration after remission (44 vs. 34 months, $p$=0.079) compared whose, without flare. Most patients with flare (85%) required restart of methotrexate, effective in 75%; only 25% needed subsequent biologic DMARDs.

**Table 1 (Abstract P227)**

| Parameter | No flare after MTX withdrawn ($n$=10) | Flare after MTX withdrawn ($n$=25) | $p$-value |
|---|---|---|---|
| Age of onset, years | 4.1 (2.4; 6.3) | 3.0 (2.2; 5.1) | 0.913 |
| Oligoarthicular, n (%) | 8 (80) | 15 (60) | 0.441 |
| Polyarthicular (RF-negative), n (%) | 1 (10) | 7 (28) | |
| ERA, n (%) | 1 (10) | 1 (4) | |
| Psoriatic, n (%) | 0 (0) | 2 (8) | |
| CRP increase at disease onset, n (%) | 0 (0) | 4 (16) | 0.289 |
| ANA- positivity, n (%) | 6 (60) | 20 (80) | 0.393 |
| HLA-B27-positivity, n(%) | 1 (10) | 3 (12) | 1.0 |
| Intraarticular corticosteroids, n(%) | 0 (0) | 15 (60) | <0.001 |
| Time to methotrexate discontinuation, months | 44.2 (35.5; 57.7) | 34.5 (27.8; 42.4) | 0.079 |
| Duration of follow-up after methotrexate discontinuation, months | 25.5 (23.1; 28.6) | 30.4 (22.8; 61.3) | 0.107 |

**Conclusion**
Intra-articular corticosteroids at disease onset and shorter methotrexate were flare risk predictors after methotrexate discontinuation in JIA patients, achieved the remission.
**Disclosure of interest**
None declared.

## P228

### Biologic switching patterns and treatment outcomes in refractory polyarticular juvenile idiopathic arthritis: a multicenter retrospective study

S. Atamyıldız Uçar[1,*], A. Uzun Bektaş[2], E. Tunce[1], N. Kara Çanlıoğlu[1], S. N. Tığrak[3], B. Öksel[4], B. Acun[5], T. Kasap[6], E. Küçük[7], G. Özomay Baykal[8], K. Öztürk[7], S. A. Bakkaloğlu[5], N. Şahin[4], S. Şener[9], S. Türkuçar[3], S. Yüksel[6], S. E. Ünsal[2], B. Sözeri[1]
[1]Pediatric Rheumatology, Umraniye Training and Research Hospital, University of Health Sciences, Istanbul; [2]Pediatric Rheumatology, Dokuz Eylül University Faculty of Medicine, Izmir; [3]Pediatric Rheumatology, Pamukkale University, Faculty of Medicine, Denizli; [4]Pediatric Rheumatology, Kocaeli University Faculty of Medicine, Kocaeli; [5]Pediatric Rheumatology, Gazi University Faculty of Medicine, Ankara; [6]Pediatric Rheumatology, Çanakkale Onsekiz Mart University Faculty of Medicine, Çanakkale; [7]Pediatric Rheumatology, Istanbul Medeniyet University Faculty of Medicine, Istanbul; [8]Pediatric Rheumatology, Mardin Training and Research Hospital, Mardin; [9]Pediatric Rheumatology, Adana City Training and Research Hospital, Adana, Türkiye
**Correspondence:** S. Atamyıldız Uçar
*Pediatric Rheumatology 2026*, **24(S1):**P228

**Introduction**
Polyarticular juvenile idiopathic arthritis (pJIA) is associated with a substantial risk of persistent disease activity and long-term functional impairment despite advances in biologic therapies.
**Objectives**
To evaluate the impact of primary and secondary non-response to the initial bDMARD on subsequent treatment outcomes in patients with pJIA requiring treatment with two or more bDMARDs.
**Methods**
This multicenter retrospective study included pJIA patients from 9 pediatric rheumatology centers in Türkiye who received at least two different bDMARDs between 2015 and 2026. Demographic, clinical, laboratory, and JADAS-27 data were retrospectively analyzed. Primary non-response was defined as failure to achieve clinically meaningful improvement within 3 months, whereas secondary non-response was defined as loss of efficacy after an initial response, both requiring treatment switch.
**Results**
A total of 74 patients were included, with a median age at diagnosis of 9.2 years; 85.1% were female and 83.8% were RF-negative. All patients initially received methotrexate as csDMARD therapy prior to biologic treatment initiation. The first-line bDMARD was predominantly an anti-TNF agent (86.5%), while 13.5% received IL-6 inhibitors. According to response to the initial bDMARD, 24.3% of patients achieved remission followed by relapse after discontinuation, 39.2% demonstrated primary non-response, and 36.5% developed secondary non-response. Failure of the first biologic was significantly associated with female gender ($p$=0.02), older age at diagnosis ($p$=0.01), and elbow involvement ($p$=0.033). Primary non-response was additionally associated with older age at diagnosis ($p$<0.001), RF positivity ($p$=0.012), and anti-CCP positivity ($p$=0.004). Among patients who relapsed after achieving remission, retreatment with a bDMARD targeting the same mechanism of action achieved an 80% response rate. In contrast, among patients with secondary non-response, switching to a different mechanism of action resulted in higher clinical response rates compared to within-class switching (53.8% vs. 28.6%), although statistical significance was not reached ($p$=0.08). Furthermore, patients with primary failure to the first bDMARD were more likely to experience primary failure to the second biologic compared with patients in the relapse group (41.4% vs. 16.7%). Overall, 23% of the cohort exhibited a resistant disease course despite exposure to at least two different bDMARD mechanisms.
**Conclusion**
Older age at diagnosis and the presence of anti-CCP antibodies appear to be important early predictors of primary non-response to initial bDMARDs. In cases of secondary non-response, switching to a different mechanism of action may provide improved clinical outcomes, whereas retreatment with the same mechanism may remain effective in patients who relapse after remission.

**Disclosure of interest**
None declared.

## P229

### High prevalence of neurodivergence in patients with JIA – a single centre experience

S. Melo Gomes[1,*], M. M. Ahmad[1], D. Adams[1], A. Ruiz[2], S. Bhatt[2], R. Florida James[2], A. Burdass[2], A. Orchard[2], V. Shivamurthy[1]
[1]Paediatric Rheumatology; [2]Guys and St Thomas NHS Foundation Trust, London, UK
**Correspondence:** S. Melo Gomes
*Pediatric Rheumatology 2026*, **24(S1):**P229

**Introduction**
Diagnosis of neurodivergence, including autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD), have been increasing in prevalence in the last few years, with reported 2.9% of children in England having an autism diagnosis. There is a well-known association between autoimmune conditions and neurodiversity, however, whilst an increased prevalence of neurodiversity in patients with juvenile idiopathic arthritis (JIA) has been suggested, existing data are limited.
**Objectives**
Assess the prevalence of neurodivergence diagnoses in a cohort of paediatric patients with JIA and its implications in patient care.
**Methods**
Patients followed in the paediatric rheumatology department during a 15month period (Jan25-Mar26) with a diagnosis of JIA and ASD/ ADHD were identified using the slicer dicer tool on Epic. Data collected included demographics, age at diagnosis, jia subtype, neurodivergence diagnosis, lab results, treatment, psychological/educational support, comorbidities and family history. Graphpad prism was used for statistical analysis.
**Results**
645 patients with JIA were identified (F-63.6%/M-36.4%; mean age 12y(5-18y)). Of these, 44 (28 F/16 M;6.82%) had a confirmed diagnosis of ASD and/or ADHD (ASD 43 (6.6%), ADHD 21 (3.2%)), considerably higher than in the general population ($p$=0.0002). An additional 15 patients (11 F/4 M) had autistic traits, 8 of which (53%) are currently awaiting or undergoing assessment, potentially increasing the prevalence of neurodivergence in this cohort to 9.1%. In the confirmed cases, 22(50%) had a JIA diagnosis before the ASD/ADHD one; 16 (36.3%) had another associated mental health condition and 8(18.2%) were followed at children and adolescents' mental health services, whilst 12 others (27.3%) had psychological support in the department; 9 (20.4%) were on adhd medication, with two of them also on SSRIs. Fifteen patients (34%) had additional health comorbidities, 3 of which were chromosomal changes. In terms of education, 10 (22.7%) had an education, health and care plan in place and 6 (13.6%) were in specialised schools. Regarding the JIA diagnosis, the most common subtypes were oligoJIA (19;43.2%) and RF negative polyarthritis (15;34%); 27(61.4%) are currently on systemic medication, including biologics in 22. In 11 patients (25%) there was documented family history of neurodivergence and 9(20.4%) had family history of autoimmune conditions. Needle phobia was documented in 19(43.2%); 4/28 girls would only accept female staff and 10 patients (22.7%) had at least one medication change due to their neurodivergence.
**Conclusion**
This study demonstrated a significantly higher prevalence of neurodivergence in JIA than in the general population, including higher prevalence in females. These results highlight the need for a highly individualised, multi-disciplinary approach as well as for considerable restructuring and adaptation of health care resources.
**Disclosure of interest**
None declared.

## P230

### Association of glucocorticoid use with serious infection in non-systemic JIA: a nested case-control study in the CARRA registry

D. Rumsey[1], E. Robinson[2], C. Correll[3], A. Dennos[4], D. B. Horton[5], E. Weller[2], T. Beukelman[6,7,*] on behalf of CARRA Registry Investigators
[1]University of Alberta, Edmonton, Canada; [2]Boston Children's Hospital, Boston; [3]University of Minnesota, Minneapolis; [4]Duke University, Durham; [5]Rutgers University, New Brunswick; [6]CARRA, Washington, USA; [7]Adelaide University, Adelaide, Australia
**Correspondence:** T. Beukelman
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**Introduction**
Chronic systemic glucocorticoid (GC) use for the treatment of non-systemic juvenile idiopathic arthritis (nsJIA) is increasingly discouraged. The risk of serious infection associated with GC use in nsJIA is uncertain.
**Objectives**
To describe GC use in nsJIA and estimate the association between GC use and serious infection using data from the CARRA Registry.
**Methods**
We assessed GC use among nsJIA participants, including chronic use (defined as current GC use documented at 2 visits >90 days and <366 days apart). We then conducted a nested case-control study within the CARRA Registry. Cases were participants with nsJIA who developed a serious infection (requiring hospitalization or IV antimicrobials) while receiving methotrexate monotherapy or an approved biologic or targeted synthetic DMARD (b/tsDMARD). Each case was matched to 5 controls without prior serious infection, using risk-set sampling and time since initiation of current DMARD therapy as the time axis; thus, cases and controls had equivalent duration of current DMARD therapy. Additional matching factors included specific current DMARD treatment, time since last CARRA Registry visit, sex, and age. GC exposure was ascertained at the most recent visit prior to the index date (infection date for cases and matched time since initiation of current DMARD for controls). Physician global assessment, patient global assessment, disease duration, number of prior b/tsDMARDs, prior use of current DMARD (i.e., not naïve use), and JIA category were assessed as potential confounders. Associations between GC use and serious infection were estimated using multivariable logistic regression.
**Results**
Among all Registry participants with nsJIA, 39.5% had ever received systemic GC, including 2.9% with chronic oral GC use following Registry enrollment. We identified 97 cases with serious infection, and all were successfully matched to 5 controls (485 total) on all factors. Median age was 13 years and 75% were female. Median time from most recent Registry visit to index date was 92 days in both cases and controls. At the most recent visit prior to index date, current oral GC use was present in 8.2% of cases and 3.7% of controls (unadjusted OR 2.3; adjusted OR 2.2 [95% CI 0.9, 5.4]). No cases had intra-articular GC at their most recent visit prior to infection, compared to 0.8% of controls.
**Conclusion**
Use of systemic GC was common overall among participants with nsJIA, but chronic oral GC use was very uncommon. Current oral GC use was associated with approximately 2-fold higher odds of serious infection, although precision was limited. No association was observed between recent intra-articular GC and serious infection.
**Disclosure of interest**
None declared.

## P231

### Escalation to systemic treatment following intra-articular steroid injection in patients with juvenile idiopathic arthritis – prevalence and risk factors

A. Kalter[1], G. Amarilyo[2], E. zagagi Yohai[1], A. Atias[1], Y. Levinsky[2,*]
[1]Pediatric B; [2]Pediatric Rheumatology, Schneider Children's Medical Center of Israel, Petah Tikva, Israel
**Correspondence:** Y. Levinsky
*Pediatric Rheumatology 2026*, **24(S1):**P231

**Introduction**
According to current guidelines for the management of oligoarticular juvenile idiopathic arthritis (JIA), intra-articular steroid injection is recommended as first-line therapy. While this treatment is highly effective in the short term, a substantial proportion of children eventually require escalation to systemic therapy. Data regarding the prevalence of subsequent treatment escalation, as well as predictors identifiable at the time of injection, are limited.
**Objectives**
To determine the proportion of children who ultimately require systemic therapy and to identify risk factors associated with treatment escalation.

**Methods**
This retrospective study was conducted at a tertiary pediatric rheumatology center. The electronic medical record system was retrospectively reviewed for the years 2015–2025 to identify all JIA cases in which initial treatment consisted of intra-articular steroid injection. Demographic, clinical, and laboratory data were collected, including disease course, subsequent need for systemic therapy, and reasons for escalation. Patients who required systemic therapy were compared with those who did not using univariate and multivariate analyses. Cox proportional hazards regression was performed to assess factors associated with time to treatment escalation.
**Results**
A total of 85 patients were included, of whom 67 (78.8%) were female. Forty-eight patients (56.5%) required escaltion to systemic therapy during the disease course. Age and sex at the time of injection did not differ significantly between groups. Patients who required systemic therapy more frequently presented with symmetric arthritis [14 (29.2%) vs. 4 (10.8%); $P = 0.040$], ankle joint involvement [17 (35.4%) vs. 4 (10.8%); $P = 0.009$], and antinuclear antibody (ANA) positivity [40 (83.3%) vs. 21 (56.8%); $P = 0.007$]. They also had a higher number of involved joints at presentation (1.75 ± 0.93 vs. 1.24 ± 0.56; $P = 0.004$) and higher erythrocyte sedimentation rate (40 ± 27 vs. 29 ± 22 mm/h; $P = 0.040$). In multivariate analysis, ANA positivity was independently associated with escalation to systemic therapy.
**Conclusion**
Over half of patients required escalation to systemic therapy. Risk factors included symmetric arthritis, ankle involvement, higher number of joints at presentation, ANA positivity, and elevated ESR. Further studies are needed to determine whether initiation of systemic therapy as first-line treatment may be warranted in patients presenting with these risk factors.
**Disclosure of interest**
None declared.

## P232

### Prolonged fever as isolated symptom in heterozygous patient with missense variant rs104895094 in MEFV gene

M. Šenjug Perica[1,*], L. Bukovac[1], A. M. Pasini[1], M. Harjaček[1]
[1]Department of Paediatric Rheumatology, Children's Hospital Srebrnjak, Zagreb, Croatia
**Correspondence:** M. Šenjug Perica
*Pediatric Rheumatology 2026*, **24(S1):**P232

**Introduction**
Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease, although it remains rare in routine pediatric rheumatology practice. We present the case of a now 16-year-old girl with atypical prolonged febrile episodes beginning at the age of 8 years.
**Objectives**
The disease initially manifested as recurrent subfebrile episodes lasting 2–6 weeks following common respiratory infections affecting both the patient and other family members. During the first 2 years, episodes occurred intermittently with temperatures up to 38°C, spontaneous resolution, and no accompanying symptoms. Laboratory findings, inflammatory markers, and microbiological investigations were repeatedly normal. One episode was associated with elevated liver enzymes attributed to cytomegalovirus infection. During the third and fourth years, episodes became longer and were accompanied by headache and fatigue, with symptom-free summers. By the end of the fourth year, the patient developed continuous fever lasting 9 months. Extensive evaluation by infectious disease specialists and pediatric rheumatologists excluded infectious, malignant, and autoimmune etiologies. Ultrasound, radiographs, scintigraphy, and MRI studies were normal. Inflammatory markers, ANA, ENA, and tumor markers remained persistently negative. NSAIDs provided only temporary symptom relief. Eurofever scores did not support an autoinflammatory disease diagnosis, and serum amyloid A testing was unavailable. In the fifth year, the patient was referred to our institution. Despite persistent febrility requiring 3 times daily ibuprofen, repeated investigations remained normal.
**Methods**
Genetic testing revealed a heterozygous missense variant rs104895094 in the MEFV gene, causing substitution of lysine with arginine at codon 695 (p.Lys695Arg). Although diagnostic criteria for FMF were insufficient, colchicine therapy (0.5 mg twice daily) was introduced, resulting in complete remission. After 8 months, the patient developed pityriasis lichenoides chronica confirmed by histopathology, which resolved with topical therapy. Following 16 months of remission and normal serum amyloid A levels, colchicine was tapered to 0.5 mg daily. Two months later, prolonged fever and fatigue recurred after a respiratory infection. Infectious workup remained negative and inflammatory markers normal. Reintroduction of colchicine 0.5 mg twice daily again resulted in complete remission. The p.Lys695Arg variant has been reported in symptomatic and asymptomatic individuals and is considered a low-penetrance variant of uncertain significance. This case highlights the diagnostic challenges of atypical FMF presentations with prolonged fever, persistently normal inflammatory markers, and uncertain MEFV variants, while emphasizing the importance of clinical response to colchicine.
**Disclosure of interest**
None declared.

## P233

### PFAPA-like clinical presentation with atypical features leading to the diagnosis of a rare inborn error of immunity

M. Bizjak[1,*], M. Debeljak[2], N. Toplak[1,3]
[1]Department for Allergology, Rheumatology and Clinical Immunology; [2]Unit for special Laboratory Diagnostics, Children's Hospital, University Medical Centre Ljubljana; [3]University of Ljubljana, Faculty of Medicine, Ljubljana, Slovenia
**Correspondence:** M. Bizjak
*Pediatric Rheumatology 2026*, **24(S1):**P233

**Introduction**
Periodic fever, aphthous stomatitis, pharyngitis, and adenitis (PFAPA) syndrome is the most common autoinflammatory disease in children. It is considered a multifactorial disorder with a genetic predisposition rather than a classic monogenic disease. Genetic testing is usually reserved for patients with atypical clinical features that raise suspicion for hereditary periodic fever syndromes or other alternative diagnoses.
**Objectives**
To present a case with a PFAPA-like clinical presentation in which atypical features led to the diagnosis of a rare inborn error of immunity.
**Methods**
Clinical data were obtained from the hospital's electronic medical record system.
**Results**
A 3-year-old boy was referred to a rheumatology clinic due to monthly fever episodes starting at one year of age, coinciding with kindergarten attendance. Episodes lasted 3–5 days and were consistently associated with tonsillitis and cervical lymphadenopathy, without aphthous ulcers. He was frequently treated with antibiotics. Hepatosplenomegaly had been noted since 5 months of age; by age three, only splenomegaly persisted. A prior bone marrow examination was normal. The patient also had recurrent respiratory infections. At presentation, growth and development were normal, with tonsillar hypertrophy, small cervical lymph nodes, and mild splenomegaly. Immunological evaluation showed elevated B-cell counts (5.13 cells 109/L (normal 2-2.1 cells 109/L), 61.2% (normal 14-44%)) with normal immunoglobulins and complement. A next-generation sequencing (NGS) autoinflammatory panel was negative. Because of suspected PFAPA syndrome, methylprednisolone on demand was suggested and after adenotonsillectomy at 3.5 years, fever episodes resolved. However, splenomegaly progressed. At age six, he had an episode of fever with gastrointestinal symptoms, massive splenomegaly, pancytopenia, and elevated inflammatory markers. No infectious cause was identified. Autoimmune lymphoproliferation syndrome (ALPS) was suspected, but further laboratory tests were inconclusive. Extended genetic testing using an NGS immunodeficiency panel revealed a gain-of-function mutation in *CARD11*, consistent with BENTA (B-cell expansion with NF-κB and T-cell anergy) disease.
**Conclusion**
The presence of splenomegaly is atypical for PFAPA syndrome and should prompt evaluation for alternative diagnoses, including inborn errors of immunity.
**Disclosure of interest**
None declared.

## P234

### A particular case of bone pain: the importance of differential diagnosis

M. D'ambrosio[1,*]
[1]Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genoa, Genoa, Italy, Genova, Italy
**Correspondence:** M. D'ambrosio
*Pediatric Rheumatology 2026*, **24(S1):**P234

**Introduction**
Chronic recurrent multifocal osteomyelitis (CRMO) is an inflammatory, non-infectious condition that affects the bones in children of all ages. The symptoms of chronic recurrent multifocal osteomyelitis vary. It may present insidiously, with recurrent or persistent low-grade fever, fatigue, and diffuse, intermittent bone pain; therefore the diagnosis might be challenging. A 4-year-and-9-month-old girl was referred to us for suspected CRMO. Her initial symptoms included limping, back pain, fever, and bilateral intra-articular effusions. An initial whole-body magnetic resonance imaging (MRI) scan revealed multiple areas of bone edema (involving pelvis, vertebrae, femurs, tibia, scapula, and sternum). A bone biopsy revealed an inflammatory infiltrate compatible with chronic osteomyelitis. Treatment with naproxen was initiated, and, given the vertebral involvement, cycles of bisphosphonates were also started, resulting in clinical improvement and normalization of the radiographic findings. Occasional episodes of pain persisted, responding to NSAIDs, and microcytosis remained evident on blood tests. Four years after the onset, the girl was readmitted due to pain unresponsive to NSAIDs. MRI revealed new areas of bone edema; furthermore, fever and pancytopenia developed after starting naproxen, so a transfusion was performed. A bone marrow aspiration revealed bone marrow aplasia and positivity for Parvovirus B19. Readmitted with persistent limping, an MRI showed areas of osteonecrosis. Suspecting an ischemic origin of the lesions, a peripheral blood smear was performed, revealing anisopoikilocytosis and target cells, while hemoglobin electrophoresis documented microdrepanocytosis with 65% HbS and increased HbA2. A new course of bisphosphonates was administered, providing pain relief, and treatment with hydroxyurea was initiated. Microdrepanocytosis is a form of hereditary hemoglobinopathy characterized by the coexistence of the sickle cell trait (HbS) and the thalassemia trait (usually beta-thalassemia). Clinically, this results in an intermediate phenotype that can sometimes remain masked, leading to nonspecific clinical manifestations and an incomplete radiological picture. In our case, the predominant presence of bone edema, the inflammatory infiltrate at histological level, the mild and inconsistent microcytic anemia, and the response to therapy with bisphosphonates and NSAIDs directed suspicion toward CRMO. Infection with Parvovirus B19, which is known to inhibit erythropoiesis, acted as a triggering event, leading to a vaso-occlusive crisis with consequent osteonecrosis in a previously subclinical hematologic condition.
**Objectives**
Through this case report we highlight the importance of considering hemoglobinopathies among the differential diagnoses in pediatric patients with persistent microcytosis and atypical bone lesions.
**Methods**
We present the case of a young girl with suspected chronic recurrent multifocal osteomyelitis (CRMO) who developed a severe vaso-occlusive crisis and osteonecrosis following infection with Parvovirus B19, leading to a diagnosis of compound sickle cell disease in heterozygotes with thalassaemia trait.
**Disclosure of interest**
None declared.

## P235

### Assessment of disease damage using the autoinflammatory disease damage index in paediatric familial Mediterranean fever: factors associated with damage

E. Esen[1,*], S. Ö. Çiçek[2], A. P. Kısaarslan[2], M. H. Poyrazoğlu[2]
[1]Pediatric Rheumatology, Bilkent City Hospital, Ankara; [2]Pediatric Rheumatology, Erciyes University, Kayseri, Türkiye
**Correspondence:** E. Esen
*Pediatric Rheumatology 2026*, **24(S1):**P235

**Introduction**
Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease in childhood, characterized by recurrent inflammatory attacks that may lead to cumulative organ damage over time. The Autoinflammatory Disease Damage Index (ADDI) is a standardized tool developed to assess irreversible damage in major autoinflammatory diseases; however, data on its use in paediatric FMF are limited.
**Objectives**
This study aimed to evaluate damage using ADDI and to identify factors associated with damage in children with FMF.
**Methods**
This retrospective study included paediatric FMF patients followed at a single tertiary centre.Cumulative damage was assessed using ADDI. To better evaluate factors associated with damage, patients with high-risk features such as M694V homozygosity and colchicine resistance were represented at a higher proportion.Patients were classified into two groups according to damage status (ADDI ≥1 vs. ADDI =0) and associated clinical and genetic factors were analysed.
**Results**
A total of 130 pediatric patients with FMF were included, of whom 49.2% were female.The median age at disease onset was 48 months(2–197), and the median age at diagnosis was 72 months (8–201).The median disease duration was 120 months(12–216). Median ADDI and ISSF scores were 0 (0–9) and 0 (0–6), respectively.M694V homozygous mutations were present in 62.3% of patients.Colchicine resistance was observed in 41.5% of patients, and 49.2% had received biologic therapy. Concomitant inflammatory diseases were present in 24.6% of the cohort.Damage (ADDI >0) was identified in 47.7% of patients.The most commonly affected domain was the musculoskeletal system (36.9%), followed by the developmental domain (19.2%). Renal damage was observed in 5.4% of patients, including amyloidosis in 2.3%.Patients with damage had significantly higher ISSF disease severity scores compared to those without damage (p=0.002). Additionally, patients with colchicine resistance had significantly higher ADDI scores than those without resistance (p<0.001).
**Conclusion**
In conclusion, damage was most commonly observed in the musculoskeletal and developmental domains. The presence of homozygous MEFV mutations, concomitant inflammatory diseases, and colchicine resistance were found to be significantly associated with damage. Higher disease severity scores also appeared to be related to increased damage accumulation. Early identification of high-risk patients and effective control of inflammation may contribute to reducing long-term complications in pediatric FMF.

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**Disclosure of interest**
None declared.

## P236

### A novel MVK variant in severe early-onset mevalonate kinase deficiency

M. Leonteva[1], A. Kozlova[1,*]
[1]Ilyinskaya Hospital, Krasnogorsk Urban District, Glukhovo, Russian Federation
**Correspondence:** A. Kozlova
*Pediatric Rheumatology 2026*, **24(S1):**P236

**Introduction**
Mevalonate kinase deficiency (MKD) is a rare autoinflammatory disease caused by pathogenic variants in the MVK gene. Clinical manifestations include recurrent fever, gastrointestinal symptoms, skin rash, lymphadenopathy, arthralgia, and marked inflammatory activity. Due to phenotypic variability, diagnosis is frequently delayed. Molecular testing is the key diagnostic tool. More than 300 MVK variants have been described, with p.(V377I) and p.(I268T) being the most common in European populations. Identification of rare variants expands the molecular spectrum of MKD and improves disease recognition.

**Objectives**
To describe a severe early-onset MKD case associated with a previously undescribed MVK variant confirmed by functional testing.
**Methods**
Clinical, laboratory, and molecular genetic data of a child with an autoinflammatory syndrome were analyzed. Targeted NGS panel testing, Sanger segregation analysis, and urinary mevalonic acid measurement were performed.
**Results**
Disease onset occurred at 2 weeks of age with rash, diarrhea with blood streaks, and recurrent fever. At 6 months, vaccination triggered fever up to 39°C with vomiting and severe diarrhea. Subsequently, monthly febrile attacks lasting 3–5 days developed, accompanied by gastrointestinal symptoms and high inflammatory activity. The patient was repeatedly hospitalized with suspected intestinal infection, enterocolitis, and sepsis. At 10 months, fever with maculopapular rash, edema of hands and feet followed by desquamation was interpreted as incomplete Kawasaki disease and treated with intravenous immunoglobulin; however, attacks persisted every 3–4 weeks. During flares, neutrophilic leukocytosis, thrombocytosis, and CRP elevation up to 141 mg/L were observed. Genetic testing identified two heterozygous MVK variants: c.83C>T p.(Ala28Val) and c.716T>G p.(Val239Gly). The A28V variant has not been previously described. The V239G variant has been reported at low frequency, while the clinical significance of both variants remains uncertain. Segregation analysis demonstrated inheritance of V239G from an asymptomatic father, while neither variant was detected in the mother. Urinary mevalonic acid was markedly elevated at 721.3 mmol/mol creatinine (normal 0.1–0.7), confirming MKD. Treatment with anakinra resulted in partial clinical improvement; persistent subfebrile episodes required escalation to canakinumab.
**Conclusion**
We describe a severe early-onset MKD case associated with a previously undescribed A28V variant in a compound heterozygous state with V239G. Functional confirmation supports the pathogenic relevance of the identified variants. This case highlights the importance of early molecular testing in children with recurrent fever and gastrointestinal manifestations and demonstrates the efficacy of IL-1 inhibition in MKD.

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**Disclosure of interest**
None declared.

## P237

**Evaluation of damage development in pediatric autoinflammatory diseases: an interim analysis of a single-center cohort**

M. O. Erkan[1,*], E. Cingoz[2], V. Cam[1], H. Ercan Emreol[1], O. Necipoglu Banak[1], H. D. Dara Kar[1], S. Ozkan[1], E. Sag[1], O. Basaran[1], Y. Bilginer[1], S. Ozen[1]
[1]Pediatric Rheumatology; [2]Pediatrics, Hacettepe University, Ankara, Türkiye
**Correspondence:** M. O. Erkan
*Pediatric Rheumatology 2026*, **24(S1):**P237

**Introduction**
Permanent damage is one of the key outcome measures determining long-term prognosis and quality of life in autoinflammatory diseases. However, data on the frequency, timing, and patterns of damage across different pediatric autoinflammatory diseases remain limited.
**Objectives**
This study aimed to evaluate the frequency of damage, time to first damage, and distribution of first damage domains in pediatric autoinflammatory diseases.
**Methods**
Pediatric patients with autoinflammatory diseases, including familial Mediterranean fever (FMF), hyper-IgD syndrome/mevalonate kinase deficiency (HIDS/MKD), and cryopyrin-associated periodic syndrome (CAPS), followed at a single center were retrospectively evaluated. Demographic, clinical, genetic, and treatment-related data were collected. Damage event rates were compared across disease groups. Time to first damage, defined as the first Autoinflammatory Disease Damage Index[1] (ADDI) score >0, was analyzed using Kaplan–Meier survival analysis and compared with the log-rank test. The distribution of first damage domains across disease groups was further explored using a Sankey diagram.
**Results**
The cohort included 79 patients with FMF (69.9%), 18 with HIDS/MKD (15.9%), and 16 with CAPS (14.2%). Damage, defined as an ADDI score >0, occurred significantly more often in HIDS/MKD (50.0%) and CAPS (43.8%) than in FMF (22.8%) ($\chi^2$=6.816, df=2, $p$=0.033). Kaplan–Meier analysis showed a significant difference in damage-free survival among disease groups (log-rank $\chi^2$=6.414, df=2, $p$=0.040). Median time to first damage was shortest in CAPS (1.99 years), followed by HIDS/MKD (2.97 years), and longest in FMF (4.05 years). Sankey-based mapping showed that growth failure was the most common first damage domain overall, while neurological damage was more prominent in CAPS and serosal and musculoskeletal damage were more frequent in FMF and HIDS/MKD.
**Conclusion**
This interim analysis shows that damage development differs significantly according to disease type in pediatric autoinflammatory diseases. Damage occurred more frequently and earlier in CAPS and HIDS/MKD than in FMF. These findings support the importance of early recognition of disease-specific damage patterns and structured long-term follow-up in children with autoinflammatory diseases.

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**Disclosure of interest**
None declared.

## P238

**Beyond juvenile idiopathic arthritis: Behçet-like autoinflammation associated with an IL6ST variant**

M. I. Menjívar-Chavarría[1,*], J. Sánchez-Manubens[2,3], L. Rodriguez[1], P. Llobet-Agulló[4,5]
[1]Rheumatology Pediatric Department, Hospital General de Granollers; [2]Clinical Head of the Pediatric Rheumatology Unit – Department of Pediatrics; [3]Research and Innovation Institute, Hospital Parc Taulí; [4]Clinical Head of the Pediatric Unit; [5]Clinical Head of the Allergy and Clinical Immunology Pediatric Department, Hospital General de Granollers, Barcelona, Spain
**Correspondence:** M. I. Menjívar-Chavarría
*Pediatric Rheumatology 2026*, **24(S1):**P238

**Introduction**
Variants affecting IL6ST, encoding the gp130 cytokine signal transducer, have recently been associated with immune dysregulation and autoinflammatory phenotypes.
**Objectives**
We report a child initially diagnosed with juvenile idiopathic arthritis (JIA) who evolved toward a Behçet-like autoinflammatory syndrome associated with an IL6ST variant.
**Methods**
Clinical, immunological, and genetic data were retrospectively reviewed.
**Results**
A 13-year-old boy was diagnosed at age 8 years with ANA-negative oligoarticular JIA characterized by recurrent right knee and ankle arthritis requiring methotrexate. Despite periods of remission, he experienced relapses. From age 12 years, he developed recurrent painful oral and genital aphthosis with residual scarring, associated with elevated serum amyloid A (318 mg/L), developmental impairment, and peculiar facies. HLA-B51 was negative and he did not fulfill classification criteria for Behçet disease. Autoinflammatory gene panel identified a

heterozygous IL6ST variant, c.1844 A> G p.(Gln615Arg), classified as a variant of uncertain significance. IL6ST encodes gp130, a shared signaling receptor involved in IL6-family cytokine pathways and STAT3 activation. Gain-of-function IL6ST variants have recently been linked to immunodeficiency with autoinflammation and dysmorphic facies (IMD94). Treatment with colchicine, corticosteroids, methotrexate, and adalimumab achieved rapid resolution of mucosal and articular manifestations, with normalization of inflammatory markers.

**Conclusion**

This case highlights the diagnostic overlap between JIA, Behçet-like disease, and monogenic autoinflammatory disorders. IL6ST variants should be considered in patients with evolving inflammatory phenotypes, mucosal disease, dysmorphic facies and immune dysregulation features.

**Disclosure of interest**

None declared.

## P239

### Delayed diagnosis and persistent healthcare burden in PFAPA: a population-based matched cohort study

M. Hamad Saied[1,2,*], B. Ghanim [2], R. Kasem Ali Sliman [2]
[1]Department of Pediatric Immunology and Infectious Diseases, Wilhelmina Children's Hospital, Utrecht, Netherlands; [2]Department of Pediatrics, Carmel Medical Center, Haifa, Israel
**Correspondence:** M. Hamad Saied
*Pediatric Rheumatology 2026*, **24(S1):**P239

**Introduction**

PFAPA (periodic fever, aphthous stomatitis, pharyngitis, and cervical adenitis) is the most common autoinflammatory syndrome in children. Despite its benign and self-limited course, diagnostic delay is common, and its impact on healthcare utilization remains poorly quantified.

**Objectives**

To evaluate healthcare utilization in children with PFAPA compared with matched controls, before and after diagnosis.

**Methods**

We conducted a population-based retrospective matched cohort study using electronic health records from Clalit Health Services (Northern District, Israel). Children ≤13 years diagnosed with PFAPA between 2011 and 2023 by pediatric rheumatologists were included. Each case was matched with 10 controls by age and sex. Healthcare utilization was assessed during the 2 years before and after diagnosis and included primary care visits, specialist consultations, laboratory tests, emergency department visits, hospitalizations, throat cultures, imaging studies, and antibiotic prescriptions. Negative binomial regression and conditional logistic regression models accounting for matching were used to estimate incidence rate ratios (IRRs) and odds ratios (ORs).

**Results**

The study included 120 children with PFAPA and 1,200 matched controls. Healthcare utilization was significantly higher among PFAPA patients across all domains both before and after diagnosis. In the prediagnosis period, PFAPA patients had higher rates of primary care visits (IRR 2.62, 95% CI 2.34–2.92), specialist consultations (IRR 2.58, 95% CI 2.10–3.18), and laboratory testing (IRR 3.15, 95% CI 2.38–3.83). Elevated odds were also observed for hospitalizations (OR 5.24, 95% CI 3.15–6.8), emergency department visits, antibiotic prescriptions (OR 6.8, 95% CI 4.5–10.3), and throat cultures (OR 11.3, 95% CI 7.5–16.9). Although utilization declined after diagnosis, it remained significantly higher than controls in adjusted analyses.

**Conclusion**

Children with PFAPA experience a substantial and persistent healthcare burden beginning well before diagnosis. Earlier recognition and structured management strategies may reduce unnecessary investigations, antibiotic use, and acute healthcare utilization, with important implications for healthcare efficiency and cost reduction.

**Disclosure of interest**

None declared.

## P240

### Not every ulcer is the same– case reports

D. Gensor[1,*], B. Slenker[1], L. Kapustova[1], E. J. Malicherova[1], A. Markocsy[1,2], M. Jesenak[1,2,3]
[1]Department of Pediatrics and Adolescent Medicine; [2]Institute of Clinical Immunology and Medical Genetics; [3]Department of Pneumology and Phthisiology, Jessenius Faculty of Medicine of Comenius University Bratislava, University Hospital in Martin, Slovakia, Martin, Slovakia
**Correspondence:** D. Gensor
*Pediatric Rheumatology 2026*, **24(S1):**P240

**Introduction**

Inborn errors of immunity constitute a heterogeneous group of genetically determined disorders that impair the function of specific components of the immune system. While classical primary immunodeficiencies are predominantly characterized by increased susceptibility to infections, a distinct subgroup is represented by autoinflammatory diseases. These conditions are defined by spontaneously occurring, non-infectious inflammation in the absence of autoantibodies and without typical features of adaptive immune activation.

**Objectives**

Autoinflammatory diseases arise from dysregulation of innate immune pathways, most commonly involving aberrant inflammasome activation, disturbances in cytokine signaling, or defects in programmed cell death. Clinically, they present with recurrent febrile episodes, mucocutaneous manifestations, arthritis, serositis, or neurological involvement. Early diagnosis supported by targeted genetic testing is essential for accurate disease classification and timely initiation of precision therapy, which significantly improves prognosis and quality of life.

**Methods**

In their presentation, the authors describe case reports of patients with confirmed monogenic autoinflammatory diseases, including HyperIgD syndrome and Familial Behçetlike autoinflammatory disease 3, outline key diagnostic challenges, and emphasize the importance of early interdisciplinary collaboration among pediatricians, immunologists, rheumatologists, and geneticists. Prompt recognition and initiation of targeted treatment are crucial for reducing morbidity and improving longterm outcomes.

**Keywords**

Autoinflammation, Monogenic diseases, hyper–IgD syndrome, Familial Behçet-like autoinflammatory disease 3, Interdisciplinary collaboration

**Disclosure of interest**

None declared.

## P242

### When the right drug is out of reach: ethical and clinical challenges in managing rare pediatric rheumatologic disorders in low– and middle–income countries

C. S. Bhat[1], N. Nayak[*,2]
[1]Paediatric Rheumatology, Rainbow Children Hospital, Bengaluru, India; [2]Paediatric Rheumatology, Bristol Royal Children Hospital, bristol, UK
**Correspondence:** N. Nayak
*Pediatric Rheumatology 2026*, **24(S1):**P242

**Introduction**

Rare rheumatologic conditions such as Complement Hyperactivation, Angiopathic Thrombosis and Protein-Losing Enteropathy (CHAPLE syndrome), and Hyperimmunoglobulin D Syndrome (HIDS) pose significant diagnostic and therapeutic challenges.(1) These diseases demand early and precise initiation of targeted biologic therapies. However, in low- and middle-income countries (LMICs), access to these life-saving treatments is severely limited. Drugs such as pozelimab (anti-C5 monoclonal antibody) and canakinumab (IL-1β inhibitor) offer disease-specific efficacy, yet are often inaccessible.(2) More commonly available alternatives like etanercept and anakinra provide only partial or non-specific immunomodulation, often resulting in suboptimal outcomes. In India, acquiring the appropriate targeted biologic remains a formidable challenge.

**Objectives**
To highlight the clinical and ethical challenges of managing rare autoinflammatory diseases in India when ideal biologics are unavailable or unaffordable, and to examine the consequences of using less effective substitutes.
**Methods**
A retrospective analysis was conducted on four pediatric patients—one with genetically confirmed CHAPLE syndrome and three with genetically confirmed HIDS. We evaluated clinical presentation, treatment strategies, drug procurement pathways, cost implications, and outcomes.
**Results**
The CHAPLE patient initially received steroids and etanercept with minimal clinical benefit. Upon genetic diagnosis, pozelimab was identified as the appropriate therapy and was acquired through a compassionate-use program enabled by international collaboration. Following treatment, the patient exhibited marked clinical improvement within two months. Of the three HIDS patients, one child was from India and two were from the Maldives. The Indian patient was managed with etanercept due to unavailability of canakinumab. The disease remained refractory, and the patient succumbed within a year. In contrast, the two Maldivian children received canakinumab after genetic confirmation—one transitioned from anakinra, and the other was started promptly post-diagnosis. Both showed significant clinical improvement and achieved remission, with costs covered under the Maldivian national health insurance program.
**Conclusion**
For rare pediatric rheumatologic disorders, access to the correct biologic is essential—not optional. Delayed or inappropriate therapy exacerbates morbidity, increases long-term costs, and undermines trust. LMICs like India require expedited drug approval processes, equitable access schemes, and enhanced international partnerships to address the stark disparities in rare disease care.

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**Disclosure of interest**

None declared.

## P243

### Real–world comparison of canakinumab therapy in biologic–naive and previously anti–interleukin–1–exposed pediatric Familial Mediterranean Fever patients

N. Kara Çanlıoğlu[1,*], S. Atamyıldız Uçar[1], E. Tunce[1], H. Öztürk Aydın[1], B. Sözeri[1]
[1]Department of Pediatric Rheumatology, Umraniye Training and Research Hospital, İstanbul, Türkiye
**Correspondence:** N. Kara Çanlıoğlu
*Pediatric Rheumatology 2026*, **24(S1):**P243

**Introduction**
Familial Mediterranean fever (FMF) is the most common autoinflammatory disease. Although colchicine remains the cornerstone of treatment, 5-10% of patients require biologic therapy due to resistance or intolerance [1]. Canakinumab is widely used in these patients; however, comparative real-world data regarding primary and secondary canakinumab use are limited.
**Objectives**
To compare clinical characteristics, flare patterns, and long-term outcomes of primary versus secondary canakinumab therapy in pediatric FMF patients.
**Methods**
This retrospective cohort study included 95 pediatric FMF patients receiving canakinumab. Patients were classified as primary ($n$=48) or secondary canakinumab users after prior anakinra exposure ($n$=47). Clinical characteristics, flare patterns, and dose intervals were evaluated. Flare-free survival was analyzed using Kaplan–Meier analysis and the log-rank test.
**Results**
Median age at evaluation was significantly higher in the primary canakinumab group than in the secondary group [227.41 (194.67–243.19) vs. 187.08 (145.86–229.52) months, $p$=0.002]. Baseline disease severity scores were similar between groups [median PRAS: 8 (7–11) in both groups, $p$=0.857]. Overall flare frequency was comparable between primary and secondary canakinumab groups (50% vs. 46.8%, $p$=0.756). However, full-dose flares were numerically less frequent in the primary group (3 vs. 8 patients, $p$=0.101). Median time to first flare was longer in patients receiving primary canakinumab [14 (10–20.5) vs. 10 (5–19) months, $p$=0.061]. Most flares occurred during dose spacing, particularly at 6–8 week dosing intervals. Kaplan–Meier analysis showed longer flare-free survival in the primary canakinumab group (35 vs. 19 months), without statistical significance (log-rank $p$=0.087). Longer prior anakinra duration (>3 months) was associated with poorer flare-free survival (HR 1.87, 95% CI 1.01–3.47, $p$=0.046).
**Conclusion**
Patients receiving primary canakinumab tended to have delayed flare occurrence and longer flare-free survival. Longer prior anakinra exposure may be associated with less favorable long-term canakinumab outcomes. Although anti-drug antibodies against anakinra are generally considered transient, persistent immunogenic effects after prolonged exposure cannot be completely excluded [2]. Larger multicenter studies evaluating anti-drug antibodies are needed to better clarify their impact on long-term canakinumab outcomes.

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**Disclosure of interest**
None declared.

## P244

### Diagnostic and therapeutic challenges in Aicardi-Goutières syndrome (AGS): single center experience

Z. Nesterenko[1,*], V. Burlakov[1], D. Pershin[1], M. Fadeeva[1], E. Raykina[2], I. Mersiyanova[2], V. Brilliantova[2], N. Kuzmenko[1], A. Mukhina[1], Y. Rodina[1], A. Shcherbina[1]
[1]Immunology; [2]Laboratory of Molecular Biology, Dmitry Rogachev National Medical Research Center of Pediatric Hematology, Oncology and Immunology, Moscow, Russian Federation
**Correspondence:** Z. Nesterenko
*Pediatric Rheumatology 2026*, **24(S1):**P244

**Introduction**
AGS is a group of monogenic disorders with a hallmark of enhanced type I interferon production. It typically presents with severe early-onset neurological manifestations and a TORCH-like syndrome, posing significant diagnostic and therapeutic challenges.
**Objectives**
To analyze clinical, genetic, and treatment response characteristics in a cohort of patients with AGS.
**Methods**
We retrospectively analyzed data of 17 patients (12 males) with AGS treated at our Center between 2014 and 2026. Genetic testing by NGS revealed pathogenic variants in the following genes: *TREX1* ($n$=4), *RNASEH2B* ($n$=5), *RNASEH2A* ($n$=2), *IFIH1* ($n$=4), *ADAR* ($n$=2). JAK inhibitors (JAKinibs) were used as a monotherapy in 5 patients, or in combination with other agents (anifrolumab, anti-IL-6, anti-IL-17) in 10 patients. SIGLEC1 (CD169) expression on monocytes as a biomarker of type I IFN-signaling activity was serially assessed. Spasticity was assessed using the Modified Ashworth Scale (MAS).

**Results**
The median age at symptoms onset was 0.1 years (range 0.0 –1.0). The median age at diagnosis was 2.0 years (range 1.0–10.1), with a mean diagnostic delay of 2.8 years. Neurological manifestations were the predominant clinical feature in 16/17 patients, followed by fever, vasculitis-like skin involvement, and other systemic manifestations. JAKinib monotherapy achieved clinical and laboratory control of autoinflammatory/autoimmune manifestations in all 5 treated patients, without significant improvement of pre-existing neurological deficits. Despite clinical stabilization and absence of overt disease progression, 4 patients receiving JAKinib as monotherapy and 2 patients treated with JAKinibs in combination with other agents (anti-IL-6, anti-IL-17) demonstrated persistently elevated SIGLEC1 expression: median index 231.5 (range 17.0–821.0, reference range ≤25). Combination therapy with JAKinibs and anifrolumab controlled autoinflammatory/autoimmune manifestations in 4 of 5 patients and resulted in neurological improvement in 1/5 (MAS score improved from 3 to 1+). Addition of anifrolumab in 5 cases was associated with marked normalization of SIGLEC1 levels: median index was 12.0 (range 4.7–15.0), with normalization observed within one day after infusion in one patient.
**Conclusion**
AGS is characterized by early-onset severe neurological involvement and marked genetic heterogeneity. SIGLEC1 (CD169) represents a reliable biomarker for disease monitoring, while JAK inhibitors and anifrolumab may contribute to halting disease progression.
**Disclosure of interest**
None declared.

## P245

### Thalidomide-responsive A20 haploinsufficiency disease in two siblings

N. F. Andrews[1,*], C. Poli[2,3], J. De la Cruz[2], A. Borzutzky[4,5]
[1]Pediatric Immunology, Allergy and Rheumatology, Pontificia Universidad Católica de Chile; [2]Program of Immunogenetics and Translational Immunology, Institute of Science and Innovation in Medicine, Faculty of Medicine, Clínica Alemana Universidad del Desarrollo, Santiago, Chile; [3]mmunology and Rheumatology Unit, Hospital Roberto del Río, Santiago, Chile; [4]Department of Pediatric Infectious Diseases and Immunology School of Medicine, Pontificia Universidad Católica de Chile; [5]Red de Salud UC CHRISTUS, Santiago, Chile
**Correspondence:** N. F. Andrews
*Pediatric Rheumatology 2026*, **24(S1):**P245

**Introduction**
Haploinsufficiency of A20, caused by heterozygous loss-of-function variants in TNFAIP3, is a monogenic autoinflammatory disorder that may mimic Behçet disease, inflammatory bowel disease or coeliac disease. Optimal treatment remains undefined, particularly in children with severe recurrent mucosal ulceration.
**Objectives**
Describe two siblings with familial A20 haploinsufficiency presenting as early-onset Behçet-like disease.
and marked clinical response to thalidomide.
**Methods**
We retrospectively reviewed clinical, serological, immunological, genetic and treatment-response data from two brothers with recurrent fever and oral, perianal and/or genital ulceration. Immune profiling included CXCL9, CXCL10, total IL-18 and interferon-stimulated gene score; ISG values ≥1.65 were considered elevated.
**Results**
The index case presented from 8 months with recurrent fever and oral/perianal ulcers. From 3 years, flares increased to every 2 weeks, with fever, oral, genital and perianal ulceration, diarrhoea and elevated inflammatory markers. Crohn disease and ocular involvement were excluded; faecal calprotectin was mildly elevated and coeliac antibodies were detected. Immune profiling showed elevated CXCL9, mildly elevated IL-18, normal CXCL10 and non-elevated ISG score. Disease persisted despite colchicine and corticosteroids, whereas thalidomide 25 mg/day induced complete resolution of fever and mucosal ulcers, maintained at 3 months. **His 13-year-old brother had recurrent severe oral ulcers and febrile flares from 6 years of age. Corticosteroids provided transient benefit and colchicine was poorly tolerated. Dermatology initiated thalidomide 50 mg three times weekly for refractory oral ulceration, achieving sustained stability for 3 years with control of mucosal and systemic flares. Coeliac disease-associated antibodies were detected. Immune profiling showed markedly elevated CXCL9, normal CXCL10 and IL-18, and elevated ISG score. Both brothers carried the same novel likely pathogenic heterozygousTNFAIP3frameshift variant, p.Arg502Serfs*195, confirming familial A20 haploinsufficiency. Their mother's childhood-onset oral/genital and oesophageal ulcers with Hashimoto thyroiditis supported autosomal dominant inheritance; her immune profile showed elevated CXCL9 with otherwise normal biomarkers.**
**Conclusion**
These cases support familial A20 haploinsufficiency as a monogenic Behçet-like disorder and suggest thalidomide as a potential steroid-sparing option for refractory mucosal and inflammatory flares.

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**Disclosure of interest**
None declared.

## P246

### Recurrent autoinflammatory arthritis mimicking juvenile idiopathic arthritis in EPCAM–associated tufting enteropathy

G. Avcı Demirciler[1], N. Gürbüz[1,*], G. Kaymak [1], E. Gülyiğit[1], T. Çetin Arıkan[1], A. H. Kamiloglu[1], S. Yücel [1], T. Turkut Tan[2], K. Çakmakkaya[3], E. Işık[2], A. Berdeli[1], G. Aksu[1]
[1]Pediatric Rheumatology; [2]Pediatric Genetic; [3]Pediatric Gastroenterology, Ege University, İzmir, Türkiye
**Correspondence:** N. Gürbüz
*Pediatric Rheumatology 2026*, **24(S1):**P246

**Introduction**
Congenital tufting enteropathy is a rare autosomal recessive enteropathy characterized by early-onset chronic diarrhea and malnutrition. Rheumatologic inflammatory manifestations have rarely been reported. We present a patient with EPCAM-associated tufting enteropathy accompanied by recurrent autoinflammatory attacks.
**Objectives**
A 17-year-old boy was followed because of chronic diarrhea, malnutrition, IGF-1 deficiency and short stature. Diarrheal symptoms had been present since infancy and worsened after complementary feeding. His parents were first-degree cousins, and his younger sister had died in early childhood because of chronic diarrhea of unknown etiology. The patient presented with recurrent episodes of ankle swelling, pain, warmth and erythematous skin lesions. Similar attacks had occurred since childhood and became monthly during the last year, lasting 2–3 days and resolving spontaneously. Fever occasionally accompanied the attacks. Morning stiffness lasting approximately 3 h was reported during attack periods. NSAID treatment was ineffective. Physical examination revealed coarse facial appearance, hypodontia, bilateral shortening of the fourth metacarpals, clinodactyly, broad thumbs and halluces, pes cavus and digital clubbing. Bilateral ankle swelling and tenderness were present. Non-raised erythematous macular lesions with a confluent pattern were observed over the dorsum of the feet and pretibial areas. Laboratory investigations demonstrated CRP 70 mg/L, ESR 36 mm/h, fibrinogen 449 mg/dL and serum amyloid A 367 mg/L. ANA, anti-dsDNA, ANCA and HLA-B27 were negative. Musculoskeletal ultrasonography demonstrated bilateral ankle synovitis together with subcutaneous edema and periarticular inflammatory changes. Skeletal survey revealed bilateral shortening of the fourth metacarpals and metatarsals. Because of recurrent inflammatory attacks and enteropathy, sulfasalazine treatment was initiated, resulting in rapid improvement in diarrhea, rash and ankle inflammation. Clinical exome sequencing later identified a homozygous *EPCAM* variant, c.346T> C (p.Cys116Arg) (NM_002354.3; hg38 chr2:47373969 T> C), was identified by clinical exome sequencing in the proband. Segregation analysis demonstrated that both unaffected parents were heterozygous carriers of the variant, consistent with autosomal recessive inheritance.

**Methods**
Although tufting enteropathy is primarily characterized by chronic diarrhea, accompanying autoinflammatory manifestations have rarely been described. In our patient, recurrent ankle arthritis, prolonged morning stiffness, transient erythematous skin lesions and markedly elevated serum amyloid A levels suggested an autoinflammatory phenotype. This case highlights that EPCAM-associated tufting enteropathy may present with recurrent autoinflammatory arthritis mimicking juvenile idiopathic arthritis.

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**Disclosure of interest**
None declared.

## P248

**Persistent fatigue in pediatric familial Mediterranean fever despite disease control: a cross-sectional study of the clinical and psychosocial burden**

Ö. Taş Aslan[1,*], F. Aydın[2], D. Öztürk[3], O. Bahçeci[2], D. Sarısoy[2], E. Erorhan[2], B. Çelikel Acar[3], Z. B. Özçakar[2]
[1]Aydın City Hospital, Aydın; [2]Ankara University Faculty of Medicine; [3]Ankara City Hospital, Ankara, Türkiye
**Correspondence:** Ö. Taş Aslan
*Pediatric Rheumatology 2026*, **24(S1):**P248

**Introduction**
Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease and requires lifelong treatment, most commonly with colchicine. In colchicine-resistant or intolerant patients, anti-interleukin-1 (anti-IL-1) agents are increasingly used. Chronic disease burden, recurrent attacks, and long-term treatment may negatively affect psychological well-being and contribute to fatigue and anxiety in children with FMF. However, data comparing fatigue and anxiety between paediatric FMF patients receiving anti-IL-1 therapy and those treated with colchicine alone are limited.
**Objectives**
This study aimed to evaluate and compare fatigue and anxiety levels in paediatric FMF patients receiving anti-IL-1 agents in addition to colchicine versus colchicine alone, and to identify potential demographic, clinical, and laboratory factors associated with fatigue and anxiety.
**Methods**
In this cross-sectional study, 73 FMF patients aged 8–18 years were enrolled from two referral centers. Group 1 included patients receiving colchicine only ($n$=45), and Group 2 included patients treated with colchicine plus anti–IL-1 agents ($n$=28). Fatigue was assessed using the Checklist Individual Strength (CIS), and anxiety was evaluated using the Revised Child Anxiety and Depression Scale–Child Version (RCADS-CV). Disease severity was measured using the International Severity Scoring System for FMF (ISSF). Outcomes were compared between groups and with healthy controls for fatigue group and a comparison was also made using the scoring system developed based on healthy children in the anxiety group.
**Results**
Both groups demonstrated significant reductions in ISSF scores after treatment ($p$<0.001). Fatigue was highly prevalent in the colchicine (91.1%) and anti–IL-1 (96.4%) groups, with no significant difference between them ($p$>0.05). However, fatigue scores were significantly higher in both FMF groups compared to healthy controls ($p$<0.001). In multivariate logistic regression analysis, sex remained independently associated with fatigue ($p$=0.011). Anxiety was identified in 15.5% of patients, with similar rates across groups ($p$>0.05).
**Conclusion**
Despite improved disease control, fatigue remains highly prevalent in paediatric FMF patients. Anxiety levels were comparable between treatment groups. These findings highlight the importance of integrated psychosocial and rehabilitative strategies in FMF management.
**Disclosure of interest**
None declared.

## P249

**Autoinflammatory features and autoimmune markers in pediatric-onset multiple sclerosis: a rheumatologic perspective**

H. H. Ozguner Kucuk[1,*], I. H. Akbeyaz[2], G. Kavrul Kayaalp[1], G. Thomas[2], O. Unver[2], D. Turkdogan[2], O. Koker[1]
[1]Division of Pediatric Rheumatology; [2]Division of Pediatric Neurology, Marmara University Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** H. H. Ozguner Kucuk
*Pediatric Rheumatology 2026*, **24(S1):**P249

**Introduction**
Emerging evidence suggests a complex interplay between autoinflammatory and autoimmune pathways in the pathogenesis of multiple sclerosis (MS), extending beyond classical demyelinating mechanisms. However, the clinical relevance of these immune dysregulation patterns in pediatric-onset MS remains insufficiently characterized.
**Objectives**
To investigate the frequency of autoinflammatory manifestations, autoimmune serological markers, and rheumatic diseases in patients with pediatric-onset MS, and to evaluate their potential associations with clinical and disease-related characteristics.
**Methods**
This single-center observational cohort study included 50 patients diagnosed with pediatric-onset MS. Demographic and clinical characteristics, magnetic resonance imaging findings, Expanded Disability Status Scale (EDSS) scores, autoinflammatory manifestations, and personal and family histories of rheumatic diseases were retrospectively evaluated. Laboratory assessments included C-reactive protein, erythrocyte sedimentation rate, oligoclonal bands (OCB), antinuclear antibody (ANA), and extractable nuclear antigen (ENA) antibodies. Associations between autoinflammatory features and disease-related parameters, including attack frequency, disease duration, and disability status, were analyzed.
**Results**
Among the patients, 72% were female, with a mean age of 18.5±2.7 years. The median age at diagnosis was 16 years (IQR: 14–17), and the median disease duration was 31.5 months (IQR:16.25–51.75). Sensory symptoms were the most frequent presenting manifestation (46%). At disease onset, 82% of patients had no systemic manifestations; recurrent fever and articular symptoms were the most commonly reported systemic findings (6% each). Clinical features suggestive of an autoinflammatory phenotype were identified in 20% of patients. A history of PFAPA syndrome during childhood was present in 12%, prior tonsillectomy in 14%, and acute rheumatic fever in 2%. MEFV gene analysis was performed in four patients. Family history revealed MS in 22%, ankylosing spondylitis in 10%, and familial Mediterranean fever (FMF), rheumatoid arthritis, and uveitis in 6% each. During follow-up, one patient developed systemic lupus erythematosus. No statistically significant associations were identified between autoinflammatory phenotype and attack frequency, EDSS score, or disease duration (p>0.05). ANA positivity was detected in 34% of patients, whereas ENA positivity was rare.
**Conclusion**
Autoinflammatory manifestations and autoimmune serological markers were observed at a considerable frequency in pediatric-onset MS, supporting the concept of broader immune dysregulation beyond conventional demyelinating processes. Although these immune-related features were not associated with disease severity or disability in the present cohort, the relatively high prevalence of rheumatologic diseases within family histories underscores the importance of rheumatologic awareness in the multidisciplinary evaluation of these patients. Larger multicenter studies are warranted to further elucidate the immunogenetic and pathophysiological links between pediatric-onset MS and autoinflammatory pathways.

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**Disclosure of interest**
None declared.

## P250

### NLRP12 variants in pediatric patients: a single–center experience

O. Necipoglu Banak[1,*], O. Basaran[1], Y. Bayindir[1], H. Ercan Emreol[1], V. Cam[1], D. Unal[1], M. O. Erkan[1], H. D. Dara Kar[1], S. Ozkan[1], E. Sag[1], Y. Bilginer[1], S. Ozen[1]
[1]Pediatric Rheumatology, Hacettepe University, Ankara, Türkiye
**Correspondence:** Ozlem Necipoglu Banak
*Pediatric Rheumatology 2026*, **24(S1):**P250

**Introduction**
Nucleotide-binding domain leucine-rich repeat-containing receptor family pyrin domain-containing 12 (NLRP12) variants have been associated with autoinflammatory phenotypes; however, their clinical significance may be difficult to interpret because reported variants range from pathogenic to variants of uncertain or conflicting significance.
**Objectives**
To describe the clinical, laboratory, genetic and treatment characteristics of pediatric patients with NLRP12 variants followed at a tertiary pediatric rheumatology center.
**Methods**
We retrospectively reviewed patients with identified NLRP12 variants who were evaluated at a single pediatric rheumatology center. Demographic data, age at symptom onset and diagnosis, clinical manifestations, acute-phase reactants, immunologic findings, genetic results, treatments and clinical outcomes were extracted from medical records and analyzed descriptively.
**Results**
Nine patients with NLRP12 variants were included. Seven patients were male (77.8%). The median age at first symptom was 2.4 years and the median age at diagnosis was 6.9 years. Recurrent fever was present in 6 patients (66.7%), while skin findings were observed in 4 (44.4%). Musculoskeletal involvement was frequent, affecting 6 patients (66.7%), mainly as arthralgia and/or arthritis. Gastrointestinal findings were the most common systemic manifestation, present in 8 patients (88.9%), predominantly abdominal pain. Oral ulcers were observed in 5 patients (55.6%). Elevated acute-phase reactants were detected in 7 patients (77.8%), including increased C-reactive protein and/or erythrocyte sedimentation rate. Genetic analysis identified 7 distinct heterozygous variants classified as conflicting, variants of uncertain significance, likely benign, or not found in databases. Therefore, no specific variants were identified, and the patients show diversity. Three patients received colchicine; two required additional interleukin-1 blockade with anakinra and/or canakinumab.
**Conclusion**
Children with NLRP12 variants may present with a broad clinical spectrum, ranging from mild or possibly incidental findings to more inflammatory phenotypes with recurrent fever and involvement of the mucosal, gastrointestinal, and musculoskeletal systems. The coexistence of variants with uncertain, conflicting, or likely benign significance highlights the need to interpret NLRP12 results in close relation to the clinical phenotype and inflammatory markers. Larger multicenter studies are needed to clarify genotype–phenotype correlations and to identify which patients may benefit from targeted anti-inflammatory treatment.

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**Disclosure of interest**
None declared.

## P253

### Chronic nonbacterial osteomyelitis of the mandible in a patient with Crohn's disease: a diagnostic challenge

H. Petrovic[1,*], S. Djordjevic[1,2], G. Susic[1], N. Ristic[3], M. Radusinovic[3]
[1]Rheumatology, University Children's Hospital, Belgrade; [2]Faculty of medicine, University of Belgrade; [3]Gastroenterology, University Children's Hospital, Belgrade, Serbia
**Correspondence:** H. Petrovic
*Pediatric Rheumatology 2026*, **24(S1):**P253

**Introduction**
Chronic nonbacterial osteomyelitis (CNO) is a rare autoinflammatory bone disorder, characterized by sterile bone inflammation and frequently associated with immune-mediated diseases such as inflammatory bowel disease (IBD). Isolated mandibular involvement is uncommon and poses a significant diagnostic challenge, particularly in distinguishing it from infectious osteomyelitis and medication-related osteonecrosis of the jaw (MRONJ).
**Objectives**
Our patient is 14 year old girl with Crohn's disease diagnosed in 2022, previously treated with infliximab from February 2023 to September 2024. In June 2024, an odontoma was incidentally identified and surgically removed. In August 2024, the patient developed recurrent mandibular swelling, pain, and increased local temperature, occasionally accompanied by low-grade fever (up to 37.3 °C). Initially suspected as infectious, symptoms partially improved with antibiotic therapy, leading to temporary discontinuation of immunosuppressive therapy; however, relapses occurred. A CT scan in September 2024 suggested osteomyelitis. Mandibular biopsy performed in November 2024 confirmed chronic osteomyelitis without identification of a causative pathogen. Although MRONJ was considered, histopathological findings did not support this diagnosis. Rheumatologic evaluation in December 2024 raised suspicion of CNO based on persistent symptoms, sterile cultures, mildly elevated inflammatory markers, nocturnal pain, and coexistence of Crohn's disease. Whole-body MRI in January 2025 demonstrated isolated mandibular involvement without other skeletal lesions. Follow-up mandibule CT supported chronic osteomyelitis of likely nonbacterial etiology. Concurrently, the patient developed reduced appetite and intermittent abdominal pain, with endoscopic confirmation of Crohn's disease relapse in March 2025. Treatment with corticosteroids and methotrexate resulted in clinical improvement, allowing gradual steroid tapering and reintroduction of infliximab in May 2025.
**Methods: Discussion**
This case highlights the diagnostic complexity of mandibular CNO, particularly in the context of underlying Crohn's disease and prior biologic therapy. The absence of microbiological confirmation, limited and transient response to antibiotics, and association with systemic inflammatory disease support a noninfectious etiology. Differentiation from infectious osteomyelitis and MRONJ is essential to ensure appropriate immunosuppressive treatment.
**Conclusion**
CNO should be considered in the differential diagnosis of mandibular pain and swelling in patients with IBD. Early recognition and a multidisciplinary approach are crucial to optimize outcomes and avoid unnecessary prolonged antibiotic use.

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**Disclosure of interest**
None declared.

## P254

### Temporal changes in diagnosis and management of pediatric familial Mediterranean fever: a 25–year single–center cohort study

P. Prencuva Akyürek[1,*], S. D. Arık[1], B. Menentoğlu[1], A. Dudaklı[1], Ö. Akgün[1], N. Aktay Ayaz[1]
[1]Division of Pediatric Rheumatology, Istanbul University, Istanbul Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** P. Prencuva Akyürek
*Pediatric Rheumatology 2026*, **24(S1):**P254

**Introduction**
Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease in childhood and is characterized by recurrent febrile attacks and serosal inflammation. Early diagnosis and timely colchicine treatment are crucial to prevent long-term complications, particularly amyloidosis and chronic inflammatory damage. Over the past decades, increasing awareness of autoinflammatory diseases, broader use of genetic testing, and advances in pediatric rheumatology care have substantially influenced the diagnostic and therapeutic approach to FMF. However, data evaluating temporal changes in clinical presentation, genetic characteristics, and treatment patterns in pediatric FMF patients over long-term follow-up periods remain limited.
**Objectives**
This study aimed to evaluate temporal changes in diagnostic delay, clinical characteristics, genetic features, and treatment approaches in pediatric familial Mediterranean fever (FMF) patients over a 25-year period.
**Methods**
This single-center retrospective cohort study included 497 pediatric FMF patients followed between 2000 and 2025. Patients were divided by year of diagnosis: Group 1 (before 2012, $n$=250) and Group 2 (2012 or later, $n$=247). Demographic, clinical, genetic, and treatment data were compared. Multivariable logistic regression identified independent predictors of colchicine-resistant disease.
**Results**
Group 2 patients were diagnosed at a younger age (median 6.0 vs. 7.4 years, $p$<0.001) with a shorter diagnostic delay (median 1.0 vs. 2.0 years, $p$<0.001). Group 2 exhibited a milder clinical phenotype, with lower rates of arthritis (15.0% vs. 28.8%, $p$<0.001), arthralgia (47.8% vs. 58.8%, $p$=0.014), erysipelas-like erythema (7.7% vs. 14.8%, $p$=0.012), and chest pain/pericarditis (18.2% vs. 27.6%, $p$=0.013). Heterozygous exon 10 MEFV variants were more frequent in Group 2 (46.2% vs. 35.2%), while homozygous exon 10 mutations predominated in Group 1 (34.8% vs. 27.1%) ($p$=0.023). Nephritic-range proteinuria declined markedly (13.6% vs. 4.5%, $p$<0.001) and biologic therapy use increased (10.8% vs. 19.8%, $p$=0.005). In multivariable logistic regression, homozygous MEFV mutations (odds ratio [OR] 6.00, 95% confidence interval [CI] 3.40–10.59), study period (Group 2 vs. 1: OR 2.81, 95% CI 1.56–5.07), arthritis (OR 1.98, 95% CI 1.05–3.74), and chest pain/pericarditis (OR 2.16, 95% CI 1.16–4.01) were independent predictors of colchicine resistance.
**Conclusion**
This 25-year cohort study demonstrates that subspecialty recognition coincided with earlier diagnosis, milder clinical phenotypes, reduced renal complications, and increased biologic use in pediatric FMF. Homozygous exon 10 MEFV mutations remained the dominant predictor of colchicine-resistant disease.
**Disclosure of interest**
None declared.

## P255

### The reshaping of HIDS: bronchiectasis and recurrent pulmonary infections in three Jordanian pediatric patients

R. Al-Laymon[1,*], M. Abu shukair[2], B. Aladaileh[3]
[1]Albashir Hospital, Amman, Jordan; [2]Pediatrics, Albashir Hospital; [3]Pediatrics, Royal medical services, Amman, Jordan
**Correspondence:** R. Al-Laymon
*Pediatric Rheumatology 2026*, **24(S1):**P255

**Introduction**
Hyperimmunoglobulinemia D syndrome (HIDS) is an autosomal recessive autoinflammatory disorder representing the milder end of the mevalonate kinase deficiency (MKD) spectrum. Caused by homozygous or compound heterozygous MVK mutations, it disrupts the cholesterol and isoprenoid biosynthetic pathways by impairing mevalonate kinase activity. Phenotypic severity correlates directly with residual enzymatic function, ranging from isolated periodic fever episodes to severe systemic metabolic dysfunction (1). While attacks can occur idiopathically, they are frequently triggered by immunization, infection, or stress (2). Notably, severe phenotypes can mimic other conditions, presenting with inflammatory bowel disease-like gastrointestinal manifestations, interstitial lung disease, or an immunodeficiency-like pattern of recurrent bacterial infections.(3).
**Objectives**
To report a rare manifestation of homozygous mutation of HIGD presenting with recurrent respiratory infections and subsequent bronchiectasis.
**Methods**
A retrospective review of the clinical presentation, laboratory investigations, imaging findings, genetic profiles, and therapeutic management of three pediatric patients presenting HIGD syndrome, recurrent pulmonary infections, and chronic cough.
**Results**
Individuals with homozygous Hyper IgD Syndrome mutations can manifest interstitial lung disease and bronchiectasis. Their frequent chest infections closely mimic a primary immunodeficiency phenotype.
**Conclusion**
Homozygous Hyper IgD syndrome behave differently in term of lung manifestation from Heterozygous type.

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**Disclosure of interest**
None declared.

## P256

### Real-world application of the 2025 EULAR/ACR classification criteria for paediatric chronic nonbacterial osteomyelitis in a tertiary cohort

R. Craveiro Costa[1], B. Luzio Vaz[2], C. Jesus e Sá[3], E. Bandeira[4], I. Marques[1], I. Madureira[1,*], C. Henriques[1], M. Paula Ramos[1], M. Conde[1]
[1]Unidade de Reumatologia Pediátrica; [2]Unidade de Pediatria Médica, Hospital de Dona Estefânia, Unidade Local de Saúde de São José, Lisboa; [3]Serviço de Pediatria, Unidade Local de Saúde do Arco Ribeirinho, Barreiro; [4]Serviço de Radiologia, Hospital de Dona Estefânia, Unidade Local de Saúde de São José, Lisboa, Portugal
**Correspondence:** I. Madureira
*Pediatric Rheumatology 2026*, **24(S1):**P256

**Introduction**
The 2025 EULAR/ACR classification criteria for paediatric chronic nonbacterial osteomyelitis (CNO) provide the first internationally validated weighted classification framework for this disease.
**Objectives**
To evaluate the retrospective applicability of these criteria in a tertiary paediatric rheumatology cohort and assess the contribution of histological weighting to classification and diagnostic timing.
**Methods**
We performed a retrospective single-centre study of paediatric patients with CNO or CNO-like autoinflammatory osteitis followed between January 2008 and May 2025. The 2025 criteria were retrospectively applied at diagnosis. Genetically confirmed monogenic autoinflammatory disorders were analysed separately as exclusion conditions. Descriptive statistics were used. Biopsied and non-biopsied patients were compared using Mann-Whitney U tests. In biopsied patients with biopsy performed before final diagnosis, time to biopsy and final diagnosis were compared using Wilcoxon signed-rank testing.
**Results**
Thirty-eight cases were reviewed. Two monogenic autoinflammatory osteitis cases, deficiency of interleukin-1 receptor antagonist (DIRA; score 44) and pyogenic arthritis, pyoderma gangrenosum and acne (PAPA) syndrome (score 53), were excluded from classification analysis. All remaining eligible CNO cases fulfilled the EULAR/ACR criteria (36/36, 100%). Mean age at diagnosis was 11.9 ± 3.0 years; 19/36 (52.8%) were

female. Median time to diagnosis was 10.5 months (IQR 6–16). Typical clavicular and/or mandibular involvement occurred in 13/36 (36.1%). Multifocal disease was present in 25/36 (69.4%), including symmetrical lesions in 7/36 (19.4%). Associated inflammatory features included arthritis in 11/36 (30.6%) and inflammatory skin disease in 6/36 (16.7%). Median classification score was 72 (IQR 65–76). Twenty-three patients underwent bone biopsy, including 9/11 (81.8%) with unifocal disease. Histological weighting contributed to classification in 17/23 (73.9%). Biopsied patients had higher total scores than non-biopsied patients (median 76 vs. 65; $p$=0.001), but scores became similar after removal of the histological domain (median 65 vs. 65; $p$=0.368). All biopsied patients remained above the ≥55-point threshold after removal of histological weighting. Among 22 biopsied patients with biopsy performed before final diagnosis, time from symptom onset to biopsy was shorter than time to diagnosis (median 6.5 vs. 10 months; $p$<0.001).

**Conclusion**

The 2025 EULAR/ACR criteria demonstrated excellent retrospective applicability, successfully classifying all eligible non-monogenic CNO cases. Histological weighting increased total scores but was not required for classification. Earlier application of the criteria without dependence on histological confirmation could reduce diagnostic and therapeutic delay in clinically and radiologically typical paediatric CNO.

**Disclosure of interest**

None declared.

## P257

### Recurrent aphthosis in childhood: clinical and laboratory clues for the suspicion of Behçet's disease

S. M. Murias[1,*], E. Pardo[1], V. Courel[1], I. Molinos[2], O. Salcedo[1], A. Gómez[2], N. Callejas[2], P. Alvarez[2], J. Rodriguez[1]
[1]Unit of Pediatric Rheumatology; [2]Rheumatology, University Hospital of Asturias, Oviedo, Spain
**Correspondence:** S. M. Murias
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**Introduction**

Recurrent aphthosis is a common reason for consultation in pediatric rheumatology. It may occur either as an isolated condition or associated with autoinflammatory or rheumatic diseases such as Behçet's disease (BD), which diagnosis is often difficult due to its incomplete and evolving presentation.

**Objectives**

The aim of this study was to describe the clinical characteristics of children with recurrent aphthosis and to identify laboratory markers that may support the suspicion of Behçet's disease (BD).

**Methods**

A retrospective observational study was conducted in patients under 18 years old followed at the Pediatric Rheumatology Unit of Hospital Universitario Central de Asturias between October 2020 and December 2025. All patients presented recurrent aphthosis as the cardinal clinical manifestation. A descriptive analysis of the data was performed using SPSS Statistics, and comparisons between variables at baseline and during flares were carried out using non-parametric tests. A p value < 0.05 was considered statistically significant. The 2015 Pediatric Behçet's Disease Group (PEDBD) classification criteria were used for the diagnosis of Behçet's disease (BD).

**Results**

Thirty patients were included (53.3% female) with the following diagnoses: PFAPA (30%), recurrent oral aphthosis (30%), incomplete BD (13.3%), hyper-IgD syndrome (6.7%), and uveitis (3.3%). Systemic symptoms were present in 73.3% of patients, mainly fever (50%), gastrointestinal symptoms (26.7%), and arthralgia (26.7%). Painful oral ulcers occurred in 60% and genital ulcers in 26.7%. Periodic episodes were reported in 46.7%, while 10% had continuous symptoms. HLA-B5 positivity was found in 26.7%. Treatments included oral corticosteroids (40%), anti-TNF agents (20%), and colchicine (10%). IgD levels were more frequently elevated during flares in patients classified as incomplete Behçet's disease, but not at baseline (no statistically significant differences were found); whereas C-reactive protein (CRP) levels showed a significant increase during flares ($p$ = 0.015).

**Conclusion**

Some patients showed clinical and laboratory features suggestive of incomplete BD, particularly genital ulcers, systemic symptoms, and elevated acute-phase reactants during flares. CRP was useful as a disease activity marker, while IgD increased preferentially during flares in suspected BD cases. Although many patients responded to topical therapy, a considerable proportion required systemic or biologic treatment with good outcomes.

**Disclosure of interest**

None declared.

## P258

### Identification of the DNASE2 c.511+5G>A variant in a group of patients and carriers suggesting a founder effect in the Turkic ethnic group

V. I. Burlakov[1,*], N. Kan[1], A. Roppelt[2], A. Avedova[1], D. Bogdanova[1], Z. Nesterenko[1], D. Akhmatova[1], E. Raykina[3], A. Gadzhiev[3], M. Gurzhikhanova[3], Y. Rodina[1], O. Schnappauf[4], I. Aksentijevich[4], A. Shcherbina[1]
[1]Department of Immunology, Dmitriy Rogachev National Center for Pediatric Hematology, Oncology, and Immunology; [2]State Budgetary Institution of Healthcare "Moscow Clinical Research Center Hospital No.52 of the Moscow City Health Department"; [3]Laboratory of Molecular Biology, Dmitriy Rogachev National Center for Pediatric Hematology, Oncology, and Immunology, Moscow, Russian Federation; [4]National Human Genome Research Institute, National Institutes of Health, Bethesda, MD, USA
**Correspondence:** V. I. Burlakov
*Pediatric Rheumatology 2026*, **24(S1):**P258

**Introduction**

DNase II deficiency is a rare autosomal recessive type I interferonopathy associated with multi-organ autoinflammatory complications, including cytopenia, arthritis, hepatitis, nephritis, and recurrent fever. DNase II is a lysosomal endonuclease predominantly expressed in macrophages, where it degrades nuclear DNA from apoptotic cells and enucleated erythroid precursors. We previously identified one patient homozygous for the splice-site variant (c.511+5G> A) in the *DNASE II* gene.

**Objectives**

Genetic testing was performed to identify disease-causing variants in two unrelated patient suspected to have a DNase II deficiency. Subsequently, we searched our database for additional carriers of the c.511+5G> A variant.

**Methods**

Sequencing was performed using targeted next-generation sequencing panel, whole-genome sequencing, and Sanger sequencing. Segregation analysis was confirmed by Sanger sequencing.

**Results**

We demonstrated that the splice-site variant (c.511+5G> A) results in exon 4 skipping and impaired DNase II activity. Patient 1 (P1), a previously reported female, was born to consanguineous parents of Tatar descent. Patients 2 and 3 (P2 and P3) were males born to unrelated non-consanguineous families of Bashkir descent. Tatars and Bashkirs are closely related subgroups of the Turkic ethnic group native to the Volga-Ural region of Russia. All three patients exhibited a similar clinical course, characterized by early-onset cytopenia, systemic inflammation, autoimmune arthritis with contractures, and decreased T-cell counts with hypergammaglobulinemia. All three patients received JAK inhibitor therapy, while P3 additionally received anifrolumab with partial clinical response. P1 underwent allogeneic hematopoietic stem cell transplantation (HSCT). All three patients were homozygous for the rare splice-site variant *DNASE2* gene c.511+5G> A, which has an allele frequency $1.8 \times 10^{-5}$ in the gnomAD v4.1.0 database. In each family, the parents were found to be heterozygous carriers. P2 had two siblings and P3 had one sibling who were also heterozygous carriers. A subsequent search of our institutional genetic database identified three additional unrelated heterozygous carriers who had undergone genetic testing for reasons other than autoinflammation. Two of these individuals were native to the same Volga-Ural region. All identified carriers reported Tatar or Bashkir descent.

**Conclusion**

We report three unrelated patients with DNase II deficiency carrying the same homozygous *DNASE II* c.511+5G> A variant and presenting with a similar phenotype, along with 3 unrelated asymptomatic carriers sharing the same ethnicity. Given the rarity of this variant in the general population, a founder effect in this ethnic group is strongly suspected.

We plan to expand our search for additional carriers and patients within these ethnic groups.

**Disclosure of interest**
None declared.

## P259

### Hemogram-derived inflammatory indices in PFAPA syndrome: a within-patient paired study

O. A. Akin[1], P. S. Çıldır[2], O. D. Tosun[1], T. D. Kuzuca[1], S. K. Kılıç[1], C. Bal[3], S. Demir[1,*]
[1]Pediatric Rheumatology; [2]Pediatrics; [3]Biostatistics, Eskisehir Osmangazi University, Eskısehir, Türkiye
**Correspondence:** S. Demir
*Pediatric Rheumatology 2026*, **24(S1):**P259

**Introduction**
PFAPA is one of the most common autoinflammatory periodic fever syndromes in childhood. Hemogram-derived composite inflammatory indices may provide practical, low-cost markers of attack-related inflammation, but have not been systematically evaluated in PFAPA using a within-patient paired design.
**Objectives**
To compare hemogram-derived inflammatory indices between attack and attack-free periods within the same patients, and to evaluate their associations with clinical disease burden and capacity to distinguish attack periods.
**Methods**
This retrospective, within-patient paired study included 57 children with PFAPA fulfilling Eurofever/PRINTO classification criteria at a tertiary pediatric rheumatology center. Patients with concurrent infection were excluded. Attack-period samples were obtained before corticosteroid administration; the interval between paired samples was 30–180 days. Neutrophil-to-lymphocyte ratio (NLR), platelet-to-lymphocyte ratio (PLR), monocyte-to-lymphocyte ratio (MLR), systemic inflammatory response index (SIRI), systemic immune-inflammation index (SII) and aggregate index of systemic inflammation (AISI) were calculated. Clinical activity was assessed by AIDAI. Paired t-test or Wilcoxon test, Spearman correlation and ROC analysis were performed. The study protocol was approved by the Eskişehir Osmangazi University Non-Interventional Clinical Research Ethics Committee.
**Results**
Thirty-one patients were male (54.4%); median attacks were 4 (2–5) in the last 6 months and 8 (5–8) in the last year. WBC was higher during attacks (13,162 ± 4,556 vs. 8,412 ± 2,218/μL; $p<0.001$) and hemoglobin lower (11.88 ± 1.20 vs. 12.16 ± 1.11 g/dL; $p=0.007$). All composite indices were significantly increased during attacks: NLR 3.14 vs. 0.96, PLR 116.4 vs. 99.6, MLR 0.397 vs. 0.159, SIRI 3.18 vs. 0.51, SII 944.9 vs. 335.4 and AISI 990 vs. 194 (all $p<0.001$ except PLR, $p=0.005$). AIDAI correlated most strongly with SII (rho=0.481) and NLR (rho=0.466). ROC analysis showed highest discrimination for SIRI (AUC=0.909), MLR (0.887), NLR (0.882), AISI (0.879) and SII (0.840), whereas PLR was limited (AUC=0.598; $p=0.066$).
**Conclusion**
SIRI, MLR, NLR, AISI and SII reflected attack-related inflammatory activation more robustly than PLR and may serve as practical, accessible biomarkers complementing existing inflammatory markers in PFAPA. Prospective multicenter validation is required before standalone clinical use.
**Disclosure of interest**
None declared.

## P260

### Molecular evidence of MEFV variants in ancient populations of Laodikeia, Western Anatolia

S. Yüksel[1,*], C. Şimşek[2], G. O. Çetin[3], S. Türel[3], T. Becerir[4], G. Kılbaş[5], S. Ayduran[6], S. Özen[7]
[1]Pediatric Rheumatology, Çanakkale Onsekiz Mart University, Çanakkale; [2]Archaeology; [3]Medical Genetics; [4]Pediatric Nephrology, Pamukkale University, Denizli; [5]Pediatric Rheumatology, Trakya University, Edirne; [6]Pediatric Rheumatology, Hatay Research Hospital, Hatay; [7]Pediatric Rheumatology, Hacettepe University, Ankara, Türkiye
**Correspondence:** S. Yüksel
*Pediatric Rheumatology 2026*, **24(S1):**P260

**Introduction**
Familial Mediterranean fever (FMF) is the most prevalent monogenic autoinflammatory disease in the Eastern Mediterranean. Despite high carrier frequencies across multiple populations, direct ancient DNA evidence for MEFV variants at the site level is entirely absent. Roman Anatolia represents an ideal context to test whether clinically relevant MEFV variation was already present nearly two millennia ago.
**Objectives**
To determine whether pathogenic MEFV variants were detectable in ancient individuals from Roman-era Laodikeia, western Anatolia (1st–5th c. CE), and to assess whether their sector-specific distribution reflected population-level variation or local burial demography.
**Methods**
Targeted next-generation sequencing of the entire MEFV gene (RefPleks MEFV panel, Gen-Era Diagnostics; Illumina NextSeq 500) was performed on molar-tooth samples from 42 ancient individuals across four necropolis sectors of Laodikeia (NW n=7, W n=19, NE n=15, S n=1), all processed under ancient-DNA clean-room conditions. Authenticity was confirmed by fragment-length distribution (mean 112.89 bp), characteristic terminal damage patterns (5′ C> T, 3′ G> A), and mean read depth (30×; range 49–99%). Variants were classified per ACMG/AMP criteria (Franklin, ClinVar, Infevers).
**Results**
MEFV variants were detected in 21 of 42 individuals (50.0%; 95% CI 34.2–65.8%), with a markedly uneven distribution across necropolis sectors. The **West Necropolis** showed the highest positivity rate (15/19; 78.9%; 95% CI 54.4–93.9%), dominated by E148Q, in a context of large family tomb complexes (Tomb 3 and Tomb 4). The **Northeast** yielded 5/15 positives (33.3%; 95% CI 11.8–61.6%), including heterozygous M694V, compound E148Q+V726A, and homozygous R761H — all from individual graves. The **South** contributed one heterozygous M694V carrier (1/1). The **Northwest** was negative (0/7; 95% CI 0.0–41.0%). Pathogenic or likely pathogenic variants (M694V, V726A, R761H) were identified in 4 individuals from the Northeast and South sectors, independent of E148Q (see Table 1).

| Sector | *n* | Positive | Rate (95% CI) | Variants Detected |
|---|---|---|---|---|
| West | 19 | 15 | 78.9% (54.4-93.9) | E148Q (hom/het), P369S, R202Q |
| Northeast | 15 | 5 | 33.3% (11.8-61.6) | E148Q, M694V, V726A, R761H |
| South | 1 | 1 | 100% (2.5-100) | - |
| Northwest | 7 | 0 | 0% (0-41.0) | - |

**Conclusion**
This is the first site-focused ancient DNA study targeting the entire MEFV gene. Pathogenic variants were present in Roman western Anatolia, placing Laodikeia within the Eastern Mediterranean MEFV landscape. E148Q enrichment in the West Necropolis reflects local family tomb clustering, not population-wide prevalence. These findings underscore the value of integrating archaeogenetics with burial context.

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**Disclosure of interest**
None declared.

## P261

### DADA2 recurrence with declining donor chimerism and ADA2 enzyme activity post–HSCT

S. Cuyx[1,*], J. Silva[2], I. Turtsevich[3], D. Eleftheriou[1]
[1]Paediatric Rheumatology; [2]Bone Marrow Transplantation Team, Great Ormond Street Hospital for Children NHS Foundation Trust, London; [3]Paediatric Rheumatology, Oxford University Hospitals NHS Foundation Trust, Oxford, UK
**Correspondence:** S. Cuyx
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**Introduction**
Deficiency of adenosine deaminase 2 (DADA2) is a monogenic autoinflammatory and vasculopathic disorder caused by biallelic pathogenic variants in *ADA2*. Clinical manifestations are heterogeneous and include vasculitis, stroke, cytopenias, immunodeficiency, and bone marrow failure. Although TNF inhibition effectively prevents vascular complications in many patients, allogeneic haematopoietic stem cell transplantation (HSCT) remains the only curative treatment for severe haematological and immunological disease.
**Objectives**
To demonstrate the relationship between declining donor chimerism, reduced ADA2 enzyme activity, and disease recurrence following HSCT in DADA2.
**Methods**
We report, with parental consent, a child with DADA2 who developed clinical relapse following HSCT.
**Results**
A female child of Turkish origin, born to consanguineous parents, presented at 4 months of age with recurrent fevers, hepatosplenomegaly, failure to thrive, microcephaly, pulmonary stenosis, and severe neutropenia. Investigations demonstrated hypocellular bone marrow, antineutrophil antibodies, and absent plasma ADA2 activity. Genetic testing identified a homozygous exon 7 deletion in ADA2, confirming DADA2. There was no clinical or radiological evidence of central nervous system involvement at diagnosis. She received rituximab and intravenous immunoglobulin; however, due to persistent neutropenia and recurrent infections, she underwent 9/10 mismatched unrelated donor HSCT at 16 months of age. Initial complete donor chimerism progressively declined from 4 months post-HSCT, paralleled by a reduction in plasma ADA2 activity. At 12 months post-transplant, she developed recurrent vasculitic rash and fevers, with ADA2 activity measuring 2 IU/L. As donor chimerism further decreased to 23% and ADA2 activity to 1.01 IU/L, she presented with gait instability. Brain MRI demonstrated an infarct in the left paramedian midbrain. Within days, she developed acute left oculomotor nerve palsy and mild left-sided weakness, accompanied by radiological progression of stroke. Treatment with etanercept, intravenous methylprednisolone, and a tapering course of oral prednisolone was initiated. No further neurological events occurred, and she achieved complete neurological recovery at 18 months' follow-up.
**Conclusion**
This case suggests that sustained donor-derived ADA2 production above a critical threshold is necessary for durable disease control following HSCT. Plasma ADA2 activity may serve as a complementary biomarker to donor chimerism for detecting graft insufficiency and impending disease recurrence. These findings have important implications for post-transplant surveillance and suggest that restoration of ADA2 enzyme activity above a critical therapeutic threshold should be a target for emerging gene therapy strategies in DADA2.
**Disclosure of interest**
None declared.

## P263

### Fever and a limp – a diagnostic conundrum

S. Ramakrishnan[*,1], V. Shivamurthy[1]
[1]Paediatric Rheumatology, Evelina London Children's Hospital, Guy's and St Thomas' NHS Foundation Trust, London, UK
**Correspondence:** S. Ramakrishnan
*Pediatric Rheumatology 2026*, **24(S1):**P263

**Introduction**
Deficiency of adenosine deaminase 2 (DADA2) is an ultra-rare genetic autoinflammatory condition which is often misdiagnosed due to its high phenotypic variability. Diagnosis is made based on a high index of suspicion, low plasma ADA2 levels and genetic testing. Treatment with TNFα inhibitors has been shown to significantly reduce ischaemic complications and vasculitic activity (1). We present an interesting case of a 5-year-old boy who presented with non-specific symptoms, but was eventually diagnosed with DADA2.
**Objectives**
Through discussion of this case, we aim to highlight the importance of considering testing for DADA2 in patients with non-specific systemic inflammatory symptoms, particularly in very young children.
**Methods**
**Case**
A 5-year-old boy presented with 2 months of intermittent fevers, limp, rash, leg pain, weight loss and night sweats. Initial blood tests showed a raised ESR, CRP and LDH, normal creatine kinase, and negative autoimmune screen. MRI of the lower limbs showed patchy multifocal muscle signal changes in both lower limbs but more severe in the right calf. The rest of his imaging and muscle biopsy were normal. He had a previous similar episode at 2 years of age, during which he was treated for presumed inflammatory arthritis. MRI of the left lower limb at this time showed myositis-like changes. Following the current presentation, he was treated for an unexplained systemic inflammatory disease with systemic steroids which resulted in improvement. He was started on a weaning regimen of oral steroids, but relapsed after stopping. He was found to have very low plasma adenosine deaminase 2 levels, and a compound heterozygous pathogenic mutation for DADA2, c.139G> T (p.Gly47Trp) and c.139G> A (p.Gly47Arg). Following this, he had recurrent relapses, but after being started on etanercept, has remained largely asymptomatic.
**Conclusion**
We describe an unusual case of DADA2 with very low levels of ADA2 enzyme activity and a compound heterozygous mutation in a 5-year-old. DADA2 has the potential to cause severe complications, especially stroke, and as such, early diagnosis and treatment with TNFα inhibitors is vital (2). Further research is needed to elucidate prognostic factors for this ultra-rare disease and to optimise management.
**Consent to publish**
Written informed consent has been obtained from the participant's parent in line with the submission guidelines.

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**Disclosure of interest**
None declared.

## P264

### From thrombocytopenia to type I interferonopathy: a case report

M. K. Karakukcu[1], Z. B. Azızoglu[2], H. Tastanoglu[3], S. B. Demir[2], S. Ozdemir Cicek[2,3,*], A. Ozcan[4], A. Pac Kisaarslan[2,3], M. Karakukcu[4], M. H. Poyrazoglu[2,3]
[1]Pediatrics; [2]Genome and Stem Cell Center (GENKÖK); [3]Pediatric Rheumatology; [4]Pediatric Hematology, Erciyes University Faculty of Medicine, Kayseri, Türkiye
**Correspondence:** S. Ozdemir Cicek
*Pediatric Rheumatology 2026*, **24(S1):**P264

**Introduction**
Pathogenic variants in the *IFIH1*(*MDA5*) gene may present with different clinical phenotypes. Aicardi-Goutières syndrome type 7 (AGS7) and Singleton-Merten syndrome type 1(SMS1). Although both disorders are associated with the same gene, their phenotypic manifestations differ: AGS7 is primarily characterized by dermatological involvement, neurological regression, and early-onset encephalopathy, whereas SMS1 is associated with skeletal abnormalities, dental dysplasia, and

aortic calcification. Here, we present a one-year-old girl evaluated for pancytopenia in whom a heterozygous *IFIH1* c.281 A> G p.Y94C variant was identified.

**Objectives: Case Presentation**

A one-year-old girl was referred for evaluation of pancytopenia. Her medical history revealed that she was born at 35 GW with a birth weight of 2200 g. At 11 months of age, thrombocytopenia had been detected during investigations performed for eczematous skin lesions, and she had subsequently been diagnosed with immune thrombocytopenic purpura and treated with IVIG, PMP, and eltrombopag. Due to the early-onset rash and thrombocytopenia, genetic analysis was performed and revealed a pathogenic heterozygous *IFIH1* c.281 A> G p.Y94C variant. On physical examination, a 2×2 cm ecchymosis was present on the left cheek, along with diffuse eczematous lesions over the abdomen. Laboratory investigations demonstrated pancytopenia, with a white blood cell (WBC) count of $1.15\times10^3$/μL, hemoglobin (Hb) level of 4.9 g/dL, and platelet (PLT) count of $5\times10^3$/μL. Cranial imaging and cardiac evaluations were normal. The interferon (IFN) score was elevated at 9.56 (cut-off: 4.60). The patient received platelet suspension transfusion for supportive treatment. Due to the elevated IFN score, ruxolitinib therapy was initiated in combination with corticosteroids. During the first month of treatment, hematological parameters improved markedly, allowing gradual tapering and discontinuation of corticosteroid therapy. At the sixth month of ruxolitinib treatment, the pre-dose trough level of ruxolitinib was 2.39 ng/mL (reference range: 9.94–50.48), while the 2-hour post-dose peak level was 67.88 ng/mL (reference range: 49.59–144.45). Complete blood count parameters were within normal limits, including Hb 11.7 g/dL, WBC $5.61\times10^3$/μL, and PLT $142\times10^3$/μL. During follow-up, treatment dosage adjustments were made according to PLT counts and IFN scores.

**Methods: Conclusion**

*IFIH1* mutations may present with a broad phenotypic spectrum. This case represents a type I interferonopathy predominantly characterized by hematological manifestations, elevated interferon score, and absence of major neurological or cardiac abnormalities.

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**Disclosure of interest**

None declared.

## P265

**Mediterranean Diet Adherence And Disease Activity In Pediatric Familial Mediterranean Fever**

B. Çinpolat[1], Z. Hazıroğlu Ökmen[1], T. Güzel[1], M. T. Karadoğan[1,*], E. E. Fırat[1], F. Çakmak[1]
[1]Basaksehir Cam and Sakura City Hospital, Istanbul, Türkiye
**Correspondence:** M. T. Karadoğan
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**Introduction**

Familial Mediterranean fever (FMF) is the most common hereditary autoinflammatory disease characterized by recurrent inflammatory attacks mediated predominantly through dysregulation of the innate immune system. Although colchicine effectively controls disease activity in most patients, environmental and nutritional factors may also contribute to inflammatory burden. Evidence regarding the relationship between Mediterranean diet adherence and disease activity in pediatric FMF remains limited.

**Objectives**

To evaluate the relationship between Mediterranean diet adherence, assessed using the Mediterranean Diet Quality Index for children and adolescents (KIDMED), and disease activity measured by the International Severity Score for Familial Mediterranean Fever (ISSF) in children with FMF.

**Methods**

This cross-sectional study included children aged ≥5 years with FMF followed at a tertiary pediatric rheumatology center. Mediterranean diet adherence was assessed using the KIDMED index, and disease activity was evaluated using ISSF scores. Demographic and clinical characteristics were recorded. The relationship between KIDMED scores and disease activity was evaluated using correlation and subgroup analyses.

**Results**

A total of 127 children with FMF were included in the study, of whom 58 (45.7%) were female. The median age of the cohort was 128 months (IQR: 108–163). The median KIDMED score was 6 (IQR: 4–7), while the median ISSF score was 1 (IQR: 1–3). According to KIDMED categories, 23 patients (18.1%) had low, 75 (59.1%) moderate, and 29 (22.8%) high adherence to the Mediterranean diet. Most patients had mild disease activity according to ISSF categories. A significant negative correlation was observed between KIDMED and ISSF scores (Spearman rho = −0.21, p = 0.019).

**Conclusion**

Our findings suggest a potential relationship between Mediterranean diet quality and disease activity in pediatric FMF. Although disease activity was generally low in this colchicine-treated cohort, dietary factors may contribute to ongoing inflammatory activity despite colchicine treatment. These findings support growing interest in nutritional approaches as complementary components of comprehensive FMF management. Further prospective multicenter studies are needed to better clarify the impact of dietary patterns on disease activity in children with FMF.

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**Trial registration identifying number**

Not applicable.

**Disclosure of interest**

None declared.

## P266

**Behçet's disease-like phenotype associated with MEFV p.Ser242Arg mutation: a novel clinical presentation**

T. Šinkovec Savšek[1,2,*], N. Emeršič[1], M. Debeljak[3], T. Avčin[1,2], N. Toplak[1,2]
[1]Department of Allergology, Rheumatology and Clinical Immunology, University Children's Hospital, University Medical Centre Ljubljana; [2]Faculty of Medicine, University of Ljubljana; [3]Clinical Institute for Special Laboratory Diagnostics, University Children's Hospital, University Medical Centre Ljubljana, Ljubljana, Slovenia
**Correspondence:** T. Šinkovec Savšek
*Pediatric Rheumatology 2026*, **24(S1):**P266

**Introduction**

Mutations in exon 2 of the *MEFV* gene, particularly p.Ser242Arg, cause pyrin-associated autoinflammation with neutrophilic dermatosis (PAAND), characterized by neutrophilic skin inflammation, recurrent fever, and arthralgia. *MEFV* variants in exon 10 are also recognized as risk factors for Behçet's disease. However, to the best of our knowledge, no case with a Behçet's disease-like phenotype has been reported in association with the p.Ser242Arg mutation.

**Objectives**

To describe two siblings with the *MEFV* NM_000243.2:c.[726 C> G]; [=] (p.Ser242Arg) mutation presenting with clinical features consistent with Behçet's disease rather than the PAAND phenotype described in connection with this mutation.

**Methods**

Case series of two sisters (ages 17 and 19 years) followed at the Department of Allergology, Rheumatology and Clinical Immunology, University Children's Hospital, University Medical Centre Ljubljana are presented. Clinical evaluation, genetic testing for autoinflammatory diseases panel, and assessment of therapeutic response were performed.

**Results**
Both patients presented with recurrent oral aphthous lesions; neither had skin conditions related to neutrophilic dermatoses. The younger sister also had recurrent genital ulcers (infections excluded), while the older sister had only one episode of genital ulcers with a confirmed HSV-1 infection. Both experienced recurrent episodes of pain and swelling of the lower extremities with elevated inflammatory markers. Genetic testing WES was performed for a panel of autoinflammatory diseases, including 60 genes. We identified the heterozygous p.Ser242Arg mutation in exon 2 of *MEFV* in both patients. Colchicine therapy was initiated. During the 4.5 years and 3.5 years follow-up, respectively, both experienced disease flares with lower extremity symptoms; deep vein thrombosis was excluded in the hospitalized patient. After increasing the dose of colchicine to 2 mg daily, both are currently in remission.
**Conclusion**
These cases demonstrate phenotypic variability in *MEFV* exon 2 mutations, with the p.Ser242Arg variant presenting as Behçet's disease-like phenotype rather than the typical PAAND. This broadens the clinical spectrum associated with this mutation and underscores the importance of genetic testing in autoinflammatory conditions, as phenotype may differ from the expected presentation.

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**Disclosure of interest**
None declared.

## P267

### PFAPA: long-term symptoms in patients without tonsillectomy

T. Moberg[1,*], K. Rydenman[1,2], S. Berg[2,3], A. Fasth[2,3], P. Wekell[1,2,3]
[1]Department of Pediatrics, NU hospital group, Trollhättan; [2]Department of Pediatrics, Institute of Clinical Sciences, University of Gothenburg; [3]Department of Pediatric Rheumatology and Immunology, Queen Silvia Children's Hospital, Gothenburg, Sweden
**Correspondence:** T. Moberg
*Pediatric Rheumatology 2026,* **24(S1):**P67

**Introduction**
Only a few studies have investigated the clinical course and long-term symptoms in patients with PFAPA who have not undergone tonsillectomy. In a previous study, we described the clinical course and long-term symptoms in patients with PFAPA in whom tonsillectomy was performed.
**Objectives**
To investigate the clinical course and long-term symptoms in patients with PFAPA who have not undergone tonsillectomy.
**Methods**
An observational cohort study with cross sectional and retrospective data. All patients diagnosed between 2006 and 2017with PFAPA at the Queen Silvia Children's Hospital, NU Hospital Group and Skaraborg Hospital, in western Sweden in whom tonsillectomy was not performed were included. Data were collected by structured telephone interviews and review of medical records. If ≥18 years old, patients were interviewed regarding current PFAPA symptoms and the clinical course during adulthood. Parents were interviewed regarding current symptoms in patients <18 years old and regarding clinical course of PFAPA during childhood in all patients.
**Results**
The study invitation was answered by 124 of 187 (66%) patients/parents and cross-sectional data were available for all these patients. In 14 patients >18 years old parents did not respond, i.e. retrospective data were available for 110 patients. At a median age of 18.1 years (range 10.5-31.2) and a median follow-up time of 16.2 years (range 8.8-24.0) from disease onset, 24/124 (19%) had ongoing febrile episodes and 34/124 (27%) had ongoing non-febrile PFAPA-related symptoms. In patients who no longer had febrile episodes, the median illness duration before remission of febrile episodes was 4.6 years (range 0.5-18.9). There was a remitting-relapsing disease course in 37/110 patients (35%) with a median symptom free interval of 3.2 years (range 1.0-12.5). Of them, 12/37 (32%) had ongoing febrile episodes at the time of follow-up, median 4.5 years after the relapse of febrile episodes (range 1.0-14.0). In the 25/37 (68%) patients in whom the febrile episodes remitted again, the median duration of the relapse was 2.0 years (1.0-5.5). For 8/37 (22%), there were several periods of relapse and remission. In patients with ongoing febrile episodes, the median interval was 140 days (range 18-365) and the median duration of episodes was 3.5 days (range 0,5-10). The intervals were significantly longer than at the onset of the disease ($p$<0.01) but no difference was found in the median duration of the febrile episodes ($p$=0,66). The most common symptoms during febrile episodes were aphthous stomatitis in 14/24 (58%), followed by pharyngitis in 13/24 (54%) and cervical adenitis in 5/24 (21%). The most common symptoms during non-febrile episodes were aphthous stomatitis in 23/34 (68%), followed by pharyngitis in 11/34 (32%), malaise in 7/34 (21%) and myalgia/arthralgia in 4/34 (12%). Aphthous stomatitis was the only symptom in 12/34 (35%). Median interval between non-febrile symptoms was 47 days (range 7-365) and median duration was 4.5 days (range 0.5-14.0).
**Conclusion**
While remission of febrile episodes occurs in most patients with PFAPA, 19% still had febrile episodes after a median follow-up time of 17 years, although generally fewer episodes than at disease onset. Non-febrile symptoms were present in 27% with aphthous stomatitis being the most common symptom. Physicians should be aware of, and patients/parents should be informed of, the heterogenous disease course in PFAPA.
**Disclosure of interest**
None declared.

## P269

### Evaluation of the relationship between enthesitis thickness and exercise–induced leg pain in patients with familial Mediterranean fever

N. Z. Özaslan[1,*], T. Özer[2], Y. Anık[2], N. Şahin[1], H. E. Sonmez[1]
[1]Division of Pediatric Rheumatology, Faculty of Medicine; [2]Department of Radiology, Faculty of Medicine, Kocaeli University, Kocaeli, Türkiye
**Correspondence:** N. Z. Özaslan
*Pediatric Rheumatology 2026,* **24(S1):**P269

**Introduction**
Familial Mediterranean Fever (FMF) is an autoinflammatory disease characterized by recurrent episodes of fever and serositis. Musculoskeletal manifestations such as arthralgia, enthesitis, and exercise-induced leg pain are common in these patients. In recent years, increased enthesis thickness and exercise-induced leg pain have been suggested to be associated with a more severe disease course in FMF.
**Objectives**
In this study, we aimed to evaluate the relationship between enthesis thickness and disease severity in pediatric patients with FMF.
**Methods**
Patients followed in the pediatric rheumatology clinic with a diagnosis of FMF were included in this study. Demographic and clinical data were obtained from patient records. Enthesis thicknesses were measured by ultrasonography. The relationship between enthesis thickness and disease severity was analyzed statistically. Increased enthesis thickness was evaluated according to ultrasonographic reference values derived from healthy Turkish children reported in the literature.
**Results**
A total of 160 patients (89 females, 71 males) with FMF were included in the study. The median age was 10 years (5–19), while the median age at symptom onset and diagnosis were 4.5 years (5–16) and 6 years (1.5–17), respectively. The median follow-up duration was 3.2 years (2–17). Musculoskeletal involvement was common; arthralgia was present in 136 patients (85%), exercise-induced leg pain in 96 (60%), myalgia in 89 (55.6%), arthritis in 26 (16.2%). Forty-six patients (28.7%) were considered colchicine-resistant. Among all patients, increased enthesis thick-

**Table 1 (Abstract P269)** Comparison of enthesis tendon thicknesses between patients with and without exercise-induced leg pain

| | Patients with Exercise-Induced Leg Pain (*n*=96) | Patients without Exercise-Induced Leg Pain (*n*=64) | *p* value |
|---|---|---|---|
| Sex, F/M | 56/40 | 33/31 | 0.398 |
| Age, years* | 10 (2–19) | 11 (4–17) | 0.476 |
| BMI* | 17.48 (12.06–34.26) | 16.04 (12.11–31.53) | 0.06 |
| Distal quadriceps tendon, cm* | 0.51 (0.36–2.54) | 0.45 (0.26–0.71) | <0.001 |
| Proximal patellar ligament, cm | 0.38 ± 0.05 | 0.36 ± 0.06 | 0.03 |
| Distal patellar ligament, cm | 0.36 ± 0.04 | 0.34 ± 0.05 | 0.02 |
| Achilles tendon, cm | 0.46 ± 0.28 | 0.41 ± 0.07 | 0.19 |
| Plantar fascia, cm | 0.29 ± 0.02 | 0.28 ± 0.03 | 0.07 |

ness was most frequently detected at the distal quadriceps tendon in 44 patients (27.5%), followed by the Achilles tendon in 15 patients (9.4%) and the distal patellar ligament in 9 patients (5.6%). Increased thickness of the proximal patellar ligament was observed in only 1 patient (0.6%), while no increased plantar fascia thickness was detected. There were no significant differences between patients with and without exercise-induced leg pain regarding age, sex, or BMI. However, patients with exercise-induced leg pain had significantly greater distal quadriceps tendon and proximal/distal patellar ligament thicknesses (Table 1). When measurements exceeding the upper reference limits adjusted for age, sex, and BMI were evaluated, increased distal quadriceps tendon thickness was significantly more frequent in patients with exercise-induced leg pain ($p$= 0,01).

**Conclusion**

Distal quadriceps tendon thickness was increased in FMF patients with exercise-induced leg pain, whereas no association was observed with colchicine resistance or overall disease severity. These findings may reflect local mechanical loading or subclinical enthesopathy rather than severe systemic disease.

**Disclosure of interest**

None declared.

## P270

### Measurement of spleen size in patients with familial Mediterranean fever

N. Z. Özaslan[1,*], T. Özer[2], Y. Anık[2], N. Şahin[1], H. E. Sonmez[1]
[1]Division of Pediatric Rheumatology, Faculty of Medicine; [2]Department of Radiology, Faculty of Medicine, Kocaeli University, Kocaeli, Türkiye
**Correspondence:** N. Z. Özaslan
*Pediatric Rheumatology 2026*, **24(S1):**P270

**Introduction**

Familial Mediterranean Fever (FMF) is an autoinflammatory disease characterized by chronic and subclinical inflammation, predominantly mediated by interleukin-1–driven inflammatory responses. Persistent inflammation and recurrent attacks may contribute to splenomegaly through activation of the reticuloendothelial system, as suggested by case series and limited studies. However, the prevalence, clinical significance, and relationship of splenomegaly with inflammatory burden in FMF have not been clearly established, and large systematic cohort studies are lacking.

**Objectives**

This study aimed to evaluate spleen size and investigate associated clinical and laboratory parameters in patients with FMF.

**Methods**

In this cross-sectional study, demographic and clinical characteristics, disease activity indicators, and routine laboratory parameters of patients with FMF were systematically recorded. Spleen dimensions were measured during routine follow-up visits using standard ultrasonography and documented. Spleen sizes were converted into Z-scores according to age-adjusted reference values. Patients with a Z-score ≥2 were considered to have splenomegaly.

**Table 1 (Abstract P270)** Comparison of spleen dimensions between colchicine-resistant and colchicine-sensitive groups

| Spleen Measurements (mm) | Colchicine-Resistant Group (*n*=45) | Colchicine-Sensitive Group (*n*=115) | *p* value |
|---|---|---|---|
| Craniocaudal diameter | 104.90 (70–142.9) | 93 (65.4–154) | 0.01 |
| Z-score | 0.57 ± 1.47 | 0.46 ± 1.13 | 0.01 |
| Splenomegaly, n (%) | 9 (20) | 11 (9.5) | 0.07 |
| Transverse diameter | 84.9 (58.1–128) | 80 (15–117) | 0.02 |
| Anteroposterior diameter | 35.6 (26.3–58.8) | 32.2 (20–62.3) | 0.03 |

**Results**

A total of 160 patients (89 females, 71 males) were included in the study. The median age was 10 years (5–19), while the median age at symptom onset and diagnosis were 4.5 years (1–16) and 6 years (1.5–17), respectively. The median follow-up duration was 3.2 years (2–17). During attacks, fever was present in 155 patients (96.8%), abdominal pain in 148 (92.5%), arthralgia in 136 (85%), and chest pain in 33 (20.6%). Forty-five patients (28.1%) were considered colchicine-resistant. The median disease severity score was 2 (0–8); moderate disease severity was detected in 48 patients (30.0%) and severe disease in 8 patients (5.0%). The median craniocaudal spleen diameter was 95 mm (65–158), the median transverse diameter was 80.7 mm (15–158), and the median anteroposterior diameter was 33 mm (20–62). Splenomegaly was detected in 20 patients (12.5%). Although there was no significant age difference between colchicine-resistant and non-resistant groups, spleen dimensions were significantly greater in the colchicine-resistant group (craniocaudal $p$=0.01, transverse $p$=0.02, anteroposterior $p$=0.03) (Table 1). Compared with patients with mild disease severity, patients in the moderate-to-severe disease group had significantly greater craniocaudal spleen diameter [103.81 (73.5–158) vs. 93 (65.4–142.9), $p$=0.004], transverse diameter [86.5 (49.1–128) vs. 80 (15–117), $p$=0.003], and higher spleen Z-scores (0.48 ± 1.45 vs. 0.47 ± 1.11, $p$=0.034).

**Conclusion**

The association between increased spleen size and colchicine resistance in patients with FMF suggests that splenomegaly may reflect subclinical inflammatory burden. However, larger prospective studies are needed to clarify the clinical significance and utility of this finding.

**Disclosure of interest**

None declared.

## P271

### Comparison of inflammatory indices, spleen size, and enthesis thickness during attack and attack-free periods in patients with familial Mediterranean fever

N. Z. Özaslan[*,1], N. Şahin[1], H. E. Sonmez[1]
[1]Division of Pediatric Rheumatology, Faculty of Medicine, Kocaeli University, Kocaeli, Türkiye
**Correspondence:** N. Z. Özaslan
*Pediatric Rheumatology 2026*, **24(S1):**P271

**Introduction**

Familial Mediterranean Fever (FMF) is an autoinflammatory disease characterized by recurrent fever and serositis attacks, with persistent subclinical inflammation during attack-free periods. Increased spleen size, enthesis thickening, and alterations in inflammatory indices may reflect ongoing inflammation.

**Objectives**

To compare clinical findings, spleen size, enthesis thicknesses, and inflammatory indices in FMF patients during attack and attack-free periods.

**Methods**

This prospective study included 36 FMF patients evaluated during both attack and attack-free periods. Clinical, laboratory, and ultrasonographic findings were prospectively recorded. Disease activity was assessed

using C-reactive protein (CRP), erythrocyte sedimentation rate (ESR), systemic immune-inflammation index (SII), systemic inflammation response index (SIRI), neutrophil-to-lymphocyte ratio (NLR), platelet-to-lymphocyte ratio (PLR), lymphocyte-to-monocyte ratio (LMR), monocyte-to-lymphocyte ratio (MLR), and platelet-to-neutrophil ratio (PNR). Spleen size and enthesis thicknesses were measured by ultrasonography and compared between attack and attack-free periods.

**Results**

A total of 36 patients (21 females, 15 males) were included in the study. The median age was 11 years (5–17), median age at symptom onset was 5 years (1–13), and median age at diagnosis was 6.5 years (2–14). The median follow-up duration was 4.7 years (1–12.3). Regarding MEFV mutations, 27 patients (75.0%) had exon 10 homozygous mutations and 9 patients (25%) had exon 10 heterozygous mutations. During attacks, all patients (100%) experienced fever, abdominal pain, and arthralgia, while 12 patients (33.3%) also had chest pain. Exercise-related leg pain was present in 25 patients (69.4%), anemia in 15 (41.7%), arthritis in 6 (16.7%), history of appendectomy in 4 (11.1%), and erysipelas-like erythema in 1 patient (2.8%). Eighteen patients (50%) were considered colchicine-resistant.The mean disease severity score was 3 (1–7).

| | During Attack (*n*=36) | Attack-Free Period (*n*=36) | *p* value |
|---|---|---|---|
| **Laboratory markers** | | | |
| CRP, mg/L | 21.65 (14.35–50.02) | 1.82 (0.71–2.63) | <0.001 |
| ESR, mm/h | 15.50 (10.25–26.75) | 7.50 (3.00–11.00) | <0.001 |
| Leukocyte, $mm^3$ | 7315 (5550–8780) | 7250 (6112–8265) | 0.9 |
| Platelet, $mm^3$ | 305,972 ± 94,819 | 315,944 ± 73,468 | 0.312 |
| SII | 491,029 (291548–794681) | 363,584 (255729–641323) | 0.027 |
| SIRI | 1202 (531–2654) | 606 (457–970) | 0.016 |
| NLR | 1.71 (1.02–2.58) | 1.19 (0.97–1.75) | 0.010 |
| PLR | 129.1 (95.2–161.8) | 119.83 (80.11–141.07) | 0.033 |
| LMR | 3.94 ± 1.93 | 5.19 ± 1.76 | 0.002 |
| PNR | 82.14 ± 34.88 | 93.16 ± 29.36 | 0.116 |
| **Spleen Measurements (mm)** | | | |
| Craniocaudal diameter | 104.43 ± 16.45 | 97.67 ± 12.19 | 0.003 |
| Transverse diameter | 86.00 ± 14.60 | 86.32 ± 13.81 | 0.088 |
| Anteroposterior diameter | 35.94 ± 4.79 | 35.08 ± 5.52 | 0.341 |
| **Enthesis Measurements (mm)** | | | |
| Distal quadriceps tendon | 5.63 ± 3.52 | 5.03 ± 0.83 | 0.323 |
| Proximal patellar tendon | 3.74 ± 0.54 | 3.84 ± 0.65 | 0.214 |
| Distal patellar tendon | 3.58 ± 0.46 | 3.56 ± 0.56 | 0.843 |
| Achilles tendon | 4.17 ± 0.69 | 4.29 ± 0.80 | 0.285 |
| Plantar fascia | 2.97 ± 0.33 | 3.01 ± 0.37 | 0.387 |

**Conclusion**

Comparison of attack and attack-free periods in FMF patients showed significant increases in inflammatory markers, particularly CRP, ESR, and inflammatory indices (SII, SIRI, NLR, PLR, and LMR), as well as spleen size during attacks. However, no significant differences were observed in enthesis thicknesses between the two periods. These findings suggest that, in addition to acute phase reactants and inflammatory indices, spleen size may also reflect disease activity and acute inflammation.

**Disclosure of interest**

None declared.

## P272

## Long-term follow-up of patients with periodic fever, aphthous stomatitis, pharyngitis, and cervical Adenitis Syndrome: preliminary report

Z. Torunoglu[1,*], O. T. Akpinar[2], E. Pinar[2], G. Aslan[2], K. C. Demirbas[2], E. Aslan[1], N. Akay[1], U. Gul[1], E. Kilic Konte[1], E. Aslan[1], A. Gunalp[1], M. Yildiz[1], S. Sahin[1], K. Barut[1], O. Kasapcopur[1]

[1]Division of Pediatric Rheumatology, Department of Pediatrics; [2]Department of Pediatrics, Istanbul Unıversity-Cerrahpasa, Cerrahpasa Faculty of Medicine, Istanbul, Türkiye

**Correspondence:** Z. Torunoglu

*Pediatric Rheumatology 2026*, **24(S1):**P272

**Introduction**

Periodic fever, aphthous stomatitis, pharyngitis, and cervical adenitis (PFAPA) syndrome is the most common non-monogenic autoinflammatory disease of childhood, typically presenting before 5 years of age with recurrent febrile episodes lasting 3–7 days and recurring every 3–8 weeks. Although PFAPA is generally considered a self-limited disorder, symptoms may persist into adolescence and adulthood, and long-term outcome data remain limited.

**Objectives**

The aim of this study was to evaluate the long-term clinical outcomes of patients with PFAPA syndrome and to define the spectrum of coexisting and incident diseases observed during follow-up.

**Methods**

This retrospective single-center cohort study included PFAPA syndrome patients at Istanbul University-Cerrahpasa Cerrahpasa Medical Faculty pediatric rheumatology department. Therapeutic efficacy was assessed retrospectively from medical records and categorized as effective, partially effective, or ineffective. Fever resolution was considered the absence of any periodic fever episodes in the past year, whereas persistent fever was defined as the continued occurrence of regularly recurring febrile episodes separated by asymptomatic intervals within the same period.

**Results**

A total of 148 patients were included, of whom 90 (60.8%) were male. The mean age at the last follow-up was 14.7 ± 2.7 years, and the median follow-up duration was 11.0 years (IQR 9.8–12.9). Complete symptom resolution was achieved in 141 patients (95.3%), with a median symptom duration of 36.0 months (IQR 18.0–60.0) and a median age at the last PFAPA episode of 60.0 months (IQR 48.0–84.0). Four patients (2.7%) had persistent symptoms, with a median symptom duration of 97.5 months (IQR 59.3–105.8). Colchicine was used in 30 patients (20.3%), with complete, partial, and no response rates of 56.7%, 16.7%, and 26.7%, respectively. Among 93 patients (62.9%) who underwent tonsillectomy or adenotonsillectomy, all experienced marked symptom improvement, and 86 (92.5%) achieved complete resolution. A family history of Familial Mediterranean Fever and Behçet's disease was present in 12.8% and 8.1% of patients, respectively. Concomitant FMF was identified in three patients (2.0%), with persistent PFAPA symptoms in only one.

**Conclusion**

In long-term follow-up, the vast majority of patients with PFAPA achieved spontaneous remission without sequelae. Persistent symptoms were uncommon, while tonsillectomy and colchicine were associated with favorable outcomes in selected patients.

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**Disclosure of interest**

None declared.

## P273

### Clinical phenotype evolution from childhood to adolescence in familial Mediterranean fever: a retrospective cohort study

Z. Torunoglu[1,*], S. Sahin[1], M. E. Bayram[2], M. Gokden[2], D. D. Tastan[2], E. Aslan[1], N. Akay[1], U. Gul[1], E. Kilic Konte[1], E. Aslan[1], A. Gunalp[1], M. Yildiz[1], K. Barut[1], O. Kasapcopur[1]
[1]Division of Pediatric Rheumatology, Department of Pediatrics; [2]Department of Pediatrics, Istanbul Unıversity-Cerrahpasa, Cerrahpasa Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** Z. Torunoglu
*Pediatric Rheumatology 2026*, **24(S1):**P273

**Introduction**
Familial Mediterranean Fever (FMF) is the most common monogenic autoinflammatory disease across the world. It is characterized by recurrent episodes of fever lasting 12–72 h accompanied by self-limiting polyserositis attacks. In clinical practice, we observe that the disease phenotype in FMF may vary during adolescence.
**Objectives**
Based on this, we aimed to evaluate the demographic and clinical characteristics of patients with FMF and to compare attack frequency, duration, and clinical manifestations across the pre-diagnosis, pre-adolescent, and adolescent periods within the same cohort.
**Methods**
This retrospective cohort study included 221 adolescents with FMF carrying homozygous or compound heterozygous pathogenic exon 10 MEFV variants who continued to experience attacks. Clinical characteristics were compared within the same patients across three periods: pre-diagnosis, post-diagnosis pre-adolescence, and adolescence under treatment.
**Results**
A total of 221 patients were included, 51.1% female. Median ages at symptom onset and diagnosis were 36 and 60 months, respectively, with a median diagnostic delay of 12 months. Fever and abdominal pain were the most frequent manifestations overall. Fever and abdominal pain were most common before diagnosis, whereas arthritis, erysipelas-like erythema, and chest pain were more frequent during adolescence. Attack duration increased significantly during adolescence ($p<0.001$). Regular colchicine use decreased from 92.7% after diagnosis to 72.9% during adolescence ($p<0.001$). Menstrual-related attacks were reported by 27.4% of female patients.
**Conclusion**
In our study, the partial increase observed in attack frequency and particularly in attack duration during adolescence suggests that this period may represent a more vulnerable phase in terms of disease activity. These findings highlight the importance of closely monitoring treatment adherence during adolescence, reassessing dose optimization, and maintaining closer clinical follow-up. Based on our findings, we conclude that the currently available classification criteria may not be equally applicable across all age groups. Therefore, future criteria sets should consider age-specific clinical priorities, emphasizing fever and abdominal pain in the pre-adolescent period and chest pain and erysipelas-like erythema during adolescence.

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**Disclosure of interest**
None declared.

## 274

### Serological immunity against vaccine-preventable diseases in children with rheumatologic diseases

I. Grafas[1], L. F. Pittet[1], G. Blanchard-Rohner[*,1]
[1]Geneva University Hospital, Geneva, Switzerland
**Correspondence:** G. Blanchard-Rohner
*Pediatric Rheumatology 2026*, **24(S1):**P274

**Introduction**
Children with rheumatologic diseases on immunosuppressive therapy are at heightened risk for vaccine-preventable infections, yet vaccination coverage and serological monitoring remain suboptimal in this population.
**Objectives**
To evaluate vaccination coverage and baseline seroprotection at diagnosis, and to describe longitudinal serological trends during follow-up.
**Methods**
Retrospective review of medical records from children under 16 years diagnosed with a rheumatologic disease at Geneva University Hospitals (HUG) between 2005 and 2023, who received immunosuppressive therapy and had an available vaccination record. Vaccination status was classified as up-to-date per the Swiss national schedule. Seroprotection was defined by established laboratory thresholds for tetanus, diphtheria, Hib, measles, varicella, pneumococcus, and hepatitis B (HBV).
**Results**
74 patients were included (median age 6 years; 73% female); diagnoses included JIA (juvenile idiopathic arthritis, 51%), uveitis (12%), and FMF (familial Mediterranean fever, 10%). Vaccination coverage at diagnosis exceeded 80% for diphtheria, tetanus, Hib, and MMR, but was suboptimal for varicella (72%) and HBV (64%). Baseline serology was performed in only 51% of patients; seroprotection was high for tetanus (91%), diphtheria (88%), Hib (100%), and measles (89%), but substantially lower for varicella (47%), pneumococcus (42%), and HBV (58%). Longitudinal monitoring was inconsistent; diphtheria and tetanus protection remained stable, while varicella and pneumococcus showed heterogeneous trends. Some patients remained measles-seronegative throughout follow-up.
**Conclusion**
Children with rheumatologic diseases frequently begin immunosuppressive therapy with incomplete vaccination coverage and undetected serological gaps, particularly for varicella, pneumococcus, and HBV. Systematic baseline serology and structured longitudinal surveillance are essential to reduce preventable infectious morbidity, in line with EULAR/PRES recommendations.
**Disclosure of interest**
None declared.

## P275

### High physician trust despite low vaccination coverage in children with immune-mediated diseases: a dual-perspective cross-sectional survey

O. Lomakina[1,*], S. Valieva[1,2], M. Soloshenko[3,4], S. Kurbanova[1]
[1]Morozov City Children's Clinical Hospital; [2]Federal State Autonomous Educational Institution of Higher Education «N.I. Pirogov Russian National Research Medical University of the Ministry of Health of the Russian Federation, Moscow; [3]ITMO University, M.Sc. Programme in Applied Genomics, St. Petersburg; [4]Alekseev Psychiatric Clinical Hospital No. 1, Moscow, Russian Federation
**Correspondence:** O. Lomakina
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**Introduction**
Children with rheumatic and other immune-mediated diseases face increased risk of severe vaccine-preventable infections, yet immunization coverage in this population remains suboptimal.
**Objectives**
To characterize vaccination practices and associated barriers among parents of children with rheumatic diagnoses and physicians managing these patients.

**Methods**
A cross-sectional survey was conducted among parents of children with established rheumatic diagnoses (*n*=256) and physicians — pediatric rheumatologists, general pediatricians, and gastroenterologists (*n*=56), recruited as a convenience sample. Questionnaires covered vaccination practices, information sources, safety perceptions, and barriers. Proportions were calculated using respondents to each question as the denominator. Between-group comparisons used chi-squared or Fisher's exact test. Wilson 95% CI were calculated for physician proportions. Odds ratios with 95% CI estimated the association between cited physician deferral and complete non-vaccination.
**Results**
Among parents, 221/256 (86.3%) identified the treating physician as the most trusted source for vaccination decisions. Of 209 who answered the vaccination question (47 missing), 18 (8.6%) reported full adherence to the national schedule, 45 (21.5%) selective vaccination, and 145 (69.4%) no vaccinations after diagnosis. Predominant diagnoses were oligoarticular JIA (32.4%), polyarticular JIA (28.5%), and systemic JIA (18.4%); vaccination rates did not differ across subgroups ($p>0.05$). Among 172 parents of incompletely vaccinated children who reported reasons, 124 (72.1%) cited a physician recommendation to defer. The association between cited deferral and complete non-vaccination was not significant (OR 1.48, 95% CI 0.70–3.15, $p=0.32$). After disease onset, 113 (44.1%) families discontinued vaccination entirely; 209 (81.6%) expressed fear of adverse effects; 74 (28.9%) considered vaccination a possible contributor to their child's disease. Among physicians, all 56 (100%) acknowledged vaccination necessity, but only 32 (57.1%, 95% CI 44.1–69.2%) actively recommended it at every visit. Only 20 (35.7%) rated their knowledge as sufficient; 35 (62.5%) as insufficient. Vaccination as a possible autoimmune trigger was considered plausible by 22 (39.3%). The most cited barriers were parental fear (69.6%) and lack of up-to-date guidelines (55.4%).
**Conclusion**
Low vaccination coverage coexists with high physician trust, pointing to structural barriers: self-reported knowledge gaps, inconsistent counseling, and absent standardized protocols. Priorities include continuing professional education and development of standardized family counseling algorithms.
**Disclosure of interest**
None declared.

## P276

### Specific fears as drivers of vaccine hesitancy in parents of children with rheumatic diseases

O. Lomakina[*,1], S. Valieva[1,2], M. Soloshenko[3,4]
[1]Morozov City Children's Clinical Hospital, Moscow, Russian Federation; [2]Federal State Autonomous Educational Institution of Higher Education «N.I. Pirogov Russian National Research Medical University» of the Ministry of Health of the Russian Federation, Moscow; [3]ITMO University, M.Sc. Programme in Applied Genomics, St. Petersburg; [4]Alekseev Psychiatric Clinical Hospital No. 1, Moscow, Russian Federation
**Correspondence:** O. Lomakina
*Pediatric Rheumatology 2026*, **24(S1):**P276

**Introduction**
Vaccine hesitancy among parents of children with chronic immune-mediated diseases differs fundamentally from the general anti-vaccination movement, often arising against an initially positive attitude toward immunization and driven by diagnosis-specific concerns. The mechanisms of this phenomenon in pediatric rheumatology have not been systematically studied.
**Objectives**
To characterize the structure of fears and beliefs related to vaccination among parents of children with rheumatic diseases and to assess their association with post-diagnosis immunization coverage.
**Methods**
A cross-sectional survey was conducted among parents of children with an established rheumatic diagnosis (*n*=256), recruited as a convenience sample. A structured questionnaire covered attitudes toward vaccination, specific concerns regarding adverse effects, beliefs about the role of vaccination in disease onset and flare, and actual immunization coverage after diagnosis. Attitude was operationalized by self-classification as positive, negative, or ambivalent. Proportions were calculated using respondents to each question as the denominator; 95% Wilson CI are reported. Between-group comparisons used chi-squared test; pairwise differences in fear proportions were assessed using z-test for two proportions.
**Results**
A pronounced gap between declared attitude and actual behavior was observed: among 100 parents with a positive attitude and available post-diagnosis data, 68 (68.0%, 95% CI 58.3–76.3%) did not vaccinate after disease onset. Coverage differed significantly across attitude groups (chi-squared=18.81, df=2, $p<0.001$): complete non-vaccination was 68.0% in positive, 58.7% in ambivalent, and 100% in negative groups. Overall, only 42 (16.4%) declared a negative attitude; 117 (45.7%) positive; 97 (37.9%) ambivalent. Of 208 who answered the coverage question (48 missing), only 18 (8.7%) received vaccinations per schedule; 145 (69.7%) received none. The most cited concern was disease flare — 216/256 (84.4%), significantly exceeding neurological complications — 20 (7.8%; z=17.38, $p<0.001$) and allergic reactions — 10 (3.9%). The belief that vaccination caused disease onset was held by 74 (28.9%); 63 (24.6%) were unsure. After disease manifestation, 113 (44.1%) families discontinued vaccination; only 54 (21.1%) adopted an individually agreed schedule.
**Conclusion**
Vaccine hesitancy in parents of children with rheumatic diseases has a specific structure shaped predominantly by fear of disease flare and beliefs about vaccination as a possible disease trigger, distinct from the general anti-vaccination movement. Educational interventions should target these diagnosis-specific concerns.
**Disclosure of interest**
None declared.

## P277

### Role of whole-body MRI in therapeutic decision-making in children with juvenile idiopathic arthritis

A. M. Pasini[*,1], M. Šenjug Perica[1], D. Dujmović Plečko[1]
[1]Department of Paediatric Rheumatology, Children's Hospital Srebrnjak, Zagreb, Croatia
**Correspondence:** A. M. Pasini
*Pediatric Rheumatology 2026*, **24(S1):**P277

**Introduction**
Assessment of disease activity in juvenile idiopathic arthritis (JIA) remains challenging, particularly in patients with minimal symptoms or when treatment de-escalation is considered. Whole-body magnetic resonance imaging (WB-MRI) enables comprehensive evaluation of inflammatory and structural musculoskeletal changes, including clinically silent lesions.
**Objectives**
To evaluate the impact of WB-MRI findings on therapeutic decision-making during therapy with conventional/biologic disease-modifying therapy in children with JIA.
**Methods**
We retrospectively analyzed 13 children with JIA who underwent WB-MRI due to assessment of disease activity, persistent symptoms, or consideration of treatment reduction. Clinical characteristics, WB-MRI findings, and subsequent therapeutic modifications were reviewed.
**Results**
The cohort included 13 patients with JIA. WB-MRI demonstrated residual inflammatory changes in most patients, predominantly minimal joint effusions involving different peripheral joints. Atlantoaxial involvement was identified in three patients. WB-MRI findings directly influenced therapeutic decisions in all patients. Treatment escalation or intensification was performed in 5/13 patients, including initiation of etanercept in two patients, shortening of tocilizumab dosing intervals in two patients, and increase of sulfasalazine dosage in one patient. Current therapy was continued without reduction in 3/13 patients due to persistent symptoms and/or residual inflammatory findings despite clinically low disease activity. Treatment de-escalation was implemented in 5/13 patients, mainly by extending dosing intervals of adalimumab or methotrexate in patients with absent or minimal inflammatory activity on WB-MRI.
**Conclusion**
WB-MRI provided clinically relevant information that significantly influenced therapeutic management in children with JIA. Detection of subclinical inflammatory changes supported individualized treatment

decisions regarding treatment escalation, continuation, or de-escalation, particularly in patients with persistent pain and morning stiffness despite limited clinical findings. Atlantoaxial joint effusion and subluxation identified in several patients were considered particularly concerning findings because cervical spine involvement in JIA may be clinically silent yet associated with risk of structural instability and neurological complications. Recognition of these changes on WB-MRI contributed to therapeutic intensification in affected patients.

**Disclosure of interest**

None declared.

## P278

### Multimodal evaluation of systemic microvascular alterations in pediatric FMF by capillaroscopy and optical coherence tomography angiography

A. Doğru Kılınç[1,*], I. Uzunkulaoğlu[2], B. Başer Taşkın[1], F. G. Demirkan[3], Ö. Akgün[1], D. Tüzün Sayın[2], N. Aktay Ayaz[1]
[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine; [2]Ophthalmology; [3]Pediatric Rheumatology, Kanuni Sultan Suleyman Training and Research Hospital, Istanbul, Türkiye
**Correspondence:** A. Doğru Kılınç
*Pediatric Rheumatology 2026*, **24(S1):**P278

**Introduction**

Familial Mediterranean fever (FMF) is an IL-1β–mediated autoinflammatory disorder where persistent inflammation can cause endothelial dysfunction and systemic microvascular injury. The degree of subclinical microvascular involvement in pediatric FMF is unknown.

**Objectives**

This study evaluated subclinical microvascular alterations in FMF using nailfold capillaroscopy (NFC) and optical coherence tomography angiography (OCTA), comparing findings according to colchicine response.

**Methods**

This cross-sectional study included 61 pediatric FMF patients classified as Group 1 (colchicine-resistant, $n$=27) and Group 2 (colchicine-responsive, $n$=34) according to EULAR criteria. NFC was used to assess capillary density and morphology, while OCTA evaluated retinal and choroidal microvascular parameters, including vessel density (VD) in the superficial and deep vascular plexuses (SVP, DVP), central macular thickness (CMT), and subfoveal choroidal thickness (SFCT). Inter-group parameters and correlations between NFC and OCTA findings were analyzed.

**Results**

Group 1 had significantly earlier symptom onset and diagnosis, higher prevalence of clinical features (chest pain, arthritis, arthralgia, exercise-induced leg pain), and higher severity scores (ISSF and Pras) (all $p < 0.05$). Group 1 exhibited a higher frequency of non-specific capillaroscopic patterns (100% vs. 35.3%) and a lower capillary density (both $p < 0.001$). Crossing capillaries, tortuous capillaries, abnormal capillaries, and dilated capillaries were observed more frequently in Group 1 ($p < 0.001$, $p < 0.001$, $p < 0.001$, and $p = 0.008$). OCTA demonstrated significantly higher bilateral SFCT and CMT in Group 1 ($p<0.005$). Group 1 showed reduced DVP-VD across all regions, including the whole image, fovea, parafovea, and perifovea. ($p = 0.021$, $p = 0.034$, $p = 0.023$, and $p < 0.001$, respectively). Significant correlations were observed between NFC and OCTA parameters: capillary density correlated positively with DVP-VD, whereas abnormal capillary features correlated negatively with DVP-VD and positively with CMT ($p<0.05$).

**Conclusion**

Colchicine-resistant pediatric FMF is associated with more pronounced microvascular alterations in both peripheral and retinal circulations, likely reflecting persistent inflammation related microvascular injury.

**Disclosure of interest**

None declared.

## P279

### Amide proton transfer (APT) CEST MRI of the knee in juvenile idiopathic arthritis: a prospective pilot study

R. Raupov[1,2,*], T. Trofimova[1,3], M. Belikova[1,3], V. Sharakova[1], M. Kostik[4]
[1]BELOOSTROV Clinic of High Technologies; [2]H. Turner National Medical Research Center for Children's Orthopedics and Trauma Surgery; [3]Saint Petersburg State University; [4]Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation

**Table 1 (Abstract P279)** Key MRI measures in active vs. inactive knee arthritis

| Parameter | Active ($n$=7) median (IQR) | Inactive ($n$=16) median (IQR) | $p$ value |
|---|---|---|---|
| Synovial thickness, mm | 3.0 (2.25–3.0) | 1.0 (1.0–1.13) | 0.0006 |
| Max enhancement, % | 225 (212.5–252.5) | 95 (65–125) | 0.017 |
| APT_mean, % | 1.44 (1.24–2.22) | 0.68 (0.35–1.60) | 0.175 |
| APT_max, % | 2.09 (1.80–3.18) | 1.25 (0.82–2.53) | 0.095 |
| APTratio | 5.14 (2.80–5.74) | 2.34 (1.53–3.59) | 0.088 |

**Correspondence:** R. Raupov
*Pediatric Rheumatology 2026*, **24(S1):**P279

**Introduction**

Dynamic contrastenhanced (DCE) MRI is an established tool for quantifying synovitis in juvenile idiopathic arthritis (JIA), but requires gadolinium administration and remains insensitive to some aspects of tissue composition. Amide proton transfer (APT) CEST MRI is a noninvasive metabolic technique that may reflect synovial protein content and microenvironment, but its role in pediatric arthritis is unknown.

**Objectives**

To assess the feasibility and exploratory value of APTCEST MRI of the knee in children with JIA and to compare APTderived metrics between clinically active and inactive joints, in relation to conventional morphological and DCEMRI parameters.

**Methods**

Single-center prospective pilot study included 23 children with JIA and prior or current involvement of the knee. At the time of MRI, 7 knees were clinically active and 16 clinically inactive. All patients underwent standardized knee MRI including anatomical sequences, DCEMRI and APTCEST. Quantitative measures comprised synovial thickness, DCE parameters (maximum enhancement, enhancement curve type) and APT metrics: mean APT signal in synovium (APTmean), maximum APT (APTmax) and APT ratio between synovium and synovial fluid (APTratio).

**Results**

Synovial thickness was markedly higher in active versus inactive knees (median 3.0 vs. 1.0 mm, $p$=0.0006). DCEMRI showed higher maximum enhancement (225 vs. 95%, $p$=0.017) and a less favourable enhancement curve type in active joints. APT metrics were increased in active knees: APTmean 1.44 vs. 0.68%, APTmax 2.09 vs. 1.25% and APTratio 5.14 vs. 2.34, with moderate–large effect sizes (Cliff's delta 0.38–0.46), although p values did not reach conventional significance (0.09–0.18) in this small sample. APT_mean correlated moderately with synovial thickness (rho=0.44, $p$=0.034), whereas correlations between APT metrics and DCE maximum enhancement were weak and nonsignificant. (see Table 1)

**Conclusion**

APTCEST MRI of the knee in children with JIA is feasible and shows higher APT values in clinically active joints, with moderate associations to synovial thickening but not to DCE enhancement in this pilot cohort. These findings suggest that APTCEST may capture complementary aspects of synovial inflammation and justify larger studies to validate APTbased metrics as potential biomarkers of disease activity in pediatric arthritis.

**Disclosure of interest**

None declared.

## P280

### Capillaroscopic alterations in idiopathic perniosis in the pediatric and adolescent population

S. M. Murias[1,*], M. Loredo[2], V. Courel[1], E. Pardo[3], I. Molinos[2], O. Salcedo[1], A. Gómez[2], N. Callejas[2], P. Álvarez[2], J. Rodríguez[1]
[1]Unit of Pediatric Rheumatology; [2]Rheumatology; [3]Rheumatology, Unit of Pediatric Rheumatology, University Hospital Central of Asturias, Oviedo, Spain
**Correspondence:** S. M. Murias
*Pediatric Rheumatology 2026*, **24(S1):**P280

**Introduction**

Periungual capillaroscopy is a key tool in rheumatology for the evaluation of the microcirculation, especially in Raynaud's phenomenon. Idiopathic perniosis is a vascular acrosyndrome related to cold exposure,

common in the pediatric population, in which capillaroscopic alterations of uncertain and generally transient significance have been described. Characterization of these findings is relevant for the differential diagnosis of connective tissue diseases in this population.

**Objectives**

To describe capillaroscopic findings in pediatric and adolescent patients with a clinical diagnosis of perniosis and to analyze their potential usefulness in the differential diagnosis with connective tissue diseases.

**Methods**

• Descriptive observational study of 11 pediatric/adolescent patients with a clinical diagnosis of perniosis, evaluated in pediatric rheumatology and transition clinics.

• Demographic variables, ANA positivity, and capillaroscopic findings were collected, including hemorrhages, dilations, megacapillaries, branching, decreased density, and presence of a visible plexus.

**Results**

Eleven patients aged between 11 and 22 years were included, 8 females and 3 males. Four patients (36.3%) had positive ANA. The most frequent capillaroscopic findings were the presence of a visible plexus (9/11; 81.8%) and capillary dilations (7/11; 63.3%), followed by hemorrhages (5/11; 45.4%). Decreased capillary density was observed in 3 patients (27.3%), and branching was found in 1 case (9.1%). No patient presented megacapillaries.

**Conclusion**

In idiopathic perniosis, nonspecific capillaroscopic alterations such as dilations or hemorrhages may be observed and should be interpreted with caution, always within the clinical and serological context; clinical follow-up is essential to establish their significance. In our series, the most frequent capillaroscopic finding was the presence of a visible plexus, and no megacapillaries or patterns suggestive of systemic sclerosis were identified.

**Disclosure of interest**

None declared.

## P281

### Applicability of OMERACT salivary gland ultrasound scoring systems in juvenile Sjögren's disease: a pilot multicenter study

S. Delli Noci[1], B. Quéré[2,3], L. Ravà[4], F. Anselmi[5], R. Nicolai[1], C. Bracaglia[1], F. De Benedetti[1], S. Magni Manzoni[1], L. Rossi-Semerano[5,*]

[1]Division of Rheumatology, IRCCS Ospedale Pediatrico Bambino Gesù Roma, Roma, Italy; [2]Rheumatology Department, CHU Hospital La Cavale-Blanche, Centre National de Référence des Maladies Auto-Immunes Rares (CERAINO); [3]LBAI, UMR1227, Univ Brest, Inserm, Brest, France; [4]Epidemiology, Clinical Pathways and Clinical Risk Unit, IRCCS Ospedale Pediatrico Bambino Gesù Roma, Roma, Italy; [5]Department of Pediatric Rheumatology, National Reference Centre for Auto-Inflammatory Diseases and Amyloidosis of Inflammatory Origin (CEREMAIA), Bicêtre Hospital, Assistance Publique-Hôpitaux de Paris (AP-HP), Le Kremlin Bicêtre, France

**Correspondence:** L. Rossi-Semerano

*Pediatric Rheumatology 2026*, **24(S1):**P281

**Introduction**

Salivary gland ultrasound (SGUS) is increasingly used in Sjögren's disease (SD), and Outcome Measures in Rheumatology (OMERACT) Grey Scale (GS) and Doppler scoring systems have been developed and validated in adult populations[1-4]; their applicability in pediatric patients has never been studied.

**Objectives**

To investigate the applicability of the OMERACT SGUS GS and Doppler scoring systems in juvenile Sjögren's disease (jSD).

**Methods**

Consecutive patients with clinically diagnosed jSD and patients seen at two tertiary care study centers without salivary gland involvement (control group) with less than 18 years were asked to participate in a 6 months period. The patients included underwent US of bilateral parotid and submandibular glands according to a standardized acquisition protocol. Images were scored using the OMERACT GS and Doppler semi-quantitative systems by 4 sonographers with different experience in SGUS, after calibration. Applicability was evaluated in terms of feasibility, acceptability, and reliability. Intra- and inter-reader agreement were assessed using Cohen's weighted κ, linear and quadratic weights, Fleiss' κ, Gwet's AC2 coefficient (<0.4 poor; 0.41-0.6 moderate, 0.61-0.8 good; >0.81 excellent agreement).

**Results**

SGUS was successfully completed in all patients (100%), 30 jSD patients and 21 controls, with a median examination time of 9 min (IQR 8–10) and excellent tolerability (97.5% fully acceptable). Intra-reader reliability was high for both GS and Doppler, with Cohen's weighted κ ranging from 0.86 to 0.96 for GS and from 0.86 to 0.98 for Doppler. Inter-reader agreement was substantial, with non- Fleiss' weighted κ of 0.775 (95% CI 0.68–0.87) for GS and 0.805 (95% CI 0.70–0.91) for Doppler. Weighted agreement analysis further confirmed these findings, with Gwet's AC2 values of 0.95 (linear) and 0.97 (quadratic) for GS, and 0.93 (linear) and 0.95 (quadratic) for Doppler. Reliability remained consistently high across readers with different levels of experience.

**Conclusion**

OMERACT SGUS scoring systems were feasible, well tolerated, and reproducible in pediatric patients with jSD by sonographers with different experience in SGUS. These findings support their use in routine clinical practice and suggest that SGUS may represent a reliable and easily implementable non-invasive tool for the assessment of salivary gland involvement in children, particularly in settings where more invasive procedures are difficult to perform.

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**Disclosure of interest**

None declared.

## P282

### Performance of OMERACT scoring systems of salivary gland ultrasound in juvenile Sjögren's disease: a multicenter pediatric study

S. Delli Noci[1], L. Ravà[2], B. Quéré[3,4], F. Anselmi[5], E. Marasco[1], A. Aquilani[1], F. De Benedetti[1], L. Rossi-Semerano[5], S. Magni Manzoni[1,*]

[1]Rheumatology Division; [2]Epidemiology, Clinical Pathways and Clinical Risk Unit, Bambino Gesù Children Hospital IRCCS, Rome, Italy; [3]Rheumatology Department, CHU Hospital La Cavale-Blanche, Centre National de Référence des Maladies Auto-Immunes Rares (CERAINO); [4]LBAI, UMR1227, Univ Brest, Inserm, Brest; [5]Department of Pediatric Rheumatology, National Reference Centre for Auto-Inflammatory Diseases and Amyloidosis of Inflammatory Origin (CEREMAIA), Bicêtre Hospital, Assistance Publique-Hôpitaux de Paris (AP-HP), Le Kremlin Bicêtre, France

**Correspondence:** S. Magni Manzoni

*Pediatric Rheumatology 2026*, **24(S1):**P282

**Introduction**

OMERACT Grey Scale (GS) and Doppler scoring systems for the assessment of salivary glands ultrasound (SGUS) are increasingly used in routine evaluation of Sjögren's disease (SD) in adults. The validity of OMERACT SGUS scorings is unknown in children.

**Objectives**

To assess the diagnostic accuracy of SGUS OMERACT GS and Doppler scoring systems in distinguishing juvenile SD (jSD) from pediatric controls.

**Methods**

In this multicenter cross-sectional study, consecutive patients aged <18 years with clinically diagnosed jSD and controls without pathology of salivary glands underwent standardized US of parotid and submandibular glands. Images were scored according to the OMERACT GS and Doppler systems[1-2]. Diagnostic performance was evaluated by assess-

ing the ability of SGUS to discriminate patients from controls, using: 1.sensitivity and specificity of a GS-based Sjögren signature (GS≥2 in at least one gland)[3]; 2.ROC curve analysis for discriminative performance and optimal cut-offs of the OMERACT scores.

**Results**

Fifty-one subjects (30 patients, with median age at disease onset of 12 years [4.2-15.7] and median age at SGUS of 15 years [4.5-18.0] and 21 age- and sex-matched controls) were included across two European study centers. The GS-Sjögren signature showed a sensitivity of 66.7% and a specificity of 100%, indicating its high specificity for the identification of salivary gland involvement in jSD, despite only acceptable sensitivity. ROC analysis demonstrated excellent discriminative ability of the OMERACT scoring systems. The total score achieved an AUC of 0.937, with an optimal cut-off of 7.5 (sensitivity 87%, specificity 95%). Both GS and Doppler scores also showed high diagnostic accuracy with AUC 0.933 and 0.910, respectively.

**Conclusion**

OMERACT SGUS scoring systems demonstrated high discriminative performance for distinguishing pediatric patients with jSD from controls. The combined OMERACT scoring system showed better overall diagnostic performance than the GS-Sjögren signature alone. These findings support the potential role of SGUS as a valuable non-invasive tool in the diagnostic evaluation of suspected jSD.

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**Disclosure of interest**

None declared.

## P283

### Morphological salivary gland ultrasound features in juvenile Sjögren's disease: comparison with pediatric controls

S. Delli Noci[1], B. Quéré[2,3], L. Ravà[4], F. Anselmi[5], G. Tarantino[1], M. I. Petrone[1], F. De Benedetti[1], S. Magni Manzoni[1], L. Rossi-Semerano[5,*]
[1]Rheumatology Division, Bambino Gesù Children Hospital IRCCS, Rome, Italy; [2]Rheumatology Department, CHU Hospital La Cavale-Blanche, Centre National de Référence des Maladies Auto-Immunes Rares (CERAINO); [3]LBAI, UMR1227, Univ Brest, Inserm, Brest, France; [4]Epidemiology, Clinical Pathways and Clinical Risk Unit, Bambino Gesù Children Hospital IRCCS, Rome, Italy; [5]Department of Pediatric Rheumatology, National Reference Centre for Auto–Inflammatory Diseases and Amyloidosis of Inflammatory Origin (CEREMAIA), Bicêtre Hospital, Assistance Publique– Hôpitaux de Paris (AP–HP), Le Kremlin Bicêtre, France
**Correspondence:** L. Rossi-Semerano
*Pediatric Rheumatology 2026*, **24(S1):**P283

**Introduction**

Salivary gland ultrasound (SGUS) is increasingly used to assess glandular involvement in Sjögren's disease (SD); however data on US morphological features in juvenile SD (jSD) remain limited.

**Objectives**

To compare SGUS morphological features between patients with jSD and pediatric controls.

**Methods**

In this multicenter cross-sectional study, consecutive patients aged <18 years with clinically diagnosed jSD and controls without salivary gland disease underwent standardized US examination of parotid and submandibular glands. Predefined morphological items were assessed for each gland on grey-scale (GS), according to the MASAI protocol[1], including parenchymal homogeneity, echogenicity, hypoechoic areas (absent, <25%, 25–50%, >50% of gland surface), hyperechoic bands (with same grading), number of intraglandular lymph nodes and presence/absence of calcifications. Comparisons were performed for each item at gland level.

**Results**

Fifty-one subjects (30 patients, with median age at disease onset of 12 years [4.2-15.7] and median age at SGUS of 15 years [4.5-18.0] and 21 age- and sex-matched controls) were included across two European study centers. In parotid glands, abnormal echogenicity and inhomogeneity were markedly more frequent in jSD than controls ($p$<0.001). Hypoechoic areas were significantly associated with jSD, often with localized or diffuse distribution, whereas controls mainly showed absent or minimal changes (right $p$=0.012; left $p$=0.007). Hyperechoic bands were more frequent in patients, although also detected in controls (right $p$=0.003; left $p$=0.106). Similarly, submandibular glands showed significant differences in echogenicity and homogeneity between the two groups (all $p$<0.001), with hypoechoic areas more commonly observed in jSD (right $p$=0.007; left $p$=0.002). In contrast, hyperechoic bands did not significantly differ between the two groups. No significant differences were observed in the number of intraglandular lymph nodes. Calcifications were rare in jSD and showed poor discriminatory value.

**Conclusion**

SGUS showed several morphological differences in jSD, mainly related to inhomogeneity, reduced echogenicity and hypoechoic areas across both parotid and submandibular glands. Hyperechoic bands appeared less specific; lymph nodes and calcifications could not discriminate between groups. Morphological SGUS assessment may provide practical support for the characterization of salivary gland involvement in children and could contribute to the early identification of suspected jSD in clinical practice.

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**Disclosure of interest**

None declared.

## P284

### Capillaroscopic assessment of microcirculation in IgA vasculitis

K. Koman[1], V. Opoka-Winiarska[*,1]
[1]Department of Pediatric Pulmonology and Rheumatology, Medical University of Lublin, Lublin, Poland
**Correspondence:** V. Opoka-Winiarska
*Pediatric Rheumatology 2026*, **24(S1):**P284

**Introduction**

IgA vasculitis (IgAV) is characterised by inflammation of small blood vessels. The primary site of the vasculitis is the skin. Capillaroscopy is an examination dedicated to the assessment of microcirculation in the skin of the nailfolds. Nailfold capillaroscopy has a proven role in the diagnosis of scleroderma spectrum diseases. However, data on the assessment of microcirculation in vasculitis are limited.

**Objectives**

The aim of the study was to characterize the microcirculation of the nailfold in patients with IgA vasculitis in the acute phase of the disease. The obtained results were compared with clinical data.

**Methods**

The study included patients diagnosed with IgA vasculitis according to the EULAR 2010 criteria. The capillaroscopic examination was performed on the first day of diagnosis, before treatment. The examination was performed with a Dino-Lite 2.0 video capillaroscope at 50x and 200x magnifications. For this preliminary analysis, we assessed microcirculation on 2 mm of the 4th finger nailfold of the non-dominant hand. The density of vascular loops, morphology, vessel size and the presence of micro-extravasations were assessed. The microcirculation pattern was determined[1]. The data from the study were related to clinical symptoms.

**Results**

The study included 38 children, 16 girls and 22 boys aged 3 to 15 years, average age of patients 6 years 10 months. All children in the study

group had typical skin lesions and arthritis, 14 children (36,8%) had gastrointestinal symptoms and only 3 (7,9%) had kidney involvement. The most common abnormal feature of microcirculation was a slight decrease in vessel density, which we found in 10 subjects (26,3%). Dilated vessels occurred in 2 (5,2%) patients and megacapillaries in none. We also did not detect any micro-extravasations. We did not find a scleroderma pattern in any patient. We found nonspecific microangiopathy in 11 subjects (28,9%). At this stage of observation, we did not find any association between microvascular changes and clinical symptoms. We are conducting further prospective observation.

**Conclusion**

Almost one third of patients with IgA vasculitis develop nonspecific microangiopathy according to the EULAR criteria[2] at the beginning of the disease. These abnormalities appear to be different than in patients with scleroderma spectrum diseases, therefore other features of microcirculation should be analyzed. Standard capillaroscopic evaluation does not seem to provide information related to disease manifestation, but it may reveal new data on vascular changes important for understanding the pathogenesis of the disease. We are conducting further prospective observation. Images and other data require further analysis, perhaps with the help of AI.

Consent of the bioethics committee no. KE-0254/53/03/2024.
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**Disclosure of interest**

None declared.

## P285

### Pulmonary involvement in juvenile-onset connective tissue diseases: a multicentre retrospective cohort study

C. Zinterl[1,2], B. P. Correia[1,2,*], C. Durão[3], S. Azevedo[4], I. Cunha[4], J. B. Lima[5], S. Sousa[2,6], V. Fraga[6], M. J. Cadório[7], J. Oliveira[7], J. Silva-Dinis[8], H. Sousa[9], F. Aguiar[10], F. Oliveira-Ramos[1,2], R. Campanilho-Marques[1,2]
[1]Paediatric Rheumatology Unit, Rheumatology Department, ULS Santa Maria; [2]Faculdade Medicina; [3]Thoracic Department, Pulmonology, ULS Santa Maria, Lisbon; [4]Rheumatology, ULS Aveiro, Aveiro; [5]Paediatric Rheumatology, ULS Santo António, Oporto; [6]Paediatric Rheumatology, ULS Almada-Seixal, Almada; [7]Rheumatology, Hospital de Coimbra, Coimbra; [8]Rheumatology, ULS São José, Lisbon; [9]Paediatrics, ULS Estuário do Tejo, Vila Franca de Xira; [10]Paediatric Rheumatology, ULS São João, Oporto, Portugal
**Correspondence:** B. P. Correia
*Pediatric Rheumatology 2026*, **24(S1):**P285

**Introduction**

Juvenile-onset connective tissue diseases (jCTDs) are rare and their pulmonary complications, in contrast to adult-onset disease, are scarcely described.

**Objectives**

To characterise the prevalence, serological and clinical associations of pulmonary involvement in jCTDs.

**Methods**

This multicentre retrospective cohort study included patients with juvenile systemic lupus erythematosus (jSLE), juvenile mixed connective tissue disease (jMCTD), juvenile-onset systemic sclerosis (jSSc), juvenile idiopathic inflammatory myopathies (jIIM), juvenile Sjögren syndrome (jSS), and overlap syndromes, followed between January 2000 and August 2025. Demographic, clinical, serological, and treatment data were collected. Pulmonary involvement was assessed using high-resolution computed tomography and pulmonary function tests. Incidence rate was calculated as new events per 100 person-years (95% CI) and prevalence as frequencies. Associations were tested using Chi-Square or Fisher's Exact Test (significance level 0.05).

**Results**

We included 276 patients (77.5% female), with median age at disease onset 13.5 years [IQR 11-16] and median disease duration 11 years [6-15.0]. Diagnoses included jSLE (*n*=181, 65.6%), jMCTD (*n*=13, 4.7%), jSSc (*n*=14, 5.1%), jIIM (*n*=38, 13.8%), jSS (*n*=15, 5.4%), and overlap syndromes (*n*=13, 4.7%). Pulmonary involvement occurred in 11.6% (*n*=32), with the highest prevalence observed in jSSc (35.7%), jMCTD (23.1%) and overlap syndromes (23.1%). Interstitial lung disease was the most common manifestation (*n*=10), predominantly nonspecific interstitial pneumonia, followed by pleural (*n*=7) and vascular (*n*=6) involvement. Median FVC was 93% [85-103] and median TLC was 97% [89-105] of predicted. Most patients received cDMARDs (95.5%), particularly mycophenolate mofetil (40.5%), and 23% received biologics, mainly rituximab (13.5%); anti-fibrotic therapy was used in two patients. The incidence rate of pulmonary involvement was 0.62 per 100 person-years (95% CI 0.35–1.0). No specific autoantibody profile or jCTD subtype predicted pulmonary disease.

**Conclusion**

Pulmonary involvement in jCTDs is infrequent but significant, with variable patterns across diseases. The absence of serological predictors supports systematic pulmonary monitoring in routine follow-up.

**Disclosure of interest**

None declared.

## P286

### Transitional care experience in juvenile idiopathic arthritis and risk of relapse: a prospective monocentric study

L. Gatti[1,*], I. Pagnini[1], J. Blagojevic[2], J. Levani[2], I. Maccora[1], E. Marrani[1], M. V. Mastrolia[1], S. Guiducci[2], G. Simonini[1,3]
[1]Rheumatology Unit, ERN-ReCONNET center, Meyer Children's Hospital IRCCS; [2]Department of Experimental and Clinical Medicine, University of Florence, Azienda Ospedaliera Universitaria di Careggi; [3]University of Florence, NEUROFARBA Department, Florence, Italy
**Correspondence:** L. Gatti
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**Introduction**

Paediatric and adult rheumatologists share a common area in Transitional care. In 2016, EULAR/PReS provided the transitional care shared recommendations for young people with rheumatic diseases (1).

**Objectives**

We describe our experience in the transitional process in Juvenile Idiopathic arthritis.

**Methods**

Observational study carried at Meyer Children's Hospital and Careggi University Hospital from July 2020 since July 2025 in: (a) retrospective analysis of patients followed at Transitional care clinic, and (b) prospective analysis of patients transited to adult care system. Clinical data were entered in a customized database.

**Results**

43 JIA patients (27 females, 62.8%) with mean age of 18.33 (IQR 18.08-18.83) were enrolled: 15 patients had oligo-arthritis, 14 polyarthritis, 9 enthesitis-related arthritis, 4 psoriasic arthritis and 1 systemic JIA. At the first transition visit, the median disease duration was 8.2 years (IQR 3.8-12.9) and 69.7% patients were on treatment (csDMARDS and/or bDMARDS). After the transition patients were followed in the adult rheumatology clinic for 31 (IQR 21-39) months and no patients were lost at follow-up. During the follow-up 8 patients (18.6%) presented a relapse; 3 patients on csDMARDs started bDMARD (anti-TNF alpha) while 5 patients already on biologic drugs were switched to different biologic agent or JAK inhibitor. Univariate analysis risk factors for relapse were represented by ANA positivity ($p$ <0.015) and active treatment (cDMARD/bDMARD) at the moment of the transition visit ($p$<0.047). Tapering of bDMARDS/csDMARDS was not performed in any patient during the follow-up due to short follow-up period.

**Conclusion**

Our transition model resulted in high rate of successful due to careful planning of the transition and close collaboration between paediatric and adult rheumatologists. Risk factors for disease relapse resulted active treatment at transition visit and ANA positivity.

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**Disclosure of interest**
None declared.

## P287

### Characteristics of patients with immune-mediated inflammatory diseases hospitalized in intensive care unit

N. Abramova[1], I. Avrusin[2], L. Firsova[3,*], A. Mikhaylova[2], O. Kozlova[4], Y. Aleksandrovich[1], M. Kostik[2]
[1]Anesthesiology, Resuscitation and Emergency Pediatrics; [2]Hospital Pediatrics; [3]Propaedeutics of Childhood Diseases named after V. Yuriev, Saint Petersburg State Pediatric Medical University; [4]Clinical Mycology, North-Western State Medical University Named after I.I. Mechnikov, Saint Petersburg, Russian Federation
**Correspondence:** L. Firsova
*Pediatric Rheumatology 2026*, **24(S1):**P287

**Introduction**
Systemic immune-mediated inflammatory diseases (sIMID) may have extremely severe course with complications, including death.
**Objectives**
To characterize patients with sIMID, hospitalized in intensive care unit (ICU).
**Methods**
Medical records of 51 patients (23 boys; 28 girls), aged 121 [60;182] months, hospitalized in the ICU, were analysed. There were 24 patients with systemic rheumatic diseases (SRD), 18 – with multisystem inflammatory syndrome in children (MIS-C), and 9 – with sepsis. The age, sex, duration of hospitalization, laboratory data, hemophagocytic indices, and the therapy were assessed.
**Results**
All the results will be presented in the order: MIS-C, SRD, sepsis groups. Children with SRD were elder (106 [64; 137], *175 [115; 193]*, 41 [19; 60] months, $p<0.01$) and had longer time before hospitalization in ICU from administration to the hospital (6 [4; 10]; *30 [3; 28]*, 7 [7;8], $p<0.01$). However, the longest stay in hospital was in sepsis group (17 [14; 26], 28 [19; 68], *32 [25; 50]*, $p=0.01$). In MIS-C group only CRP levels were higher (*173.6 [129.6; 235.3]*, 66 [6.4; 113], 131 [101; 213] mg/L, $p=0.006$). In sepsis group, the following parameters were higher: maximal hart rate (120 [102;140], 112 [110; 140], *137 [125; 150]*, $p=0.049$), AST (61 [30; 137], 41.6 [22; 63.6], *135 [91.5; 208]* U/L, $p=0.02$), bilirubin (11.6 [7.3; 15.9], 8.7 [4.6; 11.2], *13.2 [10.1; 37.2]* mcmol/L, $p=0.02$), D-dimer (7.9 [3.9; 13.3], 2.23 [0.7; 9], *14.45 [8.4; 16.7]* mg/L, $p=0.049$). In SRD group, creatinine levels (41.1 [31.2; 121.3], *121.9 [62.4; 179]*, 45 [42; 87] mcmol/L, $p=0.03$) and the incidence of corticosteroid therapy were higher (13%, *23%*, 4%, $p=0.004$). SRD group demonstrated the lowest levels of hemoglobin (79 [67;101] g/L), AST (41,6 [22;63.6] U/L), bilirubin (8.7 [4.6; 11.2] mcmol/L) and D-dimer (2.23 [0.77;9]) levels. Hemophagocytic indices did not differ. Significant differences in therapy and outcomes are presented in Table 1.
**Conclusion**
Thus, the obtained data highlight the need for an individualized approach to therapy in patients with immune-mediated inflammatory diseases.
**Disclosure of interest**
None declared.

**Table 1 (Abstract P287)** Differences in therapy and outcomes

| Parameter, *Me (25%; 75%) | MIS-C | SRD | Sepsis | *p* |
|---|---|---|---|---|
| Immunosuppressive therapy initiated in ICU, n (%) | 16 (88.9) | 9 (37.5) | 4 (44.4) | **0.003** |
| Following Immunosuppressive therapy, n (%) | 18 (100.0) | 19 (79.2) | 6 (66.7) | **0.051** |
| Delayed invasive mycosis, n (%) | 0 (0) | 6 (25) | 0 (0) | **0.022** |
| Lethal outcome, n (%) | 1 (7.7) | 10 (41.7) | 2 (15.4) | **0.028** |

## P288

### Gaps in transition care: limited and delayed transition discussions in pediatric rheumatology

L. Berben[1,2,*], A. D. Chop[1], L. Kirkpatrick[1], T. M. Kazmerski[1], K. Torok[1]
[1]Department of Pediatrics, University of Pittsburgh School of Medicine, Pittsburgh, USA; [2]University Children's Hospital Basel, Basel, Switzerland
**Correspondence:** L. Berben
*Pediatric Rheumatology 2026*, **24(S1):**P288

**Introduction**
Structured transition from pediatric to adult healthcare is imperative for adolescents and young adults with chronic rheumatologic conditions. However, healthcare transition (HCT) practices remain inconsistently implemented. Evaluating current HCT practices and identifying gaps within clinical settings is essential to improve continuity of care and long-term outcomes.
**Objectives**
To describe characteristics of HCT practices and compare the characteristics of patients with or without documented HCT discussions at a single academic pediatric rheumatology clinic and its adult counterpart.
**Methods**
We retrospectively reviewed the electronic health records (EHRs) of people aged 16–26 years with a diagnosis of a pediatric rheumatologic disease and at least one consultation in the pediatric rheumatology clinic within the past two years. We examined patient demographics, HCT outcomes and HCT discussion documentation. We descriptively analyzed results. Independent sample t-test were used to compare the characteristics of patients with and without documentation of HCT discussions.
**Results**
We included 473 patients with a chronic rheumatologic condition (68% female; 60% with juvenile idiopathic arthritis). The mean age at the most recent rheumatology clinic visit was 19.6 years (SD 2.5; range 16–26). Overall, 20% of patients transferred to adult care, 65% remained in pediatrics, and 13% were lost to follow-up. The mean age at transfer was 22 years (SD 2.5, range 17–28). HCT discussions were not documented in 75% of EHRs, with variation across groups (pediatric care: 84%; adult care: 40%; lost to follow-up: 84%). Patients with documented HCT discussions were significantly older than those without (mean difference= 2.21 years, $p< 0.001$).
**Conclusion**
HCT discussions are infrequently documented among patients with chronic rheumatologic conditions, particular among those remaining in pediatric care or lost to follow-up, highlighting substantial gaps in current HCT practices. The association between older age and documented discussions suggests that HCT planning may occur later than recommended. These findings underscore the need for earlier integration of HCT care into routine clinical practice.
**Disclosure of interest**
None declared.

## P289

### Transitional care in Swiss rheumatology: findings from the HEROES study in adolescents and young adults

L. Berben[1,2,*], M. Matter[1], M. Schraner[1], M.-L. Daly[1,2], L. Strahm[1], T. Daikeler[2], A. Woerner[1] on behalf of the HEROES Study Team
[1]University Children's Hospital Basel; [2]University Hospital Basel, Basel, Switzerland
**Correspondence:** L. Berben
*Pediatric Rheumatology 2026*, **24(S1):**P289

**Introduction**
Successful transition from pediatric to adult care in adolescents and young adults (AYAs) requires structured transitional care (TC) programs. In Swiss rheumatology clinics, TC practices vary considerably across centers. The Rheumatology Transition for Young People in Switzerland (HEROES) study aims to develop, implement, and evaluate a standardized TC program across Swiss rheumatology centers. The study is recruiting AYAs and collecting baseline data to inform program development and enable future evaluation of its effectiveness.
**Objectives**
To describe baseline characteristics of participating AYAs.

**Methods**
Data were collected using interviewer-administered questionnaires, including standardized and investigator-developed instruments. Health-related quality of life (HRQoL) was assessed using the EQ-5D-3L, and medication adherence using the Basel Assessment of Adherence to Immunosuppressive Medications Scale (BAASIS©).
**Results**
A total of 75 participants from seven pediatric rheumatology clinics in German-speaking Switzerland were included. Participants were aged 14–22 years (mean 17.0, SD 1.57). The majority of the participants were female (72%). Among 67 participants receiving medication, 25 (37.3%) reported missing at least one dose in the previous four weeks, and 9 (13.4%) reported missing three or more doses. School or work absenteeism in the past six months due to their rheumatic disease was reported by 69.3% of participants, with a mean of 8.9 days (range 0.5–80). The mean EQ-5D-3L utility score was 0.82 (SD 0.22; range 0.12–1.00), indicating overall good HRQoL with substantial variability. Problems were most frequently reported in the pain/discomfort (44%) and anxiety/depression (40%) dimensions. The mean EQ VAS score (self-rated health, 0–100) was 75.8 (SD 18.8; median 78).
**Conclusion**
Baseline findings highlight relevant challenges in medication adherence, disease burden, and HRQoL among AYAs with rheumatic diseases. These results provide important guidance for the development of a standardized TC program tailored to the needs of this population.
**Disclosure of interest**
None declared.

## P290

### Clinical profile and outcomes of children with juvenile idiopathic arthritis (JIA) receiving biological disease-modifying antirheumatic drugs (tocilizumab, adalimumab, etanercept): single-centre experience from Georgia

M. Lekishvili[1,*], M. Ioseliani[1], K. Mamamtavrishvili[1] on behalf of L. Melikidze, T. Feradze, K. Gvinefadze, C. Fanchulidze
[1]Center of Adults and Pediatric Rheumatology, New Hospitals, Tbilisi, Georgia
**Correspondence:** M. Lekishvili
*Pediatric Rheumatology 2026*, **24(S1):**P290

**Introduction**
Juvenile idiopathic arthritis (JIA) is the most common chronic rheumatic disease of childhood, and biologic disease-modifying antirheumatic drugs (bDMARDs) have significantly improved disease outcomes.
**Objectives**
We aimed to evaluate bDMARD use and treatment outcomes among patients with JIA in Georgia.
**Methods**
A total of 569 patients were included in this study between 2015 and 2025 years. The most frequent subtype was oligoarticular (57.1%), polyarticular (18.3%), systemic (19.5%), enthesitis related arthritis (ERA) (2.9%), juvenile psoriatic arthritis (JPsA) (2.1%). ANA positive was 122 (21.4%), with eye injuries 41 (7.2%). 206 patients of them treated with biologic therapy. We used etanercept (98 ), adalimumab(24 ), tocilizumab ( 105 ), 21 patient used 2 or 3 of them.
**Results**
In 206 patients who took biological DMARDs, with a female predominance ( 67.9%). subtupes requiring biologic therapy- oligoarticular (41.1%), polyarticular (25.9%), systemic (26%), ERA (2.9%), JPsA (4.1%). Significant improvement in clinical and laboratory disease activity parameters was observed after biologic treatment initiation. remission without medication was achieved in 27.6% of patients, in 14.5% of them began flare mean time 7month(1-23mont) . Remission with medication was achieved in 55% after a median of 7.5 (4-16) weeks, 38% of them with monotherapy , while 24.7% showed partial response or inadequate response . The most striking advers events were allergic reaction in 9 cases (2 adalimumab, 7 tocilizumab), 1 case tuberculosis (etanercept) .
**Conclusion**
bDMARD therapy demonstrated favorable clinical outcomes in our centre patients with JIA, with most patients achieving remission or low disease activity. The introduction of biologic agents led to a reduction in both the duration and dosage of glucocorticoid therapy. Adverse events were mostly mild and manageable.
**Disclosure of interest**
None declared.

## P291

### Assessment of functional ability and quality of life in young adult patients with juvenile idiopathic arthritis (JIA): the impact of diagnostic delay

M. Stavrakidou[1,2,3,*], A. A. Garyfallos[2], T. Dimitroulas[2], M. Trachana[3], D. Dimopoulou[2]
[1]Asklepeio Physiotherapy Clinic; [2]4th Internal Medicine Clinic, Hippokration Hospital, Aristotle University, Clinic of Connective Tissue Diseases, Clinic for Adult Patients with Pediatric Rheumatic Diseases; [3]Hippokration Hospital, Aristotle University, First Dept of Pediatrics, Pediatric Immunology and Rheumatology Referral Center, THESSALONIKI, Greece
**Correspondence:** M. Stavrakidou
*Pediatric Rheumatology 2026*, **24(S1):**P291

**Introduction**
Juvenile Idiopathic Arthritis (JIA) often persists in adulthood, affecting physical and psychosocial well-being. Despite modern therapeutic options, diagnostic delay remains a challenge that may affect long-term disease outcomes. However, evidence regarding the impact of this delay on functional status and Health-Related Quality of Life (HRQoL) specifically in the young adult JIA population remains scarce in the international bibliography.
**Objectives**
The objective of this study is to assess functional capacity and HRQoL in young adult patients with JIA and to investigate their correlation with diagnostic lag time.
**Methods**
Assessment tools included the HAQ-DI for functional capacity and the SF-36 for HRQoL (Physical Health – PH, Mental Health – MH). Statistical analysis involved Pearson correlation and subgroup comparisons.
**Results**
31 adult patients (mean age 21.9 years) were evaluated. The mean diagnostic lag time for the entire cohort was 15.1 months. A significant negative correlation was found between lag time and both SF-36 PH ($r$ = -0.44, $p$ = 0.013) and MH ($r$ = -0.41, $p$ = 0.022) scores. Subgroup analysis revealed that patients with Enthesitis/spondylitis-related JIA (ERA) experienced a substantially longer diagnostic delay compared to other subtypes (29.3 vs. 8.4 months, $p$ = 0.002). The ERA subgroup demonstrated significantly higher functional disability (HAQ-DI: 0.31 vs. 0.01, $p$ = 0.004) and lower physical health scores (PH: 66.8 vs. 84.3, $p$ = 0.001).
**Conclusion**
Diagnostic delay is a major predictor of poorer functional and quality-of-life outcomes in young adults with JIA. Patients with the ERA subtype are particularly vulnerable to prolonged lag times, leading to increased long-term disability. These findings underscore the necessity for early diagnosis and the development of specialized therapeutic rehabilitation programs to mitigate long-term physical and functional burden.
**Disclosure of interest**
None declared.

## P292

### Beyond the acute phase: autoimmune and lymphoproliferative outcomes 20 years after Kawasaki disease

M. Hamad Saied[1,2,*], I. Halabi [1], R. Kasem Ali Sliman [1]
[1]Department of Pediatrics, Carmel Medical Center, Haifa, Israel; [2]Department of Pediatric Immunology and Infectious Diseases, Wilhelmina Children's Hospital/University Medical Center, Utrecht, Netherlands
**Correspondence:** M. Hamad Saied
*Pediatric Rheumatology 2026*, **24(S1):**P292

**Introduction**
Kawasaki disease (KD) is an acute systemic vasculitis of childhood characterized by immune dysregulation that may extend well beyond the acute inflammatory phase. However, the long-term risks of autoimmune diseases and lymphoproliferative malignancies in KD survivors remain poorly defined.

**Objectives**
To determine whether children with KD have elevated 20-year risks of autoimmune and lymphoproliferative outcomes compared with matched controls.
**Methods**
Retrospective matched cohort study using the Clalit Health Services database (2002–2022). Children with KD ($n$=2,126) were matched 1:5 to controls ($n$=10,630) by sex and birthdate (±30 days) and followed through December 2024. Outcomes included psoriasis, vitiligo, immune thrombocytopenia (ITP), Hodgkin lymphoma (HL), non-Hodgkin lymphoma (NHL), hypothyroidism, type 1 diabetes mellitus (T1DM), celiac disease, and inflammatory bowel disease (IBD). Cox proportional hazards models yielded adjusted hazard ratios (AHRs) with 95% confidence intervals at 2, 5, 10, 15, and 20 years.
**Results**
KD was associated with significantly elevated psoriasis risk from 2 years (AHR 3.03, 95% CI 1.19–7.73) through 15 years (AHR 1.50, 95% CI 1.01–2.23), attenuating to non-significance at 20 years. Vitiligo reached significance at 10 years (AHR 1.38, 95% CI 1.03–1.86). HL risk was markedly elevated at 15 years (AHR 14.99, 95% CI 1.55–145.12) and 20 years (AHR 7.95, 95% CI 1.32–48.00). No significant associations were found for hypothyroidism, T1DM, ITP, celiac disease, IBD, or NHL.
**Conclusion**
KD is associated with selective, long-term elevations in psoriasis, vitiligo, and HL risk, each with distinct temporal patterns, supporting pathway-specific immune dysregulation rather than generalized autoimmunity. These findings support targeted dermatologic and hematologic awareness in KD survivors.
**Disclosure of interest**
None declared.

## P293

### Long-term outcomes of transition from pediatric to adult care in familial Mediterranean fever: a comparative analysis of clinical and treatment characteristics

Z. Torunoglu[1,*], E. Aslan[1], N. Akay[1], U. Gul[1], E. Kilic Konte[1], E. Aslan[1], A. Gunalp[1], M. Yildiz[1], S. Sahin[1], K. Barut [1], S. Ugurlu[2], O. Kasapcopur[1]
[1]Division of Pediatric Rheumatology, Department of Pediatrics; [2]Division of Rheumatology, Department of Internal Medicine, Istanbul Unıversity-Cerrahpasa, Cerrahpasa Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** Z. Torunoglu
*Pediatric Rheumatology 2026*, **24(S1):**P293

**Introduction**
Familial Mediterranean fever (FMF) is the most common monogenic autoinflammatory disease and requires lifelong follow-up. Transition of care is a structured process that transfers adolescents with chronic diseases from pediatric to adult healthcare services.
**Objectives**
This study aimed to evaluate the long-term follow-up results of the transition process from pediatric to adult rheumatology care in FMF patients carrying a biallelic pathogenic variant in exon 10 of the MEFV gene and to compare the clinical course, follow-up, and treatment characteristics between the pediatric and adult rheumatology care.
**Methods**
This retrospective single-center study included 107 patients with childhood-onset FMF who transitioned from pediatric to adult rheumatology care through a joint transition clinic between 2014 and 2023. Clinical and treatment characteristics were compared before and after transition. Successful transition was defined as at least one adult rheumatology visit within the first year, and active follow-up as at least one visit within the past year.
**Results**
A total of 107 patients were included, 51.4% of whom were female, with a mean age of 29.1 ± 2.7 years. Median pre- and post-transition follow-up durations were 13 (9–16) and 7 (5–8) years, respectively. Successful transition to adult rheumatology care was achieved in 100 patients (93.4%), although only 74 (69.2%) remained in active follow-up. The most common genotype was M694V homozygosity (64.5%). Attack prevalence decreased significantly after transition (97.2% vs. 77.6%, $p$<0.001). Abdominal pain, fever, arthritis, and arthralgia were significantly less frequent in post-transition period, whereas chest pain and erysipelas-like erythema remained unchanged. No patient developed amyloidosis.
**Conclusion**
In conclusion, this study is the first to include a homogeneous FMF cohort carrying two allelic pathogenic variants in exon 10 of the MEFV gene and to present long-term follow-up data after transition from the pediatric to the adult rheumatology care, comparing clinical characteristics, follow-up and treatment differences in the same patient group. Future prospective, multicentre studies incorporating standardised transition protocols will reveal in greater detail the true impact of structured transition programmes on FMF patients, not only on initial access to adult care but also on long-term disease control, treatment compliance, subclinical inflammation management, and continuity of follow-up.

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**Disclosure of interest**
None declared.

## P296

### Implementation and clinical impact of pharmacogenetics, therapeutic drug monitoring and MedReview in pediatric rheumatology and gastroenterology patients

P. Dalla Zuanna[1], G. Paternuosto[1], C. Debora[1], A. Parzianello[2], M. Klanjscek[2], R. Cecchin[3], S. Sindici Forgiarini[2], M. Franzin[1], B. Bellich[1], G. Ponis[1], M. Lucafò[2], R. Franca[2], A. Fabretto[1], R. Addobbati[1], A. Tommasini[1], A. Taddio[1,*], S. Pastore[1], M. Bramuzzo[1], P. Rossi[4], M. P. Trojniak[1], J. Polesel[3], G. Decorti[2], E. Cecchin[3], A. Arbo[5], G. Stocco[1]
[1]Institute for Maternal and Child Health – IRCCS "Burlo Garofolo"; [2]University of Trieste, Trieste; [3]Centro di Riferimento Oncologico di Aviano (CRO) IRCCS, Aviano; [4]Centro Regionale di Farmacovigilanza, Friuli Venezia Giulia; [5]Institute for Maternal and Child Health – IRCCS "Burlo Garofolo", Aviano, Italy
**Correspondence:** A. Taddio
*Pediatric Rheumatology 2026*, **24(S1):**P296

**Introduction**
TDM, pharmacogenetics (PGx) and MedReview are precision medicine tools that optimize drug efficacy and reduce adverse drug reactions (ADRs).
**Objectives**
This study evaluates how TDM, PGx and MedReview impact pediatric drug efficacy and safety through clinical counseling. It further reports the prevalence of out-of-range drug levels and actionable genotypes in children.
**Methods**
NCT06822959, a multicenter prospective study (IRCCS Burlo Garofolo/CRO Aviano, Italy), enrolled pediatric rheumatology and gastroenterology patients on adalimumab, infliximab, methotrexate (MTX) or azathioprine (AZA). Biologics TDM used immunochromatography/ELISA; AZA (TGN/MMPN) used HPLC-DAD; MTX (ITPA and polyglutamates) used HPLC-UV and LC-MS/MS. Ten TaqMan-analyzed variants defined actionable genotypes. MedReview evaluated drug-drug interactions by INTERCheck/Lexicomp/Medscape. A counseling provided to clinicians addressed TDM, PGx and interactions. Efficacy (clinical scores) and ADRs (CTCAE ≥2 or grade 1 causing therapy changes) were analyzed via Kaplan–Meier and log-rank tests.
**Results**
Enrolled 162 pediatric patients (median 12.9y, 90 F; 119 rheumatology, 43 gastroenterology) over 776 visits (2023–2025). We performed 742 TDM, 902 pharmacogenetic analyses and 739 counselings. Without the study, only 39.5% of TDM, 14.3% of pharmacogenetics and no counseling would have been performed. TDM was out-of-range in 35.5%; 32.8% of genotypes were actionable (94.4% of patients had ≥1). Identified 17 interaction types and 69 relevant ADRs. Treatment changes

occurred in 25 patients for ADRs and 25 for lack of efficacy. In monotherapy, subtherapeutic TDM linked to shorter failure-free survival ($p$=0.025), while supratherapeutic TDM (excluding biologics given their predominant dose-independent toxicity) linked to shorter toxicity-free survival ($p$=0.014).

**Conclusion**

Integrating TDM, PGx and MedReview optimizes pediatric prescribing by identifying risks for treatment failure or ADRs. High rates of out-of-range drug levels and actionable genotypes confirm their clinical utility. PGx and MedReview analyses are ongoing.

**Disclosure of interest**

None declared.

## P297

### Familial paradoxical flu-like reactions to anti-TNF therapy in HLA-B27-positive enthesitis-related arthritis: a possible pharmacogenetic predisposition

P. Dalla Zuanna[1], C. Debora[1], G. Caiffa[2], M. Lucafò[2], A. Tommasini[1], A. Taddio[1,*], M. Faleschini[1], A. M. R. Bianco[1], A. P. D'adamo[1], G. Stocco[1], S. Pastore[1]

[1]Institute for Maternal and Child Health – IRCCS "Burlo Garofolo"; [2]University of Trieste, Trieste, Italy

**Correspondence:** A. Taddio

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**Introduction**

Anti-TNF agents effectively treat enthesitis-related arthritis (ERA), yet paradoxical reactions and immunogenicity are rising concerns. Genetic variants, specifically *FCGR3A* and *HLA-DQA1*, may drive anti-drug antibody (ADA) formation and these associated adverse events.

**Objectives**

To describe a familial case of recurrent systemic reactions to multiple anti-TNF agents in pediatric ERA and explore possible pharmacogenetic mechanisms.

**Methods**

Clinical history and pharmacogenetic data were retrospectively reviewed in a 17-year-old girl with HLA-B27-positive ERA and her mother affected by ankylosing spondylitis, both experiencing adverse reactions to anti-TNF therapy. Whole exome sequencing was performed.

**Results**

While on adalimumab (40 mg/2 weeks), the patient developed recurrent flu-like reactions (fever, arthralgia) ~12 h post-dose, unresponsive to prednisone and causing discontinuation after 7 months. Subsequent etanercept (50 mg/weekly) triggered similar episodes (fever, arthralgia, asthenia, headache, pain with ocular movement) 6–10 h post-injection, lasting 48–72 h. Laboratory/ophthalmologic results were normal; dose reduction failed. TDM was not performed. Because similar reactions to anti-TNF agents had also occurred in the patient's mother, pharmacogenetic testing was performed, revealing in both subjects *FCGR3A* rs396991 homozygous VV genotype, *HLA-DQA1* exon 2 variants (p.C34Y, p.F41S, p.R70W), and *TNFRSF1B* rs1061622 heterozygous variant (p.M196R), with an additional *TNFRSF1B* rs3397 3'UTR variant in the mother. Literature data indicate that the *FCGR3A* VV genotype combined with *HLA-DQA1* risk variants confers an approximately 10-fold increased risk of ADA development against monoclonal anti-TNF agents such as infliximab and adalimumab, potentially enhancing immune-mediated adverse events.

**Conclusion**

This case suggests an inherited predisposition to paradoxical systemic reactions during anti-TNF therapy, likely driven by enhanced Fcγ receptor-mediated activation and immunogenicity linked to *FCGR3A* and *HLA-DQA1*, with a possible contributory role of *TNFRSF1B*. Occurrences with both adalimumab and etanercept support class-related immune dysregulation over molecule-specific intolerance. Pharmacogenetic profiling may identify pediatric patients at risk, enabling personalized strategies. Sanger confirmation is ongoing.

**Disclosure of interest**

None declared.

## P298

### Direct versus gradual colchicine initiation in pediatric familial Mediterranean fever: a multicenter comparative study

A. Doğru Kılınç[1,*], Z. Hazıroğlu Ökmen[2], B. Başer Taşkın[1], P. Prencuva Akyürek[1], S. Doğantan[2], A. Tanatar[3], F. G. Demirkan[4], Ö. Akgün[1], F. Çakmak[2], N. Aktay Ayaz[1]

[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine; [2]Pediatric Rheumatology, Basaksehir Cam and Sakura City Hospital; [3]Pediatric Rheumatology, Marmara University Faculty of Medicine; [4]Pediatric Rheumatology, Kanuni Sultan Suleyman Training and Research Hospital, Istanbul, Türkiye

**Correspondence:** A. Doğru Kılınç

*Pediatric Rheumatology 2026*, **24(S1):**P298

**Introduction**

Familial Mediterranean fever (FMF), a prevalent monogenic autoinflammatory disorder, is primarily treated with colchicine. While age-based target doses are well defined, there is no standardized approach to initiation, with either the full target dose or gradual dose escalation from a lower starting dose. Current EULAR guidelines support single or divided daily dosing but provide no consensus on the optimal initiation protocol.

**Objectives**

This study aimed to compare direct target dose initiation with gradual dose escalation of colchicine in FMF patients with respect to adverse event profiles, dose-reduction requirements, and treatment adherence.

**Methods**

This ongoing preliminary multicenter longitudinal study included pediatric FMF patients fulfilling the Eurofever criteria with at least 3 months of follow-up after colchicine initiation. Colchicine treatment was initiated according to EULAR recommendations. Patients were classified into either the Gradual Escalation Group (Group 1), in which the dose was increased weekly by 0.5 mg/day until the target dose was reached, or the Direct Group (Group 2), in which the target dose was initiated directly. Demographic characteristics, baseline and follow-up laboratory parameters, and colchicine-related adverse events were recorded for all participants.

**Results**

The study included 38 patients: Group 1 (G1, $n$=24) and Group 2 (G2, $n$=14). All completed the first month, while 20 patients (G1: $n$=15, G2: $n$=5) completed the 3-month follow-up. No significant differences were observed between groups regarding sex, age at onset/diagnosis, family history, baseline attack frequency, or clinical manifestations. At day 15, diarrhea occurred in 7 patients (all G1; duration: 1–6 days); they were advised to wash the tablet coating. By month 1, diarrhea persisted in only 3 patients (mild) and completely resolved in all by month 3. Nausea affected 4 patients (all G1) at day 15; it persisted in 2 and newly developed in 1 at month 1, resolving fully by month 3. Vomiting occurred in 1 patient (G1) at day 15 only. Transient neutropenia developed in 2 patients (G1) and resolved spontaneously. Myalgia was reported in 1 patient at day 15 and 2 at month 1 (all G1); none had elevated creatine kinase, and all resolved by month 3. No adverse events were reported in Group 2.

**Conclusion**

Direct target dose initiation of colchicine in pediatric FMF patients is a safe and well-tolerated alternative to gradual dose escalation, as transient and mild adverse effects were exclusively observed in the escalation group and resolved spontaneously without requiring dose modifications.

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**Disclosure of interest**

None declared.

## P300

### Real-world effectiveness of secukinumab in juvenile idiopathic arthritis: a single-center observational study

D. Kairatova[1,*], M. Assylbekova[1], Z. Mukusheva[1], Z. Albekova[1], A. Dildabayeva[1], A. Akhmambetova[1]
[1]Rheumatology Program, Clinical Academic Department of Pediatrics, "University Medical Center" Corporate Fund, Astana, Kazakhstan
**Correspondence:** D. Kairatova
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**Introduction**
Secukinumab is an IL-17 inhibitor that has demonstrated its efficacy in enthesitis-related arthritis (ERA) and juvenile psoriatic arthritis (jPsA) [1]. However, real-world data remain limited regarding its effectiveness in patients who switched to secukinumab because of refractory disease courses or adverse effects from previous therapy.
**Objectives**
To assess the real-world effectiveness of secukinumab in JIA patients by evaluating disease activity using JADAS10 and inflammatory markers after switching therapy.
**Methods**
We conducted an ambispective observational study of 22 patients with JIA who were switched to secukinumab. Analysis included 13 patients who had available follow-up data. JADAS10, ESR, and CRP were evaluated at baseline and at 3-month intervals after treatment initiation when available. Continuous variables were summarized using median and interquartile range. Changes from baseline to the latest available follow-up were assessed using the Wilcoxon signed-rank test for paired observations.
**Results**
Of 22 patients switched to secukinumab at our center, only 13 had paired follow-up data. 3 were lost to follow-up, and 6 had not yet reached the 3-month assessment. Out of these 13 patients, 7 had ERA or jPsA, and 6 had other JIA, including polyarticular JIA in 5 patients and oligoarticular JIA in 1 patient. Three-month follow-up data were available for 13 patients, 6-month data for 8 patients, and 12-month data for 6 patients. The median follow-up time is 6 months [IQR 5-14]. Median JADAS10 decreased from 17 [13.8-20] at baseline to 4 [3-8] at the latest follow-up ($p$=0.002). Median ESR decreased from 21.5 [10.0-35.1] to 8.0 [6.0-12.0] mm/h ($p$=0.033), while CRP changed from 2 [0.4-12.0] to 1.08 [0.6-2.14] mg/L ($p$=0.635). Psoriatic skin lesions were controlled in patients with jPsA. Two patients received concomitant tofacitinib. Dose adjustments were required in three patients, and five patients had temporary treatment interruptions at some point in the treatment due to drug unavailability. At the latest follow-up, secukinumab was continued with all 13 patients. No adverse events were reported. Written informed consent was obtained from the participants and their parents.
**Conclusion**
Secukinumab was associated with an improvement in disease activity and ESR at the latest available follow-up, while CRP demonstrated no significant change. Further observation of newly switched patients and longer follow-up are needed to confirm these findings.

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**Disclosure of interest**
None declared.

## P301

### Can fasting improve methotrexate tolerance in JIA? A multicenter PERA study from Türkiye

Ç. Yildiz[1,*], B. Küçükali[1], B. Öksel[2], B. Menentoğlu[3], N. Kara Çanlıoğlu[4], E. S. Arslan Kocamaz[5], T. Güzel[6], H. K. Zora[7], T. Kasap[8], S. Şener[9], E. N. Dizman[10], E. Aliyev[11], H. Adıgüzel Dündar[12], H. E. Sönmez[2], N. Şahin[2], N. Aktay Ayaz[3], B. Sözeri[4], B. Kasap[5], F. Çakmak[6], S. S. Kılıç[7], S. Yüksel[8], K. Öztürk[10], D. Gezgin Yıldırım[1], S. A. Bakkaloğlu[1] on behalf of The Pediatric Rheumatology Academy-Research Group (PeRA-RG)
[1]Pediatric Rheumatology, Gazi University Faculty of Medicine, Ankara; [2]Pediatric Rheumatology, Kocaeli University Faculty of Medicine, Kocaeli; [3]Pediatric Rheumatology, İstanbul University İstanbul Faculty of Medicine; [4]Pediatric Rheumatology, Ümraniye Training and Research Hospital, İstanbul; [5]Pediatric Rheumatology, İzmir City Hospital, İzmir; [6]Pediatric Rheumatology, Başakşehir Çam And Sakura Cıty Hospital, İstanbul; [7]Pediatric Rheumatology, Uludağ University Faculty of Medicine, Bursa; [8]Pediatric Rheumatology, Çanakkale Onsekiz Mart University, Çanakkale; [9]Pediatric Rheumatology, Adana City Hospital, Adana; [10]Pediatric Rheumatology, Medeniyet University Faculty of Medicine, İstanbul; [11]Pediatric Rheumatology, Lokman Hekim University Faculty of Medicine, Ankara; [12]Pediatric Rheumatology, DR. BEHCET UZ PEDIATRIC DISEASES AND SURGERY TRAINING AND RESEARCH HOSPITAL, İzmir, Türkiye
**Correspondence:** Ç. Yildiz
*Pediatric Rheumatology 2026*, **24(S1):**P301

**Introduction**
Methotrexate (MTX) is a widely used disease-modifying antirheumatic drug in pediatric rheumatologic diseases (1). However, gastrointestinal and behavioral adverse effects may lead to intolerance, negatively affecting treatment adherence and outcomes (2). MTX intolerance, assessed by the Methotrexate Intolerance Severity Score (MISS), has been reported in 25–75% of patients (2,3).
**Objectives**
This study evaluated MTX intolerance in juvenile idiopathic arthritis (JIA), focusing on the impact of fasting versus non-fasting intake and associated demographic and clinical factors.
**Methods**
A total of 325 JIA patients from 12 centers in the Turkish Pediatric Rheumatology Academy (PeRA) study group were included. Demographic and MTX-related data were retrospectively collected from medical records. Fasting status, route of administration, and MTX intolerance assessed by MISS were analyzed in relation to demographic and clinical variables.
**Results**
Among 325 patients, 60.3% were female and oligoarticular JIA was the most common subtype (57.8%). MTX intolerance was associated with longer follow-up (39 vs. 24 months, $p$=0.0002) and longer MTX exposure (26.5 vs. 16 months, $p$=0.00008). Oral administration was linked to higher intolerance rates compared with subcutaneous use ($p$=0.016), and intolerance was more frequent in patients taking MTX with food ($p$=0.027). Nausea ($p$<0.001) and vomiting ($p$=0.001) were significantly associated with intake status, whereas abdominal pain and behavioral symptoms were not. A borderline association was observed between ALT elevation and intolerance ($p$=0.071), while no associations were found with AST elevation, cytopenia, demographic features, ILAR categories, or autoantibody status.
**Conclusion**
Our study demonstrated that MTX intolerance is more frequent in patients receiving oral MTX and in those taking the drug with food. Additionally, longer treatment duration was associated with higher intolerance risk. Nausea and vomiting were the most prominent manifestations of intolerance, whereas abdominal pain and behavioral symptoms were not significantly associated. Although a borderline association was observed with ALT elevation, no significant relationship was found with AST elevation or cytopenia. The lack of association with demographic features, ILAR categories, and autoantibody status suggests that MTX intolerance is primarily influenced by administration route, timing of intake, and treatment duration rather than disease phenotype. These findings suggest that administering MTX in a fasting state or via subcutaneous route may improve tolerability and potentially enhance treatment adherence in patients with JIA.

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**Disclosure of interest**
None declared.

## P302

### Interferon-targeted therapy with anifrolumab in monogenic pediatric systemic lupus erythematosus due to C1Q deficiency: a case report

S. M. Alqahtani[1,2,*], A. M. Asiri[2], M. A. Aljuaid[2]
[1]Pediatrics, Najran University, Najran; [2]Pediatric Rheumatology, Prince Sultan militray midical city, Riyadh, Saudi Arabia
**Correspondence:** S. M. Alqahtani
*Pediatric Rheumatology 2026*, **24(S1):**P302

**Introduction**
Monogenic systemic lupus erythematosus (SLE) secondary to complement deficiencies, including C1Q deficiency caused by C1QA mutations, is a rare and severe type of lupus that can be characterized by early onset and refractory disease. Inhibiting the interferon pathway has proved to be an effective treatment option, although there is little evidence in pediatric monogenic lupus.
**Objectives**
This case provides a clinically relevant teaching point: in pediatric patients with refractory lupus, particularly those with suspected or confirmed monogenic disease, targeted therapies based on underlying immunopathology should be considered. Precision medicine approaches may improve outcomes in this challenging population.
**Methods**
We describe a 10-year-old female with genetically verified C1Q deficiency who had persistent and severe mucocutaneous disease, with recurrent skin rash, ulcerations, and oral sores, despite long-term immunosuppressive and biologic treatment. After a disease flare, the patient was started on anifrolumab, a monoclonal antibody against type I interferon receptor. She showed significant clinical improvement and full recovery of mucocutaneous lesions, restoration of laboratory parameters, and successful discontinuation of corticosteroids after seventh monthly infusions. It was well tolerated with no serious side effects. The case demonstrates the possible effectiveness and safety of anifrolumab in the treatment of refractory cutaneous manifestations in children with monogenic lupus, which is linked to interferon pathway dysregulation. Further studies are needed to establish its role in this population.

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**Disclosure of interest**
None declared.

## P303

### Intravenous immunoglobulin in severe pediatric connective tissue diseases: a tertiary center experience

M. Correia Natal[1,2], F. Gomes[3], J. P. Moreira[4], P. J. Pereira[3], I. Cardoso[5], B. Fernandes Esteves[1,2], R. Nicolau[6], D. Milkowska Mikiel [7], S. Ganhão[8], F. Aguiar[2,8,*], M. Rodrigues[9], I. Brito[9]
[1]Rheumatology Department, ULS São João; [2]Department of Medicine, Faculty of Medicine, University of Porto, Porto; [3]Rheumatology Department, ULS Braga, Braga; [4]Department of Pediatric Cardiology, ULS São João, Porto; [5]Pediatrics Department, ULS Gaia/Espinho, Gaia/Espinho; [6]Rheumatology Department, ULS Trás-os-Montes e Alto Douro, Vila Real; [7]Pediatrics Department, ULS Matosinhos, Matosinhos; [8]Pediatric and Young Adult Rheumatology Unit; [9]Pediatrics Department, ULS São João, Porto, Portugal
**Correspondence:** F. Aguiar
*Pediatric Rheumatology 2026*, **24(S1):**P303

**Introduction**
Intravenous immunoglobulin (IVIG) is a widely used therapy with pleiotropic immunomodulatory and anti-inflammatory properties. It is used as adjunctive therapy in severe, refractory or infection-limited pediatric connective tissue diseases (CTD).
**Objectives**
To describe the indications, efficacy and safety of IVIG in pediatric CTD patients followed at a tertiary pediatric rheumatology unit.
**Methods**
Retrospective descriptive study of CTD patients treated with IVIG. Demographic data, diagnosis, indication, treatment regimen, clinical and laboratory response, corticosteroid exposure and adverse events were reviewed.
**Results**
Eight patients were included, 6/8 female, with a median age of 11.5 years (range 3–17). Diagnoses were juvenile systemic lupus erythematosus (JSLE, *n*=4), juvenile dermatomyositis (JDM, *n*=3) and overlap syndrome (*n*=1). IVIG was used for refractory disease (*n*=4), severe hematological relapse (*n*=2) and severe disease at onset (*n*=2), including one patient with concomitant bacterial infection. All patients showed clinical and laboratory improvement. In JDM, improvement in muscle strength was accompanied by a marked reduction in muscle enzymes (median CK 3219.5 to 222 U/L). In JSLE, hematological recovery was observed, including resolution of severe thrombocytopenia ($<10\times10^9$/L). Mean prednisolone dose decreased from 42.5 to 8.75 mg/day at 3 months. IVIG was well tolerated, with only one mild adverse event (self-limited fever) and no treatment discontinuations.

| Diagnosis | IVIG indication | Main manifestations | Outcome after IVIG |
|---|---|---|---|
| JDM | Refractory disease | Weakness, rash, dysphagia; CK 4601 U/L | Improved strength/dysphagia, rash resolution; CK 209 U/L |
| JDM | Refractory disease | Weakness, myalgia, dyspnea; CK 1087 U/L | Muscle strength normalization, symptom resolution; CK 114 U/L |
| JDM | Severe onset/ remission induction | Myalgia, inability to walk, edema, rash; CK 2269 U/L | Walking unaided, rash improvement; CK 235 U/L |
| JDM-JSLE overlap | Severe onset + infection | Fever, purpura, hepatosplenomegaly, rash; anti-dsDNA >800, CK 4170 U/L | Clinical stabilization, no new infections; anti-dsDNA 498, CK 1888 U/L |
| JSLE | Refractory bicytopenia | Leukopenia, thrombocytopenia | WBC $1.01\rightarrow6.81\times10^9$/L; PLT 43→201 |

| Diagnosis | IVIG indication | Main manifestations | Outcome after IVIG |
|---|---|---|---|
| JSLE | Refractory pancytopenia + MAS | Fever, pallor, pancytopenia; ferritin 1703 ng/mL | Asymptomatic; Hb/WBC/PLT recovery, ferritin 1218 ng/mL |
| JSLE | Refractory thrombocytopenia | Oral bleeding; PLT $<10\times10^9$/L | No new bleeding; PLT $350\times10^9$/L |
| JSLE | Severe onset, thrombocytopenia | Petechial rash; PLT $<10\times10^9$/L | Rash resolution; PLT $268\times10^9$/L |

**Conclusion**
In this cohort, IVIG proved to be a safe and effective option for the management of severe manifestations across different pediatric CTD. Acting as a valuable therapeutic bridge, it allowed rapid clinical stabilization and a significant corticosteroid-sparing effect. Its safety profile in severe and infection-prone settings further reinforces the relevance of IVIG as an adjunctive therapeutic option in pediatric rheumatology.
**Disclosure of interest**
None declared.

## P304

### Tumor necrosis factor-α inhibitor-related autoimmune disorders in pediatric rheumatology practice: a multicenter nationwide study

D. Gezgin Yildirim[1,*], B. Küçükali[1], N. Akay[2], S. N. Yoğun[3], E. Tunce[4], H. Taştanoğlu[5], A. Doğru[6], Y. E. Bayrak[7], S. Şener[8], M. O. Erkan[9], R. Torun[10], S. Ayduran[11], N. G. Kocamaz[12], E. Küçük[13], O. A. Akın[14], Ç. Yıldız[1], E. Aslan[2], F. Demirkan[15], B. Kasap Demir[16], R. M. Kışla Ekinci[17], S. Yüksel[18], S. Türkuçar[11], E. Sağ[9], N. Şahin[7], E. Çelikel[3], K. Öztürk[13], S. Ozdel[12], A. Paç Kısaarslan[5], N. Aktay Ayaz[6], B. Sözeri[4], E. Ünsal[10], Ö. Kasapçopur[2], S. A. Bakkaloğlu[1]
[1]Department of Pediatric Rheumatology, Gazi University Faculty of Medicine, Ankara; [2]Department of Pediatric Rheumatology, İstanbul University-Cerrahpaşa Faculty of Medicine, İstanbul; [3]Department of Pediatric Rheumatology, Ankara Bilkent City Hospital, Ankara; [4]Department of Pediatric Rheumatology, Umraniye Training and Research Hospital, İstanbul; [5]Department of Pediatric Rheumatology, Erciyes University Faculty of Medicine, kayseri; [6]Department of Pediatric Rheumatology, İstanbul University Faculty of Medicine, İstanbul; [7]Department of Pediatric Rheumatology, Kocaeli University Faculty of Medicine, Kocaeli; [8]Department of Pediatric Rheumatology, Adana City Hospital, Adana; [9]Department of Pediatric Rheumatology, Hacettepe University Faculty of Medicine, Ankara; [10]Department of Pediatric Rheumatology, Dokuz Eylül University Faculty of Medicine, İzmir; [11]Department of Pediatric Rheumatology, Pamukkale University Faculty of Medicine, Denizli; [12]Department of Pediatric Rheumatology, Ankara Etlik City Hospital, Ankara; [13]Department of Pediatric Rheumatology, İstanbul Medeniyet University Faculty of Medicine, İstanbul; [14]Department of Pediatric Rheumatology, Eskişehir Osmangazi University Faculty of Medicine, Eskişehir; [15]Department of Pediatric Rheumatology, Kanuni Sultan Süleyman Training and Research Hospital, İstanbul; [16]Department of Pediatric Rheumatology, İzmir City Hospital, İzmir; [17]Department of Pediatric Rheumatology, Çukurova University Faculty of Medicine, Adana; [18]Department of Pediatric Rheumatology, Onsekiz Mart University Faculty of Medicine, Çanakkale, Türkiye
**Correspondence:** D. Gezgin Yildirim
*Pediatric Rheumatology 2026*, **24(S1):**P304

**Introduction**
TNF-α inhibitors may cause intrinsic compensatory adaptive changes of the innate immune system, leading to TNF-α inhibitor-related autoimmune disorders (TIRAIDs).
**Objectives**
The purpose of the study is to evaluate the demographic characteristics, clinical features, and management strategies of TIRAIDs in pediatric rheumatology practice.
**Methods**
The medical records of 71 pediatric patients treated with a TNF-α inhibitor for a rheumatologic disease and who exhibited any TIRAIDs during a 10-year period were retrospectively evaluated.
**Results**
The prevalence of TIRAIDs was 2.8% among 2450 patients. There was a female predominance ($n$=40, 56%). TIRAIDs were observed in 49 patients (69%) receiving etanercept, 16 patients (23%) receiving adalimumab, and 6 patients (8%) receiving infliximab. The mean time to TIRAIDs was 21.6 (median 23.7) months. Non-infectious uveitis (38%) was the most common TIRAID, followed by paradoxical psoriasis (PP) (25%), lupus-like disease (8%), multiple sclerosis (MS) (7%), episcleritis (7%), and hidradenitis suppurativa (1%). In 58% of patients, treatment was switched to another TNF-α inhibitor, while in 42%, it was switched to a non–TNF-α biological therapy. Complete resolution of TIRAIDs was observed in 63% of the patients. C-reactive protein (CRP) levels and Juvenile Arthritis Disease Activity Score-27 (JADAS27) were significantly higher during the development of TIRAIDs in patients with juvenile idiopathic arthritis (JIA) compared to the remission period ($p = 0.000$ and $p = 0.041$, respectively).
**Conclusion**
TNF-α inhibitor-related autoimmune disorders (TIRAIDs) may develop after the prescription of TNF-α inhibitors. Among TIRAIDs, non-infectious uveitis and PP were the most frequently observed manifestations, predominantly occurring during etanercept treatment. Disease activity tended to increase during the onset of TIRAIDs. Discontinuation of TNF-α inhibitor therapy and switching to another biological treatment may be effective in achieving clinical remission. • Raising awareness about the risk of TIRAIDs during TNF-α inhibitor treatments and routinely monitoring patients is essential for early detection and effective treatment of TIRAIDs.
**Disclosure of interest**
None declared.

## P305

### Treatment resistant rapidly progressive childhood IgG4-related disease with orbital and sinonasal involvement responsive to radiotherapy

G. Kaymak[1,*], S. Kamer[2], M. Palamar[3], S. İmamoğlu[4], N. Gürbüz[1], S. Yücel[1], A. H. Kamiloğlu[1], E. Gülyiğit[1], T. Çetin Arıkan[1], G. Avcı Demirciler[1], T. Bozkurt Vardar[2], A. Berdeli[1], G. Aksu[1]
[1]Department of Pediatric Rheumatology; [2]Department of Radiation Oncology; [3]Department of Ophthalmology, Ege University; [4]Department of Pediatric Rheumatology, Izmir City Hospital, Izmir, Türkiye
**Correspondence:** G. Kaymak
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**Introduction**
IgG4-related disease (IgG4-RD) is a rare fibroinflammatory disorder, rare in children. Orbital involvement is relatively common, whereas sinonasal involvement is infrequent (1). Corticosteroids and steroid-sparing agents represent the first-line therapy. In refractory cases, an alternative therapeutic modality, although less commonly employed, is radiotherapy. However there is no currently established therapeutic option for long-term maintenance (2).
**Objectives**
We present a pediatric-onset case of IgG4-RD concomitant orbital and sinonasal manifestations demonstrating aggressive progression and resistance to conventional therapies, while highlighting the role of radiotherapy.
**Methods**
The clinical features, radiological findings, histopathological data and therapeutic outcomes of the patient were retrospectively evaluated.
**Results**
A seven-year-old female patient presented with strabismus and unilateral visual acuity loss. Radiological imaging revealed a destructive lesion infiltrating the sphenoid sinus, orbital apex, and cavernous sinus. Histopathological examination confirmed the diagnosis of IgG4-RD. Initial treatment with corticosteroids and methotrexate resulted in a partial response. Following a five-year stable period, disease was observed with bilateral optic nerve involvement and progressive visual acuity decline, coinciding with puberty and the initiation of oral contraceptives. Despite intensive immunosuppressive therapy including pulse methylprednisolone, mycophenolate mofetil, azathioprine, and rituximab, the disease showed exhibited refractory progression. Surgical resection was not indicated due to anatomical complexity. Low-dose radiotherapy (20 Gy) was subsequently initiated. The entire treatment was completed in 10 fractions, conducted every weekday.

Pre-radiotherapy best-corrected visual acuity continued to decrease; counting fingers from 30 cm in the right eye and counting fingers from 1 m in the left eye. Post-radiotherapy, radiological regression was observed, with partial recovery in visual acuity, thereby halting the progression; however, optic atrophy persisted.For this report, written informed consent was obtained from the patient's legal guardians for the publication of this case report and any related clinical images and data.

**Conclusion**

This case demonstrates that pediatric-onset IgG4-RD concomitant orbital and sinonasal manifestations can exhibit an aggressive and refractory course, highlighting the importance of early intervention. Puberty and hormonal factors may be implicated in disease relapse. Radiotherapy represents an effective therapeutic option in resistant cases and should be considered when conventional treatments are exhausted.

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**Disclosure of interest**

None declared.

## P306

### Janus kinase inhibitors for children with multiple epiphyseal dysplasias manifesting features of hips osteoarthritis

A. Kozhevnikov[1,2,*], E. Melchenko[1], V. Kenis[1]
[1]H. Turner National Medical Research Center for Children's Orthopedics and Trauma Surgery; [2]Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation
**Correspondence:** A. Kozhevnikov
*Pediatric Rheumatology 2026*, **24(S1):**P306

**Introduction**

Multiple epiphyseal dysplasia (MED) is a rare clinically and genetically heterogeneous skeletal disorder characterized by delayed ossification of epiphyses, leading to early-onset severe osteoarthritis of hips and knees. Clinical and instrumental manifestations of osteoarthritis in children with MED mainly mimic of juvenile idiopathic arthritis (JIA). Long term use of NSAIDs and methotrexate therapy didn't reduce inflammatory. According to some authors the use of DMARDs also is inefficacy.

**Objectives**

**The aim of this study** was to evaluate the efficacy of targeted synthetic DMARDs and inhibitors TNF-alpha in children with MED associated with severe hip osteoarthritis.

**Methods**

The study included 8 children with MED (3 children – MED type 1, COMP gene mut; 5 children – MED type 4, SLC26A2 gene mut; mean age 9,5 ± 1,0 years 87,5% girls). All children had clinical and MRI evidence of osteoarthritis of one or both hips and suffered from severe pain (≥ 60 mm on VAS) requiring daily NSAID. Four children received etanercept (0.8 mg/kg/week) and other – tofacitinib (10 mg/day). The average follow-up of DMARDs was 12 [8; 16] months. All children were tested negative for antinuclear antibodies (ANA) and HLA B27 gene. In our cohort sacroiliitis was not detected by MRI in any of the children.

**Results**

The analysis revealed that all children who received etanercept (two children with MED type 1 and two with MED type 4) regularly felt hip pain (≥ 40 mm on VAS). MRI detected persistent signs of osteoarthritis. Children treated of tofacitinib (one with MED type 1 and three with MED type 4) for more than 6 months were reducing of pain (≤ 20 mm on VAS) and NSAID requirements. Children who showed experience inadequate response to inhibitors TNF-alpha were switched tofacitinib. After 6 months of targeted synthetic DMARDs treatment three children (two with MED type 1 and one with MED type 4) had reduction hip pain and decrease MRI signs of osteoarthritis. This fact served as the basis for the continuing treatment with targeted synthetic DMARDs. Analysis of instrumental picture showed that DMARDs included of targeted synthetic drugs and inhibitors TNF-alpha in long-term treatment prevented progressive of dislocation hip among children. However, jak inhibitors (78,5%) showed significant benefits in reducing hip pain and osteoarthritis sings as measured by MRI.

**Conclusion**

The use of anti-inflammatory drugs for long-term in children with MED manifesting features of severe hips osteoarthritis debatable. Targeted synthetic DMARDs and TNF-alpha inhibitors are considered as one of the ways to suppress of osteoarthritis activity, reduce pain and prevent the development of hip dislocation. Evidence-based choice for long-term used jak inhibitors may be rational in children with MED.

**Disclosure of interest**

None declared.

## P307

### Late-onset agranulocytosis manifesting as gingival swelling following B-cell depletion therapy with rituximab or inebilizumab: two case reports

K. Yamazaki[1,*], M. Mouri[1], E. Toyofuku[1], K. Kawahata[1]
[1]Department of Rheumatology and Allergology., St. Marianna University School of Medicine, Kawasaki, Japan
**Correspondence:** K. Yamazaki
*Pediatric Rheumatology 2026*, **24(S1):**P307

**Introduction**

B-cell depletion therapy (BCDT), including anti-CD20 antibody (rituximab) and anti-CD19 antibody (inebilizumab), has been associated with late-onset neutropenia, defined as an unexplained reduction in absolute neutrophil count (ANC) occurring at least four weeks after the final BCDT administration.

**Objectives**

To report two cases in which gingival swelling was the initial manifestation of agranulocytosis during BCDT maintenance therapy.

**Methods**

**Case 1**

A 17-year-old female with childhood-onset neuromyelitis optica spectrum disorder and mixed connective tissue disease was receiving maintenance therapy with inebilizumab, prednisolone (4.5 mg/day), and cyclosporine. Five weeks after the last inebilizumab dose, gingival swelling developed, followed by pyrexia. Laboratory investigations revealed an ANC of 29/μL. Initiation of piperacillin/tazobactam and valaciclovir increased the ANC to 2200/μL by day 6.

**Case 2**

A 21-year-old female receiving maintenance therapy for systemic lupus erythematosus and antiphospholipid syndrome with rituximab, prednisolone (4 mg/day), mycophenolate mofetil, and tacrolimus developed stomatitis and gingival swelling five weeks after the final rituximab dose. Subsequent symptoms included pyrexia, odynophagia, and a right lingual tonsillar abscess. The ANC was 0/μL. Administration of piperacillin/tazobactam, valaciclovir, and granulocyte colony-stimulating factor led to ANC recovery by day 9. Both patients tested negative for IgM antibodies to herpes simplex virus, with no increase in paired IgG.

**Discussion**

The pathogenesis of late-onset neutropenia associated with BCDT involves complex interactions among immunological homeostasis and hematopoietic cell communication. Proposed mechanisms include excessive secretion of B-cell-activating factor during B-cell reconstitution, leading to lineage competition and arrest of neutrophil maturation (1); T-cell large granular lymphocyte expansion inducing neutrophil apoptosis(2); and trogocytosis-mediated transfer of CD20 antigens to neutrophils, increasing their susceptibility to antibody-dependent cellular cytotoxicity (3).

**Conclusion**

Late-onset neutropenia should be considered in patients receiving BCDT. These cases suggest that minor oral symptoms, such as gingival swelling, may serve as early indicators of late-onset neutropenia in this population. Clinical caution is essential for the prompt diagnosis and management of life-threatening agranulocytosis in BCDT.

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**Disclosure of interest**
None declared.

## P308

### Effectiveness of anakinra in Kawasaki Disease Shock Syndrome in a 6-year-old boy

M. Tsinti[1,*], K. Zarra[1], E. Tsitsami[1]
[1]Pediatric Rheumatology Unit First Department of Pediatrics, National and Kapodistrian University of Athens Medical School,'Children's Hospital, Greece, Athens, Greece
**Correspondence:** M. Tsinti
*Pediatric Rheumatology 2026*, **24(S1):**P308

**Introduction**
Kawasaki Disease Shock Syndrome (KDSS) is a rare severe form of KD in which patients present with vasodilatory shock, hypotension with poor perfusion, and possibly myocardial dysfunction. Evidence suggests the effectiveness of anakinra in resolution of both hyperinflammation and Coronary Artery Lesions.
**Objectives**
to highlight the effectiveness of SC anakinra and pulse steroids in the treatment of IVIG-refractory KDSS.
**Methods**
Clinical case presentation.
**References: Results**
A 6.5-year-old boy was admitted with fever up to 39.7 °C for 4 days, abdominal and testicular pain, maculopapular rash, conjunctivitis, cervical lymphadenitis, cheilitis, and edema of the extremities. Markers of inflammation were elevated (CRP 150 mg/L, PCT= 3 µg/l). Broad spectrum antibiotics were initiated. Initial cardiac evaluation revealed ectasia of the left main coronary artery (z LMCA-score 2.15), without cardiac dysfunction. IVIG 2 g/kg and aspirin 70 mg/kg/24 were administered. On the 3rd day of hospitalization clinical picture deteriorated with fever worsening and paralytic ileus hypotension and myocardial dysfunction (reduction of the ejection fraction, new onset mild metroid and tricuspid regurgitation. CRP, PCT and NTpro-BNP increased (280 mg/L, 12.5 µg/l and 24780 pg/ml). Hypoalbuminemia 2.5 g/dl was detected. The patient was transferred to PICU for 48 h for support with inotropes. Three pulses methylprednisolone 30 mg/kg and sc anakinra 5 mg/kg/24 h were administered. Aspirin was continued 5 mg/kg/24 h. Resolution of hyperinflammation and restoration of cardiovascular disfunction were achieved 24 h after treatment intensification. After discharge from the PICU, he developed pancreatitis successfully treated with low-fat diet. Mild dilation on the root of LMCA is still detectable in the 6-month-follow-up. Steroids were discontinued after 6 weeks and anakinra after 6 months.
**Trial registration identifying number: Conclusions**
In this 6-year-old boy with KDSS and clinical and laboratory features of severe hyperinflammation and cardiac dysfunction that evolved despite IVIG and aspirin treatment, anakinra along with pulse methylprednisolone led to rapid resolution of hyperinflammation and resolution of cardiac abnormalities. Prompt initiation of immunomodulatory therapy may lead to favorable outcome and recovery of cardiac injury.
**Disclosure of interest**
None declared.

## P309

### Outpatient versus inpatient therapy approaches in the nursing care of children with rheumatic diseases

J. Mattei[1,*], K. Mördrup[2,3]
[1]Kinderklinik Garmisch-Partenkirchen gGmbH, Garmisch-Partenkirchen, Germany; [2]The institution for Women's and Children's Health, Karolinska Institutet, Sweden, Solna; [3]Karolinska University Hospital, Astrid Lindgren Children's Hospital, Stockholm, Sweden
**Correspondence:** J. Mattei
*Pediatric Rheumatology 2026*, **24(S1):**P309

**Introduction**
In many countries, the nurse acts as the central point of contact within their area of responsibility and provides outpatient care. In countries where patients are treated as inpatients, care is generally provided in the hospital during acute flare-ups or for specific interventions such as infusion therapies.
**Objectives**
We compared the different approaches to find out the advantages and disadvantages.
**Methods**
As part of the collaboration for a PReS Educational Course, we analyzed different nursing therapy approaches in Sweden and Germany. These were then compared and our experiences as experts in the respective setting were included.
**Results**
(1) Inpatient therapy approach in Germany. The total prevalence of children affected in Germany lies between 15,000 [i] and 20,000 [ii]. Each year, just under 8,000 cases receive inpatient treatment [iii]. This therefore accounts for a significant proportion. For the comparison, the concept of a specialized hospital was discussed. A special concept was developed in which approaches from different professional groups can be ideally combined. [iv] The nursing specialist is the permanent contact person on the ward, coordinates appointments and supports the patient during their stay according to their individual needs. The advantage for the affected families is that the organizational and time effort is lower than with individual outpatient appointments with specialized therapists at various locations. (2) Outpatient therapeutic approach in Sweden. In sweden most patients are treated as out patients. The goal is to minimize the impact of the illness on the patients and their entire family. In most cases, families are assigned a nursing specialist as a point of contact following the diagnosis. When possible, appointments with different health care professionals are coordinated. The respective goal to enable those affected to lead their daily lives largely unimpeded is the same. Ultimately, what is crucial for the success of therapy is that it is carried out multi-professionally, individually and holistically.
**Conclusion**
Regardless of the setting, therapy and the nurse's role encompass counseling, guidance, concrete support, and prevention. The aim is to enable patients and their relatives to lead an independent, normal daily life despite the chronic illness. The structure and organization of a country's entire healthcare system are decisive in determining how this goal can ideally be achieved.

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**Disclosure of interest**
J. Mattei Employee with: Low conflict value, K. Mördrup Employee with: Low conflict value.

## P310

### Comparison of systemic corticosteroid therapy practices between pediatric and adult rheumatologists: a multicenter survey study

Ö. Özenli Yağcı[1,*], Ç. Yıldız[1], D. Gezgin Yıldırım[1], E. Bilgin[2], S. A. Bakkaloğlu[1]
[1]Department of Pediatric Rheumatology, Gazi University Faculty of Medicine, Ankara; [2]Department of Rheumatology, Sakarya University, Sakarya, Türkiye
**Correspondence:** Ö. Özenli Yağcı
*Pediatric Rheumatology 2026*, **24(S1):**P310

**Introduction**
The indications, dosage and duration of systemic corticosteroid treatment may vary among pediatric rheumatologists (PRs) and adult rheumatologists (ARs) in daily clinical practice (1).

**Objectives**
This study aims to evaluate and compare practices, perceptions, and attitudes of PRs and ARs regarding the use of systemic corticosteroids.
**Methods**
An online survey was administered to rheumatologists in Türkiye to assess about their use of systemic corticosteroids in the management of rheumatic diseases. The questionnaire addressed preferred corticosteroid formulations, dosing and tapering strategies, monitoring and preventive measures for adverse effects, and vaccination practices.
**Results**
Sixty PRs and forty ARs participated in the study. Prednisolone was most commonly preferred by PRs while almost all ARs used methylprednisolone. Weight-based dosing was favored by PRs whereas ARs preferred fixed-dose regimens. PRs more frequently preferred longer pulse corticosteroid therapy, whereas ARs most commonly used 3-day pulse regimen. In contrast, ARs reported significantly longer durations of bridging and maintenance therapy. Delirium was reported significantly more often by ARs as a complication ($p$ =0.003). In pre-treatment evaluation, PRs more commonly assessed peripheral blood smear ($p$ <0.001) whereas ARs more frequently screened for hepatitis serology ($p$ =0.014). As preventive strategies, PRs were significantly more likely to request ophthalmologic examination ($p$ =0.005). Immunization assessment for meningococcal, measles-mumps-rubella and varicella vaccines was significantly more common among PRs ($p$ >0.005, for all).
**Conclusion**
Significant heterogeneity exists between PRs and ARs in systemic corticosteroids practices, highlighting the need for standardized protocols for dosing, preventive strategies, and complication monitoring.

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**Disclosure of interest**
None declared.

## P311

### Methotrexate-related gastrointestinal intolerance in juvenile idiopathic arthritis: clinical characteristics and therapeutic outcomes

H. H. Ozguner Kucuk [1,*], G. Kavrul Kayaalp[1], O. Koker[1]
[1]Division of Pediatric Rheumatology, Marmara University Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** H. H. Ozguner Kucuk
*Pediatric Rheumatology 2026*, **24(S1):**P311

**Introduction**
Juvenile idiopathic arthritis (JIA) is the most common chronic rheumatic disease of childhood. Methotrexate (MTX) remains the cornerstone first-line conventional disease-modifying antirheumatic drug; however, treatment-related adverse effects, particularly gastrointestinal intolerance, represent a major challenge affecting treatment adherence and therapeutic continuity.
**Objectives**
To evaluate the frequency and clinical characteristics of MTX-related intolerance and adverse effects in patients with JIA and to assess subsequent management strategies and clinical outcomes.
**Methods**
In this retrospective single-center study, the medical records of 144 patients diagnosed with JIA were reviewed. Demographic data, clinical characteristics, and laboratory findings were collected. JIA subtypes, MTX dose and route of administration, concomitant therapies, frequency and characteristics of intolerance symptoms, time to symptom onset, and management approaches following intolerance were evaluated.
**Results**
MTX-related gastrointestinal intolerance was identified in 34 of 144 patients (23.6%). Among affected patients, 52.9% were female, with a mean age of 14.4±3.38 years and a mean age at disease onset of 12.2±3.4 years. JIA subtypes included oligoarticular JIA (44.1%), enthesitis-related arthritis (32.4%), polyarticular JIA (20.6%), and psoriatic arthritis (2.9%). All patients were receiving subcutaneous MTX. The mean time to development of intolerance was 8.2±4.53 months. Nausea was the most reported symptom (73.5%), followed by combined nausea and vomiting (17.6%) and anticipatory nausea (8.8%). Following the development of intolerance, MTX was discontinued in 58.8% ($n$=20) of patients, treatment intervals were prolonged in 29.4% ($n$=10), and symptomatic improvement after folic acid supplementation was observed in 8.8% ($n$=3). Among patients discontinuing MTX, 75% ($n$=15) were switched to biologic therapy, 10% ($n$=2) continued concomitant biologic treatment, and 15% ($n$=3) remained off treatment because of sustained inactive disease. MTX-related elevation of liver enzymes was detected in 9 patients (6.3%). In eight patients, MTX was temporarily interrupted and successfully reintroduced following normalization of liver enzyme levels. In one patient, MTX was permanently discontinued because of sustained clinical remission.
**Conclusion**
MTX-related gastrointestinal intolerance is a frequent complication in patients with JIA and represents an important cause of treatment modification and escalation to biologic therapy. In contrast, MTX-associated hepatotoxicity is generally transient and manageable with close monitoring and timely intervention. These findings highlight the importance of early recognition and individualized management of MTX-related adverse effects to optimize treatment adherence and maintain long-term disease control.
**Disclosure of interest**
None declared.

## P312

### When treatment triggers trouble: a single centre retrospective review of Drug Hypersensitivity Reactions (DHRs) to biologic agents in a paediatric rheumatology population

P. Whooley[1,*], M. Kelleher[2], A. Stafford[2], O. Killeen[1], E. MacDermott[1], A. Byrne[2], K. Gallagher[1]
[1]Rheumatology; [2]Allergy, Childrens Health Ireland, Dublin, Ireland
**Correspondence:** P. Whooley
*Pediatric Rheumatology 2026*, **24(S1):**P312

**Introduction**
The increasing use of biologic agents in the management of rheumatological conditions has led to increasing reports of DHRs. Such reactions may cause morbidity and treatment discontinuation. Despite this, there remains limited data describing DHRs in this population.
**Objectives**
This study aims to establish a comprehensive database of biologic-associated DHRs of all patients treated with infliximab, rituximab and tocilizumab at the National Centre for Paediatric Rheumatology (NCPR), Ireland. Data gathered will be used to inform and develop guidelines for biologic-related DHRs.
**Methods**
A retrospective chart review conducted at the NCPR included all patients who received infliximab, rituximab and tocilizumab between January 2020 and August 2025. Ethical approval was obtained. Reactions were classified using Common Terminology Criteria for Adverse Events.
**Results**
91 patient charts were reviewed. 45 patients received infliximab; 12 DHRs were identified among eight patients (female $n$=5; 62.5%, male=3; 37.5%). 23 patients received rituximab; 11 DHRs were identified among 10 patients (female $n$=7; 70%, male=3; 30%). No DHRs to tocilizumab were identified among 23 patients. Reactions to infliximab were classified as mild (6/12, 50%), moderate (4/12, 33.3%), severe (1/12, 8.3%), and life-threatening (1/12, 8.3%). Reactions to rituximab classified as mild ($n$=2,18.2%), moderate ($n$=8,72.7%), and severe ($n$=1,9.1%). 11/12 (91.7%) DHRs to infliximab occurred after multiple exposures (mean infusion number 14.9). In contrast, the majority of reactions to rituximab occurred during first exposure ($n$=8, 72.7%). Concomitant immunosuppressive agents were being taken in 10/12 (83.3%) infliximab reactors and in 9/11 (81.8%) rituximab reactors. The Allergy team were consulted in 8/12 (66.7%) of infliximab reactions but only 3/11 of rituximab reactions (27.3%). Infliximab infusions were re-commenced on the same day in 5/12 DHRs (one with rate adjustment) and on a later date in 1/12 DHR (without rate adjustment). Infliximab was permanently discontinued following 5/12 DHRs. Rituximab was recommenced on the same day in 5/11 reactions (4/11 with rate adjustment), recommenced on a different day without rate adjustment in 1/11 (9.1%), and subsequently

administered via desensitisation in 1/11 (9.1%). Rituximab was permanently discontinued following 1/11 (9.1%) DHR. The remaining 3/11 (27%) continued rituximab treatment without issue.

**Conclusion**

This is the first analysis of biologic-related DHRs conducted at the NCPR. DHRs to biologics are common in this cohort and cause significant morbidity. Infliximab was much more likely to be discontinued post-DHR compared to rituximab. Consistent engagement with the Allergy team is crucial to achieve optimal patient outcomes.

**Disclosure of interest**

None declared.

## P313

### Early methotrexate monitoring in juvenile idiopathic arthritis: a review of toxicity and adherence to guidelines

P. Pankhania[*,1], J. M. MacMahon[2]
[1]School of Clinical Medicine, University of Cambridge; [2]Department of Paediatric Rheumatology, Cambridge University Hospitals NHS Foundation Trust, Cambridge, UK
**Correspondence:** P. Pankhania
*Pediatric Rheumatology 2026*, **24(S1):**P313

**Introduction**

Methotrexate (MTX) is a first-line conventional synthetic DMARD in the treatment of Juvenile Idiopathic Arthritis (JIA). While the recommended rate of early monitoring for toxicity has reduced, the paediatric evidence base on MTX safety and monitoring in the initiation period is limited.

**Objectives**

To evaluate current evidence on MTX toxicity, appropriate blood monitoring frequencies, associated risk factors, and adherence to monitoring guidelines in the first six months of JIA treatment.

**Methods**

Literature search conducted using MEDLINE, Embase, PubMed and Cochrane. Studies reporting toxicity outcomes, monitoring frequency, risk factors, or adherence to monitoring guidelines during the first six months of MTX treatment for JIA were included. All study designs were considered. Full-texts and abstracts were included. Studies that were non-English, published before 2000, adult population only, or not specifying the nature of adverse reactions were excluded. Reference lists of included studies were searched.

**Results**

319 studies were extracted. Following removal of duplicates and screening, a total of 22 publications met eligibility criteria. A further 3 were identified through reference screening. Abnormalities in transaminases were frequently reported, although were often transient and associated with low rates of permanent MTX discontinuation. High temporary discontinuation rates suggest possible over-intervention. Interpretation is limited by significant heterogeneity, including definitions of toxicity, monitoring frequency, and length of follow-up. While laboratory abnormalities were reported to peak specifically in the first month and six months of treatment, few studies reported the timing of abnormalities, and none compared monitoring frequencies. Few risk factors have been identified. Limited evidence suggests a role for polymorphisms to MTX pathway genes, starting dose, concomitant therapy and concurrent infection as potential risk factors for toxicity. However, findings are inconsistent, and roles in persistent, significant abnormalities are unclear. Adherence to monitoring guidelines is low, with frequent over- and under-monitoring. No paediatric studies evaluated adherence specifically in the first six months of treatment.

**Conclusion**

Clinically significant MTX toxicity in the first six months of JIA treatment is rare. Clinical practice is hindered by limited evidence on appropriate monitoring frequencies, unvalidated risk factors, and low guideline adherence. Given the peak in laboratory abnormalities during treatment initiation, studies evaluating adherence to current monitoring guidelines are needed to assess this possible safety gap and prevent interruption to care.

**Disclosure of interest**

None declared.

## P314

### A unique clinical case of major gynaecological surgery because of severe congenital malformation in fibrodysplasia ossificans progressiva: management under palovarotene and JAK inhibition

I. Nikishina[1], S. Arsenyeva[1,*], V. Matkava[1], S. Valieva[2], S. Kurbanova[2], S. Kravchuk[3], D. Gorokhov[4], S. Korotkova[5]
[1]Paediatric, V.A. Nasonova Scientific Research Institute of Rheumatology; [2]Paediatric; [3]Anesthesiology; [4]Intensive care; [5]Gynecological, Morozov City Children's Clinical Hospital, Moscow, Russian Federation
**Correspondence:** S. Arsenyeva
*Pediatric Rheumatology 2026*, **24(S1):**P314

**Introduction**

Fibrodysplasia ossificans progressiva (FOP) is an ultra-rare genetic disorder in which trauma, any invasive procedures and surgery may trigger inflammatory flare-ups followed by heterotopic ossification. Therefore, surgical treatment is usually avoided whenever possible. However, severe non-rheumatological conditions may create situations in which surgery becomes inevitably.

**Objectives**

To describe a rare case of major gynaecological surgical operation required due to a life-threatening congenital abnormality of internal genital development in adolescent girl with severe FOP receiving palovarotene and JAK inhibitor therapy.

**Methods**

A 16-year-old girl with genetically confirmed FOP had a severe, rapidly progressive course of disease and typical clinical features, multiple heterotopic ossifications, functional restriction and joint contractures. Large joints synovitis, sacroiliitis, confirmed by CT and MRI and cervical spine ankylosis were observed as rheumatological manifestations She was currently treated with palovarotene and upadacitinib (tofacitinib previously). She had primary amenorrhoea and recurrent lower abdominal pain since the age of 15 years.

**Results**

Pelvic imaging revealed a complex malformation of the internal genital organs with impaired menstrual outflow: incomplete uterine septum/bicornuate uterus, cervical canal obstruction/atresia, hematometra, hematocervix and bilateral hematosalpinx. Because of huge difficulties with anesthesia due to cervical spine ankylosis and high risk of flare-ups development after invasive procedures, initial conservative hormonal treatment was prescribed (Dienogest). However, symptoms and obstructive changes increased. Surgical treatment was considered unavoidable. Due to progressive blood accumulation and persistent pain, cervical canal bougienage and laparoscopic puncture with evacuation of hematosalpinx, hematocervix and hematometra were initially attempted. Due to severe malformation of the internal genital organs and total adenomyosis multidisciplinary laparoscopic hysterectomy, bilateral salpingectomy, bilateral ovarian biopsy and ovarian tissue resection for cryopreservation were performed. Despite the expected anaesthetic risks in FOP, orotracheal intubation with videolaryngoscopic control was performed without complications. The postoperative period was clinically stable. No respiratory, haemorrhagic complications or early clinically evident of FOP flare-up were observed as a result of multidisciplinary team's work. Before these surgical procedures premedication with intravenous dexamethasone with gradual tapering was performed twice (during 5 days). Palovarotene and upadacitinib were not withdrawal during all period. Follow up 8 months revealed no flare-up including area of anterior abdominal wall muscles.

**Conclusion**

This case illustrates that even major gynaecological surgery may be possible in some patients with FOP in inevitably cases and the procedure is performed under premedication. The absence of flare-up in a patient treated with palovarotene and JAK inhibition is important and may support further discussion of perioperative strategies in FOP. Longer follow-up is required to assess delayed heterotopic ossification after surgery.

**Disclosure of interest**

None declared.

## P315

### Treatment with Janus kinase (JAK) inhibitors in pediatric rheumatology

S. Barreiro Pérez[1], C. Udaondo[1,*], A. Montserrat[2] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, M. A. Muñoz[3] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, Á. Villarejo[4] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, L. Martín Pedraz[5] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, J. Sánchez[6] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, D. Clemente[7] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, L. Fernández[8] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, S. Guillén[9] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, A. Remesal[1], S. Murias[10] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, R. Alcobendas[1], B. Bravo[11] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology, C. Calvo[12] on behalf of Working Group on JAK Inhibitor Therapy of the Spanish Society of Pediatric Rheumatology
[1]Pediatric Rheumatology, Hospital Universitario La Paz; [2]Pediatric Rheumatology, Hospital Universitario Sant Joan de Deu, Madrid; [3]Pediatric Rheumatology, Hospital Universitario Son Espases, Palma de Mallorca; [4]Pediatric Rheumatology, Hospital Universitario 12 de Octubre, Madrid; [5]Pediatric Rheumatology, Hospital Regional Universitario de Málaga, Málaga; [6]Pediatric Rheumatology, Parc Taulí Hospital Universitari, Sabadell; [7]Pediatric Rheumatology, Hospital Universitario Niño Jesús, Madrid; [8]Pediatric Rheumatology, Hospital Universitario Virgen del Rocio, Sevilla; [9]Pediatric Rheumatology, Hospital Universitario de Getafe, Getafe; [10]Pediatric Rheumatology, Hospital Universitario Central de Asturias, Oviedo; [11]Pediatric Rheumatology, Hospital Universitario Virgen de las Nieves Granada, Granada; [12]Pediatrics and Infectious Diseases, Hospital Universitario La Paz, Madrid, Spain
**Correspondence:** C. Udaondo
*Pediatric Rheumatology 2026*, **24(S1):**P315

**Introduction**
Janus kinase inhibitors (JAKi) are immunomodulatory drugs recently approved for several inflammatory diseases in adults and, to a more limited extent, in pediatric patients, where their use is mostly off-label.
**Objectives**
The aim of this study was to describe the indications and adverse events associated with JAKi use in pediatric rheumatology.
**Methods**
This was a multicenter, retrospective, observational study conducted across eleven Spanish pediatric rheumatology departments. Patients aged 0–17 years who received treatment with JAKi between January 1, 2018, and December 31, 2023, were included.
**Results**
A total of 50 patients were included, of whom 19 (38%) were male. The most frequent indication for treatment was juvenile idiopathic arthritis (JIA) (32 patients, 64%). Nine patients (18%) received treatment with two different JAK inhibitors. The median age at initiation of the first JAKi was 11.42 years (IQR 7.96–13.77). Tofacitinib was the most commonly used JAKi (24 cases, 48%). Overall, 34 patients (57.6%) remained on treatment with a JAKi, including 28 on their first JAKi and 6 on a second agent. Among patients who discontinued treatment, the median treatment duration was 14 months (IQR 7.0–22.0). Reasons for discontinuation included therapeutic failure in 76% of cases, adverse events in one patient, and disease resolution in another. Concomitant medications were required in 41 patients (82%), with oral corticosteroids being the most frequently used (17 patients, 34%). Regarding safety, 37 patients (74%) remained free of adverse events. A total of 23 adverse events were reported, including 18 infections (30.5%) and 4 hematological abnormalities (6.8%). Most events were mild (19 cases, 32.2%), and no severe adverse events were reported.
**Conclusion**
JAK inhibitors represent an effective therapeutic option for pediatric patients with inflammatory diseases, particularly JIA. Most patients tolerated treatment well, with predominantly mild adverse events.
**Disclosure of interest**
None declared.

## P316

### Patterns of switching the first biologic therapy in juvenile idiopathic arthritis: a single-center real-world study

D. Sat[1,*], M. Assylbekova[1], Z. Mukusheva[1], Z. Albekova[1], A. Dildabayeva[1], A. Akhmambetova[1]
[1]Rheumatology Program, Clinical Academic Department of Pediatrics, "University Medical Center" Corporate Fund, Astana, Kazakhstan
**Correspondence:** D. Sat
*Pediatric Rheumatology 2026*, **24(S1):**P316

**Introduction**
Biologic therapies have transformed the management of juvenile idiopathic arthritis (JIA), although a proportion of patients require treatment switching due to inadequate response or adverse events. Repeated biologic failure may reflect a difficult-to-treat disease phenotype characterized by persistent inflammation and reduced therapeutic response. Real-world evidence on switching patterns in pediatric JIA remains limited in Central Asian populations.
**Objectives**
To evaluate reasons, patterns, and outcomes of switching the first biologic therapy in children with JIA in a tertiary pediatric rheumatology center.
**Methods**
We conducted a retrospective observational study of JIA patients who switched their first biologic therapy between 2018 and 2025. Clinical and demographic data, JIA subtype, first biologic agent, duration before switching, reasons for discontinuation, subsequent therapy, and outcomes were extracted from medical records. Descriptive statistics were applied.
**Results**
A total of 20 patients were included. The cohort was predominantly female (70%), with a mean age at disease onset of 7.8 years (10 months–17 years). Subtypes included oligoarticular JIA (40%), RF-positive polyarticular (25%), RF-negative polyarticular (15%), and enthesitis-related arthritis (20%). Etanercept was the most commonly used first biologic (60%), followed by adalimumab (40%) and tocilizumab (20%). Mean duration of first biologic therapy was 1 year 8 months (6 months–5 years 3 months), with longer drug survival in oligoarticular JIA and earlier switching in polyarticular JIA and ERA. Secondary inefficacy was the leading reason for switching ($n$=16), followed by primary inefficacy ($n$=3), uveitis ($n$=3), poor adherence ($n$=2), and persistent leukocytosis ($n$=1). After switching, patients received adalimumab ($n$=6), secukinumab ($n$=6), infliximab ($n$=3), or tocilizumab ($n$=6). IL-17 inhibition was more frequently used in ERA, while repeated TNF inhibitor failure was mainly observed in polyarticular JIA. Clinical improvement after switching was observed in 65% of patients, while 35% had persistent or partially controlled disease activity. Two patients demonstrated a difficult-to-treat course with sequential exposure to three biologics.
**Conclusion**
Biologic switching was common in JIA, most frequently due to loss of efficacy. Most patients achieved clinical improvement after switching. Repeated biologic failure was mainly observed in polyarticular JIA and ERA, suggesting distinct refractory phenotypes requiring individualized therapeutic strategies.

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**Disclosure of interest**
None declared.

## P317

### Predictors of relapse after bDMARD withdrawal in juvenile idiopathic arthritis: a multicenter study

S. Yücel[1,*], N. Gürbüz[1], T. Çeltik[2], B. E. Kösalı[3], G. Kaymak[1], E. Gülyiğit[1], T. Çetin Arıkan[1], A. H. Kamiloğlu[1], G. Avcı Demirciler[1], Ö. Altuğ Gücenmez[3], B. Bora[2], S. E. Ünsal[2], A. Berdeli[1,4], N. Kütükçüler[1,5], G. Aksu[1,5]
[1]Department of Pediatric Rheumatology, Ege University Faculty of Medicine; [2]Department of Pediatric Rheumatology, Dokuz Eylul University Faculty of Medicine; [3]Department of Pediatric Rheumatology, University of Health Sciences, Dr. Behçet Uz Children's Hospital; [4]Molecular Genetics Laboratory, Department of Pediatrics; [5]Department of Pediatric Immunology, Ege University Faculty of Medicine, Izmir, Türkiye
**Correspondence:** S. Yücel
*Pediatric Rheumatology 2026*, **24(S1):**P317

**Introduction**
Disease relapse after biologic withdrawal is still a major challenge in juvenile idiopathic arthritis (JIA). The predictors and optimal timing of withdrawal remain unclear.
**Objectives**
We aimed to evaluate factors associated with relapse after withdrawal of biologic disease-modifying anti-rheumatic agents (bDMARDs) in patients with JIA.
**Methods**
Patients diagnosed with JIA from 2010 to 2025 according to International League of Associations for Rheumatology (ILAR) classification criteria who received bDMARDS, discontinued treatment after achieving remission, and followed for at least one year after withdrawal were retrospectively evaluated in this multicenter study. Fisher's exact test, Mann-Whitney U test, and chi-square tests were used to compare data. Univariate regression was used to analyze risk factors of relapse. Cox regression and Kaplan-Meier curves were constructed to assess predictors of outcome.
**Results**
A total of 71 patients whose bDMARDS were withdrawn after remission were evaluated; 5 of them were diagnosed with Still's disease. Relapse after bDMARD withdrawal was observed in 49 (69%) of them, 28 (39,4%) being within the first year of cessation. Still's disease group was excluded from the sensitivity analysis. No categorical variables were associated with relapse. Longer biologic treatment duration ($p$=0.015), inactive disease duration before withdrawal ($p$=0.044), and clinical remission duration before withdrawal ($p$=0.034) were associated with sustained remission. No significant differences in relapse-free survival were observed according to ANA positivity, history of uveitis, or withdrawal strategy. Kaplan–Meier and Cox regression analyses did not demonstrate independent predictors of relapse.
**Conclusion**
Longer biologic treatment duration and prolonged inactive disease/remission duration before withdrawal were associated with sustained remission after bDMARD discontinuation in non-systemic JIA patients.

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**Disclosure of interest**
None declared.

## P318

### Impact of methotrexate withdrawal regimen on relapse risk in children with juvenile idiopathic arthritis: analysis of the Moscow registry

N. Babich[1], V. Sevostyanov[2,*], S. Valieva[1], S. Kurbanova[1], O. Lomakina[1], E. Zholobova[1,3]
[1]Moscow City Center of Pediatric Rheumatology, Morozov Children's City Clinical Hospital; [2]Department of Propedeutics of Children's Diseases; [3]Department of Children's Diseases, Federal State Autonomous Educational Institution of Higher Education I.M. Sechenov First Moscow State Medical University of the Ministry of Health of the Russian Federation (Sechenov University), Moscow, Russian Federation
**Correspondence:** V. Sevostyanov
*Pediatric Rheumatology 2026*, **24(S1):**P318

**Introduction**
Continuing MTX for 12 months (vs. 6 months) after remission significantly reduces relapse risk [1]. However, realworld MTX withdrawal (abrupt or gradual) and relapse predictors are understudied; one recent study found no association of withdrawal regimen with relapse, but NSAIDs doubled the hazard [2]. We therefore evaluated JIA relapse frequency after MTX withdrawal according to regimen and clinical factors using a large retrospective registry.
**Objectives**
To evaluate the frequency of JIA relapses after MTX withdrawal according to withdrawal regimen and additional clinical factors.
**Methods**
A retrospective cohort study based on the Moscow Registry of Children with Rheumatic Diseases (data locked on 01.09.2025). We included patients with JIA (ILAR criteria) who received MTX and had a documented MTX withdrawal after ≥6 months of inactive disease status. Withdrawal regimens were classified as abrupt (discontinuation within ≤2 weeks) or gradual (increasing the interval between doses and/or reducing the dose). Relapse was defined as recurrence of active arthritis or uveitis requiring therapy intensification. Statistical analysis: descriptive statistics (median, interquartile range); frequency comparisons – $\chi^2$ test (or Fisher's exact test). Odds ratio (OR) with 95% confidence interval (CI), absolute risk difference (ARD), and number needed to harm (NNH) were calculated. Timetorelapse was analysed using Kaplan–Meier curves and the logrank test. Statistical significance was set at $p<0.05$.
**Results**
Among 1,297 JIA patients, 786 (71.3%) received MTX. MTX withdrawal was documented in 137 patients (10.6% of all JIA). Median age at withdrawal – 12 [9; 14] years. Withdrawal regimen was known for 90 patients (65.7%): abrupt – 74 (82.2%), gradual – 16 (17.8%). The groups did not differ in age, sex, or JIA subtype ($p>0.05$ for all comparisons).
Relapse rates:
Overall relapse rate – 20.4% (28/137).
Abrupt withdrawal – 29.7% (22/74).
Gradual withdrawal – 12.5% (2/16).
Relapse risk was higher after abrupt withdrawal: OR = 2.96 (95% CI: 0.62–14.1, $p$=0.245). Absolute risk difference = 17.2%, NNH = 6 (i.e., one additional relapse occurs for every 6 patients withdrawn abruptly). Median time to relapse: abrupt withdrawal – 11.5 months [7; 16]; gradual withdrawal – 16 months [13; 19]. Logrank $p$=0.12. Relapse rates: polyarticular RFnegative – 22.2% (8/36), oligoarticular – 20.5% (18/88), systemic – 12.5% (1/8). Differences between subtypes were not statistically significant ($p$=0.78).
**Conclusion**
In realworld practice, MTX withdrawal is most often abrupt. Abrupt discontinuation shows a >2.9fold higher relapse risk (not significant, possibly due to small gradualwithdrawal sample). One additional relapse per 6 abrupt withdrawals. JIA subtype does not affect risk. Prospective studies are needed.

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**Disclosure of interest**
None declared.

## P319

### Clinical characteristics and outcomes of pediatric patients with rheumatic diseases treated with anakinra: a single-center experience from Kazakhstan

A. Otegen[1], Z. Mukusheva[*,1]
[1]Pediatric rheumatology, CF "University medical center", Astana, Kazakhstan
**Correspondence:** Z. Mukusheva
*Pediatric Rheumatology 2026*, **24(S1):**P319

**Introduction**
Pediatric rheumatic diseases are associated with significant morbidity and life-threatening complications, including macrophage activation syndrome (MAS). Dysregulation of the interleukin-1 (IL-1) pathway plays a central role in systemic inflammation. Anakinra is an important therapeutic approach, yet real-world data remain limited.
**Objectives**
To evaluate the clinical characteristics, indications, treatment response and outcomes of pediatric patients with rheumatic diseases treated with Anakinra in a tertiary referral center.
**Methods**
We retrospectively analyzed 18 pediatric patients with rheumatic diseases treated with Anakinra between 2021 and 2025. Median age was 6 years (0.9–16); 44.4% were male (*n*=8) and 55.6% female (*n*=10). Clinical features, laboratory findings, indications, treatment duration, response, and adverse events were evaluated.
**Results**
Most patients had systemic juvenile idiopathic arthritis (77.8%, *n*=14), followed by Kawasaki disease (11.1%, *n*=2), systemic lupus erythematosus (5.6%, *n*=1), and juvenile dermatomyositis (5.6%, *n*=1). Macrophage activation syndrome (MAS) occurred in 50.0% (*n*=9). In sJIA patients, fever resolved within 72 h after anakinra initiation, with rapid improvement in systemic symptoms and inflammatory markers. Median CRP decreased from 79.75 to 1.93. In MAS patients, median HScore fell from 190 to 0, indicating rapid control of hyperinflammation. Complete remission was achieved in 11.1% (*n*=2), while others had partial remission or persistent activity. Anakinra enabled glucocorticoid reduction/discontinuation in many cases. Dose escalation was required in 11.1% (*n*=2) due to flare or insufficient response, and 16.7% (*n*=3) with refractory sJIA had persistent activity after steroid tapering. Subcutaneous anakinra was administered at 2–5 mg/kg/day; two patients required intravenous administration (2 and 10 mg/kg/day). Adverse events included one mild injection-site reaction and one case of hepatotoxicity requiring treatment discontinuation. No serious infections or treatment-related mortality occurred.
**Conclusion**
Anakinra demonstrated high effectiveness and a favorable safety profile in children with severe rheumatic diseases across different diagnoses, including life-threatening MAS. These real-world data support early IL-1 blockade as a key therapeutic strategy for rapid disease control and improved clinical outcomes in complex pediatric patients.
**Disclosure of interest**
None declared.

## P320

### Sulfasalazine-induced DRESS syndrome in a child with recurrent flares during corticosteroid tapering

V. A. Podzolkova[1,2,*], S. A. Davtian[1], A. B. Kazumova[3], O. Y. Loskutova[2]
[1]Children's Diseases, Sechenov First Moscow State Medical University; [2]Fantasy Children's Clinic; [3]Sechenov First Moscow State Medical University, Moscow, Russian Federation
**Correspondence:** V. A. Podzolkova
*Pediatric Rheumatology 2026*, **24(S1):**P320

**Introduction**
Drug Reaction with Eosinophilia and Systemic Symptoms (DRESS) is a severe hypersensitivity reaction that can be triggered by sulfasalazine, a drug commonly used in paediatric rheumatology. Rheumatologists should be aware of this syndrome to ensure prompt diagnosis, avoid drug re-administration, and recognise potential relapses during glucocorticoid tapering.
**Objectives**
To present a clinical case of DRESS syndrome in a child with recurrent synovitis, with a focus on relapses triggered by glucocorticoid withdrawal.
**Methods**
A 4yearold boy with a 6month history of recurrent, asymmetrical synovitis of the knees, ankles and left hip. Laboratory tests showed elevated ESR (45 mm/h) and CRP (32 mg/L), but negative ANA, RF, HLAB27. Sulfasalazine was started at 15 mg/kg/day, increased to 30 mg/kg/day. Joint symptoms improved markedly within 10 days. Three weeks after starting sulfasalazine, the boy developed high fever (39.0 °C), facial oedema, and a pruritic rash on the auricular, cheek and cervical areas. The parents discontinued the drug. Initial evaluation in a community hospital suggested an acute respiratory infection. After reexposure to sulfasalazine (15 mg/kg/day), fever recurred within 4 h (38.7 °C), accompanied by worsening facial erythema and pruritus. Despite cetirizine, the condition progressed: fever up to 39.8 °C, generalised rash, severe pruritus. Intramuscular dexamethasone (8 mg) provided transient stabilisation, but the rash continued to spread. The child became irritable and developed an episode of confusion with disorientation and incoherent speech, which was interpreted as a clinical marker of central nervous system involvement. Laboratory tests revealed leukocytosis ($22.17\times10^9$/L) and absolute eosinophilia ($1.33\times10^9$/L). PCR from tonsillar material was positive for human herpesvirus 6. Intravenous dexamethasone (4 mg/day for 3 days) was given during hospitalisation. After discharge, fever and rash recurred, confirming the diagnosis of DRESS. Using the RegiSCAR criteria, the patient scored 6 points (fever >38.5 °C, lymphadenopathy, eosinophilia, typical rash, CNS involvement, lasting >15 days, exclusion of other causes), which corresponds to a definite DRESS diagnosis. Prednisolone was started at 0.9 mg/kg/day and tapered over 3 weeks. When the dose was reduced below 0.15 mg/kg/day, a relapse of cervical rash and pruritus occurred. Symptom control was achieved with cetirizine, and maintenance prednisolone (0.15 mg/kg/day) was continued for another 2 weeks, after which it was tapered over 4 weeks without further relapse. One month after complete withdrawal, the child remained symptomfree, but recurrent transient synovitis of the hip persisted, requiring NSAIDs. This case underlines the importance of prompt DRESS recognition, the risk of reexposure, and the need for prolonged glucocorticoid therapy with very slow tapering to avoid relapse. A multidisciplinary approach (paediatric rheumatologist, allergist, clinical pharmacologist) and precise differential diagnosis are essential.

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**Disclosure of interest**
None declared.

## P321

### Single-center real-world experience of multitargeting therapy with tofacitinib and palovarotene combination for patients with fibrodysplasia ossificans progressive

V. Matkava[1,*], I. Nikishina[1], S. Arsenyeva[1]
[1]Paediatric, V.A. Nasonova Scientific Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** V. Matkava
*Pediatric Rheumatology 2026*, **24(S1):**P321

**Introduction**
Fibrodysplasia ossificans progressiva (FOP) is an ultra-rare monogenic disease characterized by episodic flare-ups and progressive heterotopic

ossification (HO) and inflammatory musculoskeletal signs. Palovarotene (PVT), a selective retinoic acid receptor gamma (RARγ) agonist, is the first approved therapy shown to reduce new HO formation in clinical trials. Tofacitinib by targeting the JAK/STAT pathway may modulate the early inflammatory flare phase that precedes endochondral ossification. We have previously reported our experience with tofacitinib in FOP patients (pts) and in recent years it has become a routine therapeutic option at our centre, replacing glucocorticoids for flare-up management (1).

**Objectives**

To evaluate the first Russian real-world experience of multitarget therapy with palovarotene and tofacitinib in patients with FOP.

**Methods**

This was a prospective real-world cohort study of 17 patients with confirmed FOP caused by pathogenic *ACVR1* variants. All patients received combined treatment with tofacitinib and palovarotene. HO progression was assessed by low-dose whole-body computed tomography. Functional status was evaluated using the Cumulative Analogue Joint Involvement Scale (CAJIS) and Childhood Health Assessment Questionnaire (CHAQ). Adverse events were recorded throughout follow-up.

**Results**

The cohort included 17 patients. All pts had a classic FOP phenotype; 70% female; 16/17 had the classic *ACVR1* c.617G> A (p.Arg206His) variant and 1 patient had an ultra-rare c.1067G> A (p.Gly356Asp) variant. In most pts (16/17) tofacitinib therapy preceded palovarotene initiation; in one girl tofacitinib was added 4 months after starting palovarotene because of an arthritis. Median age at tofacitinib initiation was 11.0 years [8.6–12.3]. Median age at palovarotene initiation was 12.4 years. Median treatment duration was 42 months [31.0–62.5] for tofacitinib and 20 months [13.5–29.3] for palovarotene. In 8 patients receiving combined therapy for more than 12 months, paired assessment showed stable total HO volume over 12 months: 279.5 $cm^3$ [46.9–710.0] at palovarotene initiation and 280.5 $cm^3$ [46.9–710.0] at follow-up. CAJIS and CHAQ scores also remained stable. Mean height increased from 155.4±11.2 cm to 158.5±10.6 cm, with mean annual growth of 3.0±2.4 cm/year. No serious adverse events were observed. Palovarotene-associated mucocutaneous adverse events occurred in 14/17 patients, dyspeptic symptoms in 3/17, and transient transaminase elevation in 1/17.

**Conclusion**

This preliminary real-world experience suggests that combined palovarotene and tofacitinib therapy may stabilize HO progression and functional status in patients with FOP, with an acceptable safety profile. The complementary targeting of inflammation and endochondral ossification may represent a promising multitarget therapeutic strategy for FOP.

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**Disclosure of interest**

None declared.

## P325

### Beyond juvenile idiopathic arthritis: unmasking a rare genetic arthropathy after years of misdiagnosis

A. A. Abushhaiwia[1,*]
[1]Pediatric Rheumatology, Tripoli Children's Teaching Hospital, Faculty of Medicine, University of Tripoli, Tripoli, Libya
**Correspondence:** A. A. Abushhaiwia
*Pediatric Rheumatology 2026*, **24(S1):**P325

**Introduction**

Camptodactyly–Arthropathy–Coxa Vara–Pericarditis (CACP) syndrome (OMIM #208250; ORPHA: 137) is a rare autosomal recessive non-inflammatory arthropathy caused by mutations in the PRG4 gene encoding lubricin, a key glycoprotein essential for synovial lubrication and joint homeostasis. Clinically, it is characterized by progressive non-inflammatory arthropathy, camptodactyly, coxa vara, and occasionally pericardial involvement. Due to its chronic joint swelling and deformities, CACP syndrome is frequently misdiagnosed as juvenile idiopathic arthritis (JIA), resulting in prolonged and unnecessary exposure to immunosuppressive and biologic therapies without clinical benefit.

**Objectives**

To report a genetically confirmed case of CACP syndrome initially misdiagnosed as seronegative polyarticular JIA, emphasizing diagnostic challenges, therapeutic implications, and the importance of early genetic recognition.

**Methods**

A case report.

**Results**

We report a male patient born in 2008 who developed chronic joint disease at 4 years of age. Initial management included corticosteroids and methotrexate. The patient had a history of parental consanguinity and no family history of rheumatic disease. Despite treatment, he developed persistent symmetrical polyarthritis with progressive limitation of joint mobility. In 2017, he was evaluated in our pediatric rheumatology clinic, where involvement of multiple joints including wrists, elbows, knees, ankles, hips, and cervical spine was noted. Laboratory investigations repeatedly showed normal inflammatory markers (ESR and CRP), and autoimmune serology was consistently negative. Despite these atypical findings, a diagnosis of seronegative polyarticular JIA was established. Treatment was escalated to subcutaneous methotrexate, followed by biologic therapy with etanercept from 2018 to 2025. However, no sustained clinical remission was achieved.The disease course was characterized by persistent joint effusions, progressive flexion contractures, genu valgum, and marked functional impairment. Radiographic evaluation demonstrated hip dysplasia with progressive femoral varus deformity consistent with coxa vara, resulting in significant gait disturbance. Given the absence of inflammatory activity and poor therapeutic response, genetic testing was performed in 2026. This revealed a homozygous pathogenic PRG4 variant (c.32548_3260del, p.Ser1085*), confirming the diagnosis of CACP syndrome.

**Conclusion**

CACP syndrome should be considered in children labeled as "juvenile idiopathic arthritis" who fail to respond to prolonged immunosuppressive and biologic therapy, particularly when inflammatory markers remain normal and structural joint damage progressively worsens. Early differentiation from inflammatory arthropathies is essential to avoid unnecessary exposure to corticosteroids, DMARDs, and biologic agents, and to ensure timely genetic counseling and appropriate orthopedic management.

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**Disclosure of interest**

None declared.

## P327

### Physical inactivity as a risk factor for later developing JIA

E. Solheim[1,*], K. L. Haftorn[2], H. Sanner[2,3], K. Risum[1,2]
[1]Oslo Metropolitan University; [2]Department of Rheumatology, Oslo University Hospital; [3]University of Oslo, Oslo, Norway
**Correspondence:** E. Solheim
*Pediatric Rheumatology 2026*, **24(S1):**P327

**Introduction**

Juvenile idiopathic arthritis (JIA) is a chronic immune-mediated disease which affects children under the age of 16, and without certain etiology. JIA causes chronic inflammation in the joints, and it is assumed that both genetic and environmental factors can influence the risk. Physical activity may have anti-inflammatory effects, and other positive health effects. Screen time is often associated with sedentary time. Sedentary time is associated with less physical activity and more juvenile obesity. However, little is known about whether physical inactivity in early childhood influences the subsequent risk of developing JIA. To our knowledge, few population-based studies have examined early-life physical inactivity and screen time in relation to the later risk of JIA.

**Objectives**
This study examined whether physical activity or screen time affects the subsequent risk of JIA.
**Methods**
This was a population-based observational study using data from the Norwegian Mother, Father, Child Cohort study (MoBa), which recruited participants during the period 1999-2008. JIA was defined through a combination of chart review and ICD-10 codes (M08, M09), linked to the Norwegian Patient Registry. Physical activity was measured as time outdoor, and screen time was measured trough time spent in front of TV/video. Time outdoor and screen time was assessed through parent-registered questionnaires when the child was 18 and 36 months old. The association were examined using logistic regression and reported as odds ratio (OR) and 95% confidence intervals (CI). The analyses were adjusted for potential confounders (sex, parity, maternal rheumatic disease, early-life antibiotic exposure, region of birth and maternal education).
**Results**
The total sample analyzed at 18 months was 75 819 mother-child pairs of which 236 children were JIA cases. At 36 months old the total sample was 53 612 with a total of 173 JIA cases. No significant association was found between time spent outdoors at 18 (OR=1.06, 95% CI:0.66-1.70) and 36 months (OR=1.64, 95% CI:0.74-3.65) and later risk of JIA. The association between screen time at 18 (OR=1.00, 95% CI:0.58-1.70) or 36 months (OR=0.85, 95% CI:0.42-1.72) and later risk of JIA was nonsignificant. Sensitivity analyses excluding children that already developed JIA prior to exposure, did not alter the results.
**Conclusion**
We found no evidence for an association between time outdoor or screen time at age 18 and 36 months, and later risk for JIA. Time outdoor and screen time were parent-reported and may be affected by information bias, potentially limiting the ability to capture the true variation in physical inactivity during early childhood. Future studies using more detailed and objective measured assessment of physical activity and sedentary behavior are warranted to further explore the role of lifestyle factors in JIA development.
**Disclosure of interest**
None declared.

## P328

### Epigenetic and transcriptomic signatures of treatment response in juvenile idiopathic arthritis: a prospective study design

L. Leko[1,2,*], B. Vrhovšek[2,3], J. Kovač[2,3], T. Avčin[1,2], M. Zajc Avramovič[1], N. Emeršič[1], B. Jenko Bizjan[2,3]
[1]Department of Allergology, Rheumatology and Clinical Immunology of the Paediatric Clinic, University Medical Center Ljubljana; [2]Faculty of Medicine, University of Ljubljana; [3]Clinical Institute for Special Laboratory Diagnostics of the Paediatric Clinic, University Medical Center Ljubljana, Ljubljana, Slovenia
**Correspondence:** L. Leko
*Pediatric Rheumatology 2026*, **24(S1):**P328

**Introduction**
Juvenile idiopathic arthritis (JIA) is the most common chronic rheumatic disease in children, with an estimated prevalence of 30 per 100,000 in Europe [1, 2]. Despite advances in treatment, a significant proportion of patients fail to achieve inactive disease or sustained clinical remission off medication [3]. DNA methylation regulates immune cell development and function, and its dysregulation has been implicated in autoimmune pathogenesis [4]. However, epigenetic predictors of treatment response in JIA remain largely unexplored [5].
**Objectives**
To identify baseline DNA methylation patterns and gene expression profiles in peripheral blood that distinguish JIA patients who achieve disease control with methotrexate (MTX) monotherapy from those requiring escalation to biologic therapy.
**Methods**
This prospective study includes 30 newly diagnosed oligoarticular and polyarticular JIA patients (ILAR criteria) at the Paediatric Clinic, University Medical Centre Ljubljana. Peripheral blood is collected at diagnosis, before immunomodulatory treatment. Patients are followed for 12 months and classified as treatment-responsive (JADAS ≤1.4 on MTX monotherapy) or treatment-refractory (requiring biologic escalation). Genomic DNA undergoes long-read whole-genome sequencing (Oxford Nanopore Technologies) enabling direct detection of 5-methylcytosine. Total RNA isolated from PAXgene tubes undergoes bulk RNA sequencing (Illumina NovaSeq 6000). Bioinformatic analysis includes differentially methylated region (DMR) identification, differential gene expression analysis (DESeq2), and multi-omics integration of methylation and expression data with pathway enrichment of immune-related processes.
**Results**
30 patients have been recruited. DNA sequencing is ongoing, with patients sequenced to date at a mean coverage of 31.7x. Read length N50 is 24 kb (range 10–30 kb), meaning half of bases in reads ≥24 kb. We anticipate identifying coordinated epigenetic-transcriptional alterations in immune pathways, particularly involving monocyte activation, T cell regulation, and proinflammatory cytokine signalling, that differentiate treatment-refractory from treatment-responsive patients at disease onset.
**Conclusion**
This multi-omics study combines long-read sequencing for methylation profiling with RNA sequencing to characterise baseline molecular differences associated with treatment response in JIA. Findings may contribute to understanding early immune dysregulation underlying treatment refractoriness and inform future development of stratified treatment approaches in paediatric rheumatology.

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**Disclosure of interest**
None declared.

## P329

### Applied aspects of lectin histochemistry of the HIP joint

A. Fedotchenko[1,*]
[1]Zaporizhzhia State Medical and Pharmaceutical University, Zaporizhzhia, Ukraine
**Correspondence:** A. Fedotchenko
*Pediatric Rheumatology 2026*, **24(S1):**P329

**Introduction**
Glycoconjugates are fundamental components of joint tissue and play a critical role in regulating innate immunity and inflammation (Bernardi et al., 2012; Li et al., 2019; Chen et al., 2021).
**Objectives**
to establish the practical significance of the lectin histochemistry of the hip joint.
**Methods**
The hip joint of intact laboratory rats by using lectins conjugated with horseradish peroxidase (HRP): peanut (PNA), soybean (SBA), Helix pomatia (HPA), wheat germ (WGA), Perca fluviatilis (PFA), Sambucus nigra (SNA), Lens culinaris (LCA), and Vicia sativa (VSA) from birth till the 90th day was studied. The detailed distribution of these lectins was described previously (Fedotchenko, 2023, 2024, 2025).
**Results**
This summary work emphasises the phenomenon of strong expression of lectin receptors at the interfaces (borders) of certain morphologically different joint structures: the modified basement membrane of the synovium (synovium-synovia interface); the most superficial zone

of the articular cartilage (cartilage-synovia interface); the capsule of the lacunae of chondrocytes (chondrocyte-interterritorial matrix interface); the marginal cartilage (cartilage-synovium interface); the growth plate (cartilage-bone interface); osteoid (bone-periosteum interface); endosteum (bone-marrow interface); articular cartilage during the endochondral ossification (ossification-cartilage interface). Glycoconjugates provide, in this case, anatomical delimitation, serving as barriers against pathogens, activated immunocompetent cells, immune complexes, etc. (Rosen 1993; Dos Santos Silva et al., 2019; Hatinguais et al., 2022; Alba et al., 2023; Nardini et al., 2024), thereby ensuring normal functioning of the joint. The abundant presence of glycoconjugates in the marginal cartilage (MC), through their ability to mediate adhesion of immune cells, immune complexes, etc., might prevent articular cartilage (AC) from being recognized by them and, consequently, from immune-mediated invasion of the marginal synovium (MS) into it, as happens in rheumatoid arthritis. Noteworthy is the strong expression of galactose and mannose residues in the MC. Galactose residues are believed to play a dual role in joint inflammation (Valero-Martinez et al., 2025). Their importance for the MC most likely lies in maintaining its structure. Mannose conjugates, in turn, reduce pro-inflammatory cytokine levels (Torretta et al., 2020), inhibit osteoclastogenesis (Dong et al., 2019), and suppress the transition of monocytes into dendritic cells (Xu et al., 2015), thereby exhibiting immunomodulatory effects. Thus, a strong expression of galactose- and mannose-specific lectins in the normal MC is of a protective nature and confirms the statement that their depletion is strongly associated with the development of RA, its severity, and poor prognosis (Graudal et al., 1998; Albrecht et al., 2014; Saevarsdottir et al., 2001; Schwedler et al., 2018; Nie et al., 2025).

**Conclusion**

The presented data reveal the clinical aspects of joint morphology and provide morphological evidence for lectin-mediated strategies in the treatment of joint immunoinflammatory diseases (Hamilton et al., 2022; Wang et al., 2021; Si et al., 2024).

**Disclosure of interest**

None declared.

## P330

### B-Regulatory cells and PD-L1 expression in juvenile idiopathic arthritis

A. Koutsonikoli[1], A. Taparkou[1], P. Pratsidou-Gertsi[1,*], V. Sgouropoulou[1], M. Trachana[1]

[1]Pediatric Immunology and Rheumatology Referral Centre, 1st Pediatric Dept, Hippokrateio General Hospital, Aristotle University of Thessaloniki, Thessaloniki, Greece

**Correspondence:** P. Pratsidou-Gertsi

*Pediatric Rheumatology 2026*, **24(S1):**P330

**Introduction**

Regulatory B-cells (Bregs) represent a functionally distinct and phenotypically diverse subset of B-cells which inhibit inflammatory responses, by suppressing T-cell activation and inducing regulatory T-cell differentiation. Bregs can function via cell–cell contact and immune checkpoint molecules such as Programmed cell Death Ligand-1 (PD-L1). Currently, no single universal marker exists for Bregs and they are commonly identified by combinations of several markers. Data regarding Bregs and PD-L1 expression in Juvenile Idiopathic Arthritis (JIA) are scarce and conflicting.

**Objectives**

To investigate whether Bregs and cellular PD-L1 expression are quantitatively dysregulated in active JIA.

**Methods**

Using flow cytometry, we determined Bregs in peripheral blood and evaluated the PD-L1 expression on B-cells, monocytes and Bregs, in JIA patients (in active and inactive state) and age and sex matched healthy controls (HCs). Bregs were identified as a percentage of the B-cell population, with 6-color flow cytometry. Since there is no universal identification phenotype, we used two proposed combinations of markers, namely: CD19+CD20+CD5+CD1d+ (Breg1) and CD19+CD20+CD24hiCD38hi (Breg2). Inactive JIA (on/off treatment) was defined according to Wallace criteria.

**Results**

This was a cross-sectional study involving 35 JIA patients (24 female) of a median (range) age of 14 (2-19) years, with a oligoarthritic (20.5%), polyarthritic (35%), psoriatic (12%), entesitis-related (20.5%), systemic (12%) course. Twenty HCs were also tested. The percentage of Breg1 was significantly higher in JIA patients [3.5% (0.1-11.8)] compared to HCs [1.5% (0.8-2.1)] ($p$=0.011). The expression of PD-L1 on B-cells was significantly higher in active [86.8% (56.4-94)] compared to inactive [66.6% (52.8-96.4)] ($p$=0.012) JIA patients. Treatment did not significantly affect this association. There was a strong positive correlation between the expression of PD-L1 on Breg2 and on total B-cells (ρ=0.57, $p$<0.001). Breg1 were also positively correlated with Breg2 (ρ=0.328, $p$=0.05).

**Conclusion**

These findings indicate that Breg production and PD-L1 expression tend to increase in JIA, possibly aiming to collaboratively suppress inflammation. Further studies are warranted to validate these results and determine whether functional dysregulation hinders their immunoregulatory effects.

**Disclosure of interest**

None declared.

## P331

### Post-dress immune dysregulation mimicking systemic inflammatory disease in a child with probable autoimmune encephalitis

T. R. Vasilev[1,*], P. Shahid[1], T. Paneva[1], D. Stamatov[1], G. Tacheva[1], I. Litvinenko[1]

[1]Department of Pediatrics, Medical University of Sofia, Sofia, Bulgaria

**Correspondence:** T. R. Vasilev

*Pediatric Rheumatology 2026*, **24(S1):**P331

**Introduction**

Autoimmune encephalitis (AE) in childhood represents a severe neuroinflammatory condition requiring early immunomodulatory therapy. Drug reaction with eosinophilia and systemic symptoms (DRESS), most commonly triggered by aromatic antiepileptic drugs, is a rare but potentially life-threatening hypersensitivity reaction associated with prolonged immune dysregulation. Increasing evidence suggests that DRESS may act as a trigger of sustained immune activation, mimicking systemic inflammatory or autoimmune diseases.

**Objectives**

To describe a pediatric case illustrating the interplay between autoimmune encephalitis, DRESS syndrome and subsequent persistent immune dysregulation, with emphasis on diagnostic challenges and immunoregulatory mechanisms.

**Methods**

We present a clinical case of a 13-year-old girl with probable autoimmune encephalitis, complicated by DRESS syndrome and a relapsing hyperinflammatory course. Clinical, laboratory, immunological and neuroimaging data were analysed longitudinally. The clinical course and therapeutic interventions were reviewed in a multidisciplinary setting.

**References: Results**

The patient initially presented with seizures and impaired consciousness, with MRI findings consistent with bilateral encephalitic involvement. Infectious and neuronal antibody panels were negative, and empirical immunotherapy with intravenous immunoglobulin and corticosteroids was initiated. During treatment with aromatic antiepileptic drugs, she developed DRESS syndrome, characterised by rash, eosinophilia, cytopenia and hepatic involvement, with CMV reactivation and HLA-associated susceptibility. Withdrawal of the offending drugs and corticosteroid therapy resulted in transient improvement; however, re-exposure led to relapse. Subsequently, the patient developed a hyperinflammatory state with pancytopenia, hyperferritinaemia, livedo-like rash and neuropsychiatric deterioration, raising differential diagnoses including relapse of AE, systemic autoimmune disease and drug-related immune dysregulation. Repeated courses of corticosteroid pulses and intravenous immunoglobulin, followed by cyclosporine, led to partial clinical and laboratory remission, although attempts at steroid tapering were associated with disease flares. Follow-up neuroimaging demonstrated regression of inflammatory lesions but progression to cerebral atrophy.

**Conclusion**

This case highlights DRESS syndrome as a potent inducer of prolonged immune dysregulation, capable of mimicking systemic inflammatory disease and complicating the course of autoimmune encephalitis. The

observed relapsing hyperinflammatory phenotype supports the concept of persistent immune activation beyond drug withdrawal. Early recognition, avoidance of re-exposure and individualized immunomodulatory strategies are essential. Multidisciplinary management and long-term follow-up are required to address the risk of chronic immune-mediated complications.

**Disclosure of interest**
None declared.

## P332

### A rare case of scleromyositis overlap syndrome in a pediatric patient in western Guatemala

M. M. Rodriguez Reyes[1], F. A. Vasquez Triminio [2,*], Z. I. Garcia Murillo[2]
[1]Kids Centro Pediátrico, Guatemala, Guatemala; [2]Pediatria, Hospital Universitario "Dr. José Eleuterio Gonzalez ", Monterrey, Mexico
**Correspondence:** F. A. Vasquez Triminio
*Pediatric Rheumatology 2026*, **24(S1):**P332

**Introduction**
Systemic Sclerosis is an autoimmune disease characterized by skin thickening and internal organ involvement. In its pediatric form, it may present as either diffuse or limited disease and may occasionally be associated with myositis or dermatomyositis. Patients with scleromyositis overlap syndrome not only develop a clinical presentation.

**Objectives**
To report a rare case in Guatemala of a 10 year old male presenting with Systemic Sclerosis overlapping with inflammatory myopathy.

**Methods**
A previously healthy 10-year-old boy presented with progressive lower extremity pain that began five months before evaluation. Due to persistent symptoms and progressive clinical deterioration, he returned one week prior to admission with worsening pain, joint swelling, and fever, leading to hospitalization with an initial suspicion of JIA. Physical examination revealed characteristic cutaneous and musculoskeletal findings suggestive of inflammatory myopathy and systemic sclerosis overlap. A heliotrope rash was observed on both eyelids, along with Gottron's papules over the metacarpophalangeal and interphalangeal joints. The patient also exhibited dry skin, skin thickening, and sclerodactyly. Range of motion was limited in both shoulders, elbows, and wrists, with inability to raise the arms above ear level, consistent with proximal muscle weakness. Functional assessment demonstrated significant muscle involvement. Laboratory evaluation showed markedly elevated muscle enzymes, CPK 2826U/L and LDH 1240 U/L, consistent with active myositis. Mild transaminase elevation was also present. Autoimmune testing revealed positive ANA with homogeneous nuclear pattern (1:80), while anti-RNP/Sm antibodies were negative.Myositis-specific and overlap antibodies were positive for anti-Ku, anti-PM-Scl100(+++), anti-PM-Scl75(+++), SRP, and PL-7+. The systemic sclerosis antibody panel was negative for anti-Scl70, anticentromere, and anti-RNA polymerase III antibodies. High-resolution chest CT: mild bilateral basal interstitial lung involvement. Nailfold capillaroscopy: edema, decreased capillary density, and dilated capillaries, supporting underlying microvascular disease. The combination of proximal muscle weakness, elevated muscle enzymes, heliotrope rash, Gottron's papules, skin thickening, sclerodactyly, abnormal capillaroscopy, and anti-PM/Scl positivity supported the diagnosis of juvenile scleromyositis overlap syndrome, a rare condition characterized by overlapping features of Myositis and Systemic Sclerosis. Treatment included prednisone, methotrexate, monthly intravenous immunoglobulin.

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**Disclosure of interest**
None declared.

## P333

### Capillaroscopic features of juvenile systemic sclerosis: results of a two-center study

I. Avrusin[1,*], V. Podzolkova[2], P. Chikova [1], E. Klochkova[1], A. Yakovlev[1], K. Aleksanyan[3], O. Kalashnikova[1], I. Chikova[1], M. Osminina[2], P. Berezhanskiy[2], M. Kostik[1]
[1]Hospital Pediatrics, Saint Petersburg State Pediatric Medical University, Saint Petersburg; [2]Department of Children's Diseases, Sechenov First Moscow State Medical University; [3]Department of Pediatric Rheumatology, Sechenov Center of Maternity and Childhood, Moscow, Russian Federation
**Correspondence:** I. Avrusin
*Pediatric Rheumatology 2026*, **24(S1):**P333

**Introduction**
Juvenile systemic sclerosis (jSSc) is a systemic immune-mediated connective tissue disorder characterised by fibroproductive changes and microvascular abnormalities. Nailfold capillaroscopy (NFC) allows real-time assessment of microcirculation. While considerable data exist on capillaroscopic patterns in adult systemic sclerosis, data on the juvenile form remain limited.

**Objectives**
To evaluate the capillaroscopic pattern in children with jSSc, with a focus on differences between younger patients and those aged ≥14 years (young adults).

**Methods**
This two-center study included 43 jSSc patients (35 girls, 8 boys) aged 4–17 years (median 14 years). NFC was performed followed the standardised criteria for systemic sclerosis and Raynaud's phenomenon [1].

**Results**
Low capillary density was found in 81.4% of patients (median 5 capillaries/mm). Capillary dilation was observed in 95.4%, and giant capillaries in 46.5%; the median maximum apex diameter was 45 µm. Abnormal capillary morphology was present in 93%, including bushy capillaries (65.1%), widened apex (65.1%), interdigitated (53.5%), and spire-shaped (48.8%). Haemorrhages were detected in 34.9%, and perivascular oedema in 57.1%. Patients aged ≥14 years at the time of capillaroscopy ($n$=22) had significantly longer disease duration before NFC (median 60 vs. 28 months, $p$=0.016) and a longer diagnostic delay (median 12 vs. 7.5 months, $p$=0.004) compared to those <14 years ($n$=21). Clinically, the older group more frequently had fingertip ulcers (22.7% vs. 0%, $p$=0.048) and gastrointestinal involvement (63.6% vs. 33.3%, $p$=0.047). On NFC, low capillary density was significantly more common in the older group (95.5% vs. 66.7%, $p$=0.021).

**Conclusion**
NFC demonstrates progressive capillary loss and dilatation in jSSc, correlating with disease duration and late manifestations (digital ulcers, GI involvement) in patients ≥14 years. It serves as a noninvasive biomarker of disease burden and a monitoring tool. Future trials should assess if early therapy improves capillaroscopic and clinical outcomes.

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**Disclosure of interest**
None declared.

## P334

### Juvenile localized scleroderma: clinical characteristics and therapeutic outcomes in a single-centre cohort

B. Rinaldi[1,*], F. Di Stasio[2], F. Minoia[3], F. Vianello[3], A. Losi[1], G. Filocamo[3]
[1]Pediatric immunorheumatology, University of Milan, Milano; [2]Pediatric immunorheumatology, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Mlano; [3]Pediatric immunorheumatology, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milano, Italy
**Correspondence:** B. Rinaldi
*Pediatric Rheumatology 2026*, **24(S1):**P334

**Introduction**
Juvenile Localized Scleroderma (JLS) is a rare inflammatory and fibrosing disorder affecting the skin and underlying tissues, with heterogeneous clinical phenotypes and possible extracutaneous involvement. Early treatments are essential to prevent progression and functional sequelae.
**Objectives**
To describe clinical characteristics, disease subtypes, therapeutic approaches, LOSCAT assessment in a monocentric pediatric cohort with JLS.
**Methods**
We conducted a single-center observational study of JLS patients at a pediatric rheumatology centre. Demographic and clinical data were collected between December 2014 and March 2026. Disease activity and damage were assessed using the Localized Scleroderma Cutaneous Assessment Tool (LOSCAT), including the modified Localized Scleroderma Skin Severity Index (mLoSSI) and the Localized Scleroderma Skin Damage Index (LoSDI), at treatment initiation and during follow-up.
**Results**
The cohort included 18 patients (10 females, 8 males). Median age at onset was 5.88 years (IQR 4.1-8.8) and median age at diagnosis was 6.88 years (IQR 4.6-9.8), with a median diagnostic delay of 11.99 months. Linear morphea was the most common subtype (8/18, 44%), including 4 craniofacial forms (2 *en coup de sabre* and 2 Parry–Romberg). Generalized morphea occurred in 5/18 (28%) and circumscribed morphea in 4/18 (22%). One patient developed a flexor tendon retraction as a complication. Topical corticosteroids were used in 12 patients, pimecrolimus in one, and tacrolimus in another. Systemic treatment was required in 16/18 patients (89%). Methotrexate was administered in 16/18 (89%); 10/16 (63%) received intravenous methylprednisolone pulses and 4/16 (22%) oral corticosteroids. Five patients (28%) required escalation to mycophenolate mofetil (MMF) because of new lesions ($n$=4) or insufficient response ($n$=1). Patients who responded to first-line therapy had baseline median mLoSSI and LoSDI scores of 3 (IQR 1–4) and 4 (IQR 3–8), respectively. At 9 months, median mLoSSI decreased to 0, while LoSDI was 3 (IQR 2–6). Time from onset to treatment initiation was 12.6 months (IQR 9.9–18.2). Patients requiring MMF had baseline median mLoSSI and LoSDI scores of 10 (IQR 7–12) and 6 (IQR 5–7), respectively. At MMF initiation, median mLoSSI and LoSDI were 8 (IQR 7–11) and 13 (IQR 7–26); after 4 months, mLoSSI decreased to 2 (IQR 0–3), while LoSDI was 11 (IQR 0–26). Time from onset to treatment initiation was 13.6 months (IQR 13.2–15.6).
**Conclusion**
Linear morphea, especially craniofacial variants, was predominant with a high need for systemic therapy. Overall, LOSCAT showed marked reduction in disease activity, with less improvement in damage scores. Patients requiring MMF showed higher LOSCAT scores at disease onset.
**Disclosure of interest**
None declared.

## P335

### Juvenile systemic sclerosis with dermatomyositis-like overlap features represents a distinct clinical phenotype: updates from the juvenile scleroderma inception cohort

I. Foeldvari[1,*], K. Torok[2], J. Klotsche[3], O. Kasapcopur[2], A. Adrovic[2], B. Feldman[2], F. Sztajnbok[2], M. T. Terreri[2], A. P. Sakamoto[2], J. Anton[2], S. Johnson[2], R. Khubchandani[2], S. Hajek[2], V. Stanevicha[2], G. Özomay Baykal[2], E. Alexeeva[2], D. Schonenberg-Meinema[2], E. Al-Abadi[2], M. Katsicas [2], S. Sawhney[2], V. Smith[2], S. Abu Al-Saoud[2], S. Appenzeller[2], T. Avcin [2], N. Cabrera[2], H. D. Dara Kar[2], S. O. Hetlevik[2], M. Kostik[2], T. Lehman[2], S. Li[2], H. Malcova[2], E. Marrani[2], C. Pain[2], A. Patwardhan[2], W.-A. Sifuentes-Giraldo[2], N. Vasquez-Canizares[2], P. Costa Reis[2], M. Janarthanan [2], M. Moll[2], D. Nemcova [2], M. J. Santos[2], C. Battagliotti[2], L. Berntson[2], B. Bica[2], J. Brunner[2], D. Eleftheriou[2], L. Harel[2], A. Horne [2], G. Horneff[2], D. Kaiser[2], T. Kallinich[2], D. Lazarevic[2], J. M. López Benítez[2], K. Minden[2], S. Nielsen[2], F. Nuruzzaman[2], P. Seferna[2], M. Sparchez[2], Y. Uziel [2], N. Helmus[1]
[1]Hamburg Centre for Pediatric and Adolescence Rheumatology; [2]jSSc collaborative group, Hamburg; [3]German Rheumatism Research Center, Berlin, Germany
**Correspondence:** I. Foeldvari
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**Introduction**
Juvenile systemic sclerosis (jSSc) is a rare and heterogeneous autoimmune disease. A subset of patients fulfilling classification criteria for jSSc exhibit clinical features characteristic of other connective tissue diseases, most commonly dermatomyositis. These patients are often referred to as having an "overlap" phenotype. The clinical relevance and disease pattern associated with this presentation in pediatric systemic sclerosis remain incompletely defined. The Juvenile Scleroderma Inception Cohort (jSScC), the largest multinational prospective cohort of jSSc patients, provides a unique opportunity to characterize this subgroup early in the disease course.
**Objectives**
To compare demographic characteristics, clinical features, autoantibody profiles, and disease damage between jSSc-overlap (OV+) and jSSc-non-overlap(OV-) patients at the time of cohort inclusion.
**Methods**
Clinical data were extracted from the jSScC up to 15 December 2025. Patients fulfilling classification criteria for jSSc were categorized as OV+ or OV- based on the presence or absence of dermatomyositis-like and other connective tissue disease overlap features at the time of cohort inclusion. Demographic data, clinical characteristics, organ involvement, autoantibody profiles, and patient- and physician-reported outcomes were compared between groups. Categorical variables were compared using $\chi^2$ or Fisher's exact tests, and continuous variables using non-parametric tests. A two-sided p value <0.05 was considered statistically significant.
**Results**
Among 291 patients included, 62 (21%) were classified as OV+. OV+ were present in 17% of patients with diffuse cutaneous jSSc and 58% of those with limited cutaneous jSSc ($p$<0.001). The proportion of patients of Caucasian origin was similar (61–72%). There were no differences in mean age at onset of Raynaud's phenomenon or first non-Raynaud manifestation. Median disease duration at inclusion was approximately 2.5 years, and 90% of patients in both groups were receiving disease-modifying therapy. Compared with OV- patients with OV+ the significant differences regaring clinical presentation of antibody pattern are described in Table 1. No significant differences were observed in other organ systems. Patient- or parent-reported global disease damage (VAS 0–100) was significantly higher in OV+(50 vs. 30, $p$=0.021).

**Table 1 (Abstract P335)** Patients characteristic with and without overlap features

| At time of inclusion in the cohort | Patients without overlap *N*=229 | Patients with overlap *N*=62 | *P* value |
|---|---|---|---|
| Anti-scl 70 | 35% (75/216) | 14% (8/56) | 0.003 |
| Anti-PMScl | 10% (12/124) | 33% (16/49) | <0.001 |
| MRSS, median (IQR) | 11 (5 – 23) *n*=219 | 7 (3 – 18) *n*=61 | 0.001 |
| Gottron Papules | 22% (50/228) | 44% (26/59) | 0.001 |
| Raynaud´s phenomenon | 94% (215/229) | 82% (51/62) | 0.004 |
| Abnormal findings on HRCT | 40% (70/175) | 61% (31/51) | 0.022 |
| Only Pulmonary Involvement | 40% (92/229) | 55% (34/62) | 0.041 |
| Presence of joints with decreased range | 58% (131/227) | 80% (48/60) | 0.002 |
| Muscle Weakness | 16% (32/206) | 36% (21/58) | 0.005 |

**Conclusion**
jSSc patients with OV+, predominantly consistent with dermatomyositis, represent a distinct clinical phenotype characterized by a predominance of limited cutaneous disease, higher anti-PM/Scl antibody positivity, increased musculoskeletal involvement, and more frequent interstitial lung disease on HRCT. Despite lower skin scores, OV+ patients report greater overall disease damage early in the disease course. Recognition of this phenotype may have important implications for disease monitoring, risk stratification, and individualized management in jSSc.
**Disclosure of interest**
None declared.

## P336

### Clinical characteristics associated with progressive skin involvement in juvenile systemic sclerosis: 12-month data from the juvenile scleroderma inception cohort

I. Foeldvari[1,*], K. Torok[2], J. Klotsche[3], O. Kasapcopur[2], A. Adrovic[2], B. Feldman[2], M. T. Terreri[2], A. P. Sakamoto[2], F. Sztajnbok[2], J. Anton[2], S. Johnson[2], M. Katsicas [2], R. Khubchandani[2], V. Stanevicha[2], S. Abu Al-Saoud[2], E. Alexeeva[2], S. Appenzeller[2], T. Avcin [2], S. Li[2], E. Marrani[2], W.-A. Sifuentes-Giraldo[2], E. Al-Abadi[2], P. Costa Reis[2], S. Hajek[2], D. Nemcova [2], D. Schonenberg-Meinema[2], C. Battagliotti[2], L. Berntson[2], B. Bica[2], J. Brunner[2], N. Cabrera[2], D. Eleftheriou[2], L. Harel[2], S. O. Hetlevik[2], G. Horneff[2], M. Janarthanan [2], T. Kallinich[2], T. Lehman[2], K. Minden[2], M. Moll[2], S. Nielsen[2], A. Patwardhan[2], V. Smith[2], M. Sparchez[2], N. Helmus[1]
[1]Hamburg Centre for Pediatric and Adolescence Rheumatology; [2]jSSc collaborative group, Hamburg; [3]German Rheumatism Research Center, Berlin, Germany
**Correspondence:** I. Foeldvari
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**Introduction**
At a recent international consensus meeting in Hamburg, a multidisciplinary expert group proposed a working definition for progressive skin involvement(prsk) in juvenile systemic sclerosis (jSSc) (Table 1). Applying this definition to longitudinal cohort data may help characterize clinical features associated with skin progression.
**Objectives**
To apply this consensus definition of prsk to the Juvenile Scleroderma Inception Cohort (jSScC) and to describe clinical characteristics associated with prsk over 12 months.
**Methods**
Data were extracted from the jSScC up to 15 December 2025. Patients were categorized according to whether they met criteria for prsk over 12 months. Demographic characteristics, clinical features, laboratory parameters, and patient-reported outcomes were compared between patients with and without prsk at cohort inclusion and after 12 months of follow-up. Categorical variables were compared using $\chi^2$ or Fisher's exact tests, and continuous variables using non-parametric tests. A two-sided p value <0.05 was considered statistically significant.
**Results**
A total of 168 patients were included; 33 (20%) met criteria for prsk. The female-to-male ratio was higher among patients with prsk+ compared with those without prsk (prsk-) (approximately 5–6:1 vs. 3.5:1). Approximately 75% of patients in both groups had diffuse cutaneous disease. Median age at first non-Raynaud manifestation was similar between groups (10.0 vs. 10.9 years). Median disease duration at baseline was 3.7 years in both groups and around 70% were of Caucasian origin. At baseline, antinuclear antibody positivity was more frequent among patients prsk- (92% vs. 79%, $p$=0.021), while no differences were found for other autoantibodies. After 12 months, creatine kinase levels were more frequently elevated among patients with prsk+ (23% vs. 9%, $p$=0.069). Median mRSS increased by 5 points over 12 months in patients with prsk+, compared with a median decrease of 7 points in patients with prsk- ($p$=0.001). Sclerodactyly at 12 months was more common among patients with prsk+ (81% vs. 65%, $p$=0.071). prsk+ was associated with a significantly higher frequency of active digital ulcerations at 12 months (30% vs. 12%, $p$=0.009). A greater proportion of patients with prsk+ had DLCO <80% predicted at 12 months (73% vs. 47%, $p$=0.073). Patient-reported global disease activity (VAS 0–100) was significantly higher at 12 months among patients with prsk+ (30 vs. 20, $p$=0.049), and patient-reported disease damage also tended to be higher (25 vs. 17, $p$=0.061).

**Table 1 (Abstract P336)** Definition of progressive skin involvement

| Items defining progressive skin disease |
|---|
| (1) an increase in modified Rodnan skin score (mRSS) of ≥2 points if prior mRSS was <10; |
| (2) an increase of ≥4 points if prior mRSS was ≥10; or |
| (3) development of a new area or extension of skin involvement. |

**Conclusion**
Application of a consensus definition for prsk identified a subset of patients with distinct clinical characteristics, including increasing skin scores, higher rates of digital ulceration, laboratory evidence of muscle involvement, and higher patient-reported disease activity over 12 months. These findings describe characteristics associated with prsk rather than true predictors.
**Disclosure of interest**
None declared.

## P337

### Gender differences in clinical patterns in juvenile systemic sclerosis: updates from the juvenile scleroderma inception cohort

I. Foeldvari[1,*], K. Torok[2], J. Klotsche[3], O. Kasapcopur[2], A. Adrovic[2], B. Feldman[2], F. Sztajnbok[2], M. T. Terreri[2], A. P. Sakamoto[2], J. Anton[2], S. Johnson[2], R. Khubchandani[2], S. Hajek[2], V. Stanevicha[2], G. Özomay Baykal[2], E. Alexeeva[2], D. Schonenberg-Meinema[2], E. Al-Abadi[2], M. Katsicas [2], S. Sawhney[2], V. Smith[2], S. Abu Al-Saoud[2], S. Appenzeller[2], T. Avcin [2], N. Cabrera[2], H. D. Dara Kar[2], S. O. Hetlevik[2], M. Kostik[2], T. Lehman[2], S. Li[2], H. Malcova[2], E. Marrani[2], C. Pain[2], A. Patwardhan[2], W.-A. Sifuentes-Giraldo[2], N. Vasquez-Canizares[2], P. Costa Reis[2], M. Janarthanan [2], M. Moll[2], D. Nemcova [2], M. J. Santos[2], C. Battagliotti[2], L. Berntson[2], B. Bica[2], J. Brunner[2], D. Eleftheriou[2], L. Harel[2], A. Horne [2], G. Horneff[2], D. Kaiser[2], T. Kallinich[2], D. Lazarevic[2], J. M. López Benítez[2], K. Minden[2], S. Nielsen[2], F. Nuruzzaman[2], P. Seferna[2], M. Sparchez[2], Y. Uziel [2], N. Helmus[1]
[1]Hamburg Centre for Pediatric and Adolescence Rheumatology; [2]jSSc collaborative group, Hamburg; [3]German Rheumatism Research Center, Berlin, Germany
**Correspondence:** I. Foeldvari
*Pediatric Rheumatology 2026*, **24(S1):**P337

**Introduction**
Juvenile systemic sclerosis (jSSc) is a rare, severe autoimmune disease with an estimated prevalence of approximately 3 per 1,000,000 children. In adult systemic sclerosis, male sex is associated with a more severe disease course and increased organ involvement. Whether similar sex-related differences are present in pediatric-onset disease remains incompletely understood. The Juvenile Scleroderma Inception Cohort (jSScC) is the largest multinational prospective cohort of patients with jSSc worldwide and provides a unique opportunity to explore gender-associated differences early in the disease course.
**Objectives**
To explore differences in clinical presentation, disease characteristics, and patient- and physician- reported outcomes between female and male patients with jSSc at the time of cohort inclusion.
**Methods**
Data were extracted from the jSScC up to 15 December 2025. Demographic characteristics, clinical features, organ involvement, and patient- and physician-reported outcomes were analyzed. The jSScC is a multinational, prospective inception cohort, as previously described. Categorical variables were compared using $\chi^2$ or Fisher's exact tests, and continuous variables using non-parametric tests. A two- sided p value <0.05 was considered statistically significant.
**Results**
A total of 291 patients were included, of whom 234 (80.5%) were female and 57 (19.5%) were male(female-to-male ratio 4:1). A higher proportion of male patients had diffuse cutaneous disease compared with females (77% vs. 66%, p=ns). Approximately 70% of patients in both groups were Caucasian. Median age at onset of the first non-Raynaud symptom was 10.9 years in both groups, and median disease duration at inclusion was approximately 2.4 years. Disease-modifying treatments were used in 91% of female and 86% of male patients. Au-

toantibody profiles did not differ between sexes. Male patients had a significantly higher modified Rodnan skin score (mRSS) compared with female patients (median 16 vs. 10, $p$=0.021). A significantly greater proportion of male patients had a 6-minute walk test (6MWT) distance below the 10th percentile (86% vs. 58%, $p$=0.047) and a body mass index (BMI) z-score ≤ −2 (25% vs. 14%, $p$=0.02). No significant differences were observed in pulmonary, cardiac, renal, or musculoskeletal involvement. Patient-reported global disease damage (VAS 0–100) was significantly higher in male patients (50 vs. 30, $p$=0.011), as was physician-rated global damage (VAS 0–100; 40 vs. 25, $p$=0.043).

**Conclusion**

Within a relatively short disease duration (~2.4 years), male patients with jSSc exhibited greater overall disease damage compared with female patients, reflected by higher skin scores, reduced functional capacity, lower BMI, and higher patient- and physician-reported damage. Unlike adult systemic sclerosis, these differences were not driven by increased cardiopulmonary or renal involvement or distinct autoantibody patterns. These findings highlight potential sex-specific disease trajectories in jSSc and underscore the importance of ongoing longitudinal evaluation as the cohort continues to expand.

**Disclosure of interest**

None declared.

## P338

### Raynaud phenomenon as the first manifestation of early juvenile systemic scleroderma: a case report using the VEDOSS approach

D. Gronych[1,*], K. Bouchalova[1], M. Janega[1], Z. Klobouková[1], T. Haiduk[1]
[1]Department of Pediatrics, University Hospital Olomouc, Olomouc, Czech Republic
**Correspondence:** D. Gronych
*Pediatric Rheumatology 2026*, **24(S1):**P338

**Introduction**

Juvenile systemic sclerosis (jSSc) is a rare connective tissue disease characterized by excessive collagen deposition leading to tissue atrophy and fibrosis. jSSc should be considered in children presenting with Raynaud phenomenon (RP) associated with pathological capillaroscopy, digital ulcers, or a positive autoantibody profile characteristic of systemic sclerosis (SSc). There are currently no validated diagnostic criteria for jSSc. In clinical practice, the ACR/EULAR 2013 classification criteria are commonly used, although they appear to have lower sensitivity and specificity in juvenile-onset disease compared to adult-onset SSc. The VEDOSS (Very Early Diagnosis of Systemic Sclerosis) approach is an algorithm emphasizing the follow-up of patients with RP, puffy fingers, and positive antinuclear antibodies (ANA). It is considered useful for identifying very early stages of systemic sclerosis. However, no studies have yet evaluated these criteria in pediatric patients.

**Objectives**

We present a case report of a pediatric patient with Raynaud phenomenon and suspected very early systemic sclerosis. The aim is to highlight diagnostic aspects of the early disease stage.

**Methods**

A 15-year-old boy was referred from a nephrology outpatient clinic due to suspected Raynaud phenomenon. His history was dominated by knee arthralgia, episodes of finger blanching and cyanosis, mild hypesthesia, and occasional oral aphthae. Symptoms began during the previous winter season. The patient was admitted for diagnostic work-up. Capillaroscopy revealed pathological findings. Initial imaging (X-ray and ultrasound of the knees, abdominal ultrasound) showed no abnormalities related to connective tissue disease. Cardiological examination revealed mild mitral valve prolapse without signs of pulmonary hypertension. Pulmonary function testing (spirometry, DLCO, and body plethysmography) demonstrated impaired gas diffusion without evidence of restrictive or obstructive lung disease. Given the suspicion of very early systemic sclerosis, the patient was managed according to a VEDOSS-based approach. Outpatient esophageal manometry revealed early esophageal motility disturbances, while low-dose lung HRCT showed no pathology. Laboratory testing revealed positive ANA (titer 1:80). During hospitalization, no ischemic complications of the acral extremities developed. Vasodilatory therapy was initiated (calcium channel blocker, naftidrofuryl, and Ginkgo biloba). Juvenile systemic sclerosis is a rare connective tissue disease with often insidious onset. This case highlights the need to further validate the VEDOSS criteria for the detection of very early systemic sclerosis in pediatric patients. Early recognition may help prevent severe organ involvement. Initial treatment focuses on management of Raynaud phenomenon, which is often the first clinical manifestation, followed by therapy targeting the underlying disease, typically including immunosuppressive treatment.

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**Disclosure of interest**

None declared.

## P339

### Differential diagnosis of juvenile scleroderma using nailfold capillaroscopy

N. Yudkina[1], A. Volkov[1], I. Nikishina[1,*]
[1]V.A.Nasonova Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** I. Nikishina
*Pediatric Rheumatology 2026*, **24(S1):**P339

**Introduction**

When treating patients with juvenile scleroderma, practicing physicians often encounter difficulties in differentiating between the systemic sclerosis (SSc) and localized scleroderma (LoSD). Despite the characteristic appearance and localization of cutaneous manifestations, extracutaneous manifestations are often encountered, including damage to the nervous system, visual organs, blood vessels, joints, muscles, esophagus, and even lungs, resembling systemic scleroderma. Furthermore, many patients are found to have positive antinuclear antibodies. The different approaches to treating these diseases dictate the need for an accurate diagnosis.

**Objectives**

To determine the role of nailfold videocapillaroscopy (NVC) in the differential diagnosis between LoSD and SSc in pediatric patients.

**Methods**

The study included 65 children aged 2 to 18 years with suspected juvenile scleroderma, referred to our center by a rheumatologist or dermatologist. All patients underwent NVC using a 200x stereomicroscope. Nailbed capillaries on fingers 2–5 of both hands were examined. Parameters assessed included capillary morphology, size, density, and the presence of hemorrhages.

**Results**

In 61 of the 65 children examined, NVC revealed a normal capillaroscopic image without signs of microangiopathy characteristic of SSc. These patients were subsequently diagnosed with localized scleroderma: 26 children were diagnosed with the plaque form, 24 with linear scleroderma affecting the face or extremities, 8 with Parry-Romberg hemiatrophy, and 7 with lichen sclerosus. In the remaining 4 of the 65 patients, NVC revealed pathological changes consistent with the scleroderma pattern: 2 children had the early scleroderma type, and 2 had the active scleroderma type. These patients exhibited clinical manifestations of SSc, including Raynaud's phenomenon, finger swelling, induration of the skin of the hands and face, and SSc-specific antinuclear antibodies (anticentromere antibodies and topoisomerase-1). Based on the results of a comprehensive rheumatological examination, all 4 children were diagnosed with SSc.

**Conclusion**

The NVC method is an important diagnostic tool. It is not complex or difficult to access, is painless, can be performed multiple times due to the absence of radiation exposure, and does not require expensive equipment. It remains indispensable in the differential diagnosis of juvenile scleroderma, as confirmed by the results of our study and the inclusion of NVC in the 2013 ACR-EULAR criteria for SSc.

**Disclosure of interest**

None declared.

P340

**Cardiac involvement in patients with juvenile systemic sclerosis**

S. Rodionovskaya[1,2], V. Komoliatova[1], G. Torosyan[1], M. Shalygina[3], I. Nikishina[1,3,*]
[1]Federal Scientific and Clinical Center for Children and Adolescents of the Federal Medical and Biological Agency of Russia; [2]Medical and Biological University of Innovation and Continuing Education of the Federal Medical Biophysical Center named after A.I. Burnazyan; [3]V.A. Nasonova Research Institute of Rheumatology, Moscow, Russian Federation
**Correspondence:** I. Nikishina
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**Introduction**
Juvenile systemic sclerosis (jSSc) is a rare autoimmune connective tissue disease of childhood. Cardiac involvement includes rhythm and conduction abnormalities and heart failure are associated with an adverse prognosis.
**Objectives**
To determine the frequency of cardiac involvement in patients with jSSc.
**Methods**
A retrospective analysis of clinical, demographic characteristics and examination findings in 11 patients (pts) with jSSc was performed. The diagnosis met the 2013 ACR/EULAR classification criteria. All pts were female; the median age at the disease onset - 11 [9.1–12.2] years, and the median disease duration at diagnosis - 0.7 [0.4–1.2] years. All pts underwent ECG, EchoCG. Holter monitoring (HM) were performed in 5 pts cardiac and cardiac MRI - 3. Follow-up ranged from 1.6 to 11.3 years. One pts suddenly died in 15 years.
**Results**
Cardiac involvement was identified in 7 pts (64%) at a disease duration of 3.1 [0.47–4.5] years. According to ECG, the conduction abnormalities were detected in 6 pts (86%): right bundle branch block in 2 cases, first-degree AV-block in 4 cases, sinus tachycardia in 3 cases and bradycardia in 1 case. Five pts had changes by HM: non-sustained ventricular tachycardia in 2 cases, frequent couple and isolated ventricular premature beats (VPB) in 3 pts and fascicular VPB in one. MRI was performed in three children with jSSc. In two cases, diffuse and focal non-ischemic myocardial changes involving the left ventricle (LV) and the anterior wall of the right ventricle were detected; these pts received biologic therapy with tocilizumab, in one case combined with antiarrhythmic therapy (amiodarone). In the 3-rd case, a 14-year-old patient with cardiac involvement at jSSc onset (VPB, LV dilation, and reduced LV ejection fraction of 54%) was started on biologic therapy with rituximab. Cardiac MRI performed after 18 months when rituximab therapy was started and showed no focal myocardial abnormalities. Ventricular fibrillation was the cause of death in a 15-year-old patient with a 6-year disease duration who had not been receiving biologic therapy, but had couple and isolated VPB by HM.
**Conclusion**
Children with juvenile systemic sclerosis had high cardiac involvement. That including conduction abnormalities and ventricular tachyarrhythmia. All children with juvenile systemic sclerosis and cardiac involvement should be performed cardiac MRI.
**Disclosure of interest**
None declared.

P341

**Successful tocilizumab experience in a pediatric patient with generalized morfea**

Z. Kucukakcali Arslan[1,*], A. Alisan[1], Y. Denli[2], S. Gunasti Topal[2], R. M. Kisla Ekinci[1]
[1]Pediatric Rheumatology; [2]Dermatology, Cukurova University Faculty of Medicine, Adana, Türkiye
**Correspondence:** Z. Kucukakcali Arslan
*Pediatric Rheumatology 2026*, **24(S1):**P341

**Introduction**
Localized scleroderma, also named as morphea is a chronic inflammatory connective tissue disease characterized by the inflammation and fibrosis of the skin and subcutaneous tissues. It presents as skin hardening and discoloration. Generalized morphea is characterized with minimum four plaques larger than 3 cm affecting at least two or more anatomical regions.
**Objectives**
In this case report, we aimed to share our successful experience on tocilizumab use in a pediatric patient with treatment resistant generalized morphea.
**Methods**
A 13-years old girl presented with complaints of redness and hardening of skin that initially started on the right thigh and later spread to both lower and upper extremities. Physical examination revealed widespread sclerotic changes in both lower extremities and linear sclerotic lesions on both arms, accompanied by mild limitation of movement in the knees, elbows, and ankles. Laboratory investigations showed no cytopenia, while mild eosinophilia (AEC: 600 cells/μL) was present. Acute phase reactants and creatine kinase levels were normal. Superficial ultrasonography demonstrated increased skin thickness and high echogenicity down to the knee level in the anterior, lateral, and medial regions of both thighs. Skin biopsy findings were consistent with morphea involving the epidermis, dermis, and subcutaneous adipose tissue. Based on clinical and biopsy findings, the patient was diagnosed with generalized morphea, and treatment with subcutaneous methotrexate 15 mg/week and oral prednisolone 30 mg/day was initiated. At the end of the second week of treatment, new lesions appeared around the umbilical region and progressively increased; therefore, intravenous tocilizumab 400 mg every 2 weeks was added at the end of the first month of treatment. After the second dose of tocilizumab, softening of the skin hardening and improvement in movement limitation were observed. Prednisolone was discontinued in the fifth month of tocilizumab treatment.

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**Disclosure of interest**
None declared.

P343

**Pediatric-onset raynaud's phenomenon preceding systemic sclerosis: a retrospective study**

A. Gkoutzourelas[1,*], A. Gkouvi[1], C. G. Katsiari[1], D. P. Bogdanos[1], T. Simopoulou[1]
[1]Department of Rheumatology and Clinical Immunology, Larissa University Hospital, University of Thessaly, Larissa, Greece
**Correspondence:** A.s Gkoutzourelas
*Pediatric Rheumatology 2026*, **24(S1):**P343

**Introduction**
Raynaud's phenomenon (RP) is common in pediatric populations and is often considered benign; however, it may represent an early manifestation of underlying connective tissue disease, including Systemic Sclerosis (SSc).
**Objectives**
To describe clinical, serological and capillaroscopic characteristics in patients with pediatric-onset RP preceding SSc.
**Methods**
We conducted a retrospective review of patients with SSc at a tertiary referral center. Patients with documented onset of RP during childhood or adolescence were included. Clinical, demographic, serological, and capillaroscopic data were extracted.
**Results**
Among 167 patients with SSc, seven patients, all female, reported onset of RP during childhood or early adolescence (age range: 11–16 years). The age at SSc diagnosis ranged from 12 to 47 years. In comparison with adult-onset RP, pediatric-onset cases demonstrated a longer mean interval between RP onset and SSc diagnosis (9.4 vs. 3.37 years), although marked variability was observed in both groups. In two patients, diagnosis was established concurrently with RP, while in three patients the SSc was diagnosed 2–5 years later. Notably, two patients demonstrated markedly prolonged latency, reaching 23 and 34 years. 5/7 patients had

**Table 1 (Abstract P343)** Characteristics of patients with pediatric-onset Raynaud's phenomenon preceding systemic sclerosis

| Patient (all female) | RP onset (years) | SSc onset (years) | SSc type | Skin | GI | ILD | Cardiac | ANA titer - SSc related abs | Capillaroscopy |
|---|---|---|---|---|---|---|---|---|---|
| 1 | 11 | 34 | dSSc | Telangiectasias, ulcers | Upper | + | - | 1/640 - Scl70 | Active |
| 2 | 15 | 17 | dSSc | - | - | - | - | 1/1280 - Scl70 | Active |
| 3 | 15 | 15 | lSSc | Telangiectasias, calcinosis | - | - | - | NR | NR |
| 4 | 13 | 47 | lSSc | Telangiectasias, ulcers | Upper | - | - | 1/640 - Scl70 | NR |
| 5 | 16 | 21 | lSSc | Telangiectasias, ulcers | - | + | PAH, Pericarditis | 1/1280 - ACA, anti-RNP | Active |
| 6 | 12 | 12 | lSSc | Ulcers | Upper | - | - | 1/320 - anti-Ro | NR |
| 7 | 15 | 17 | lSSc | - | - | - | - | 1 | |

Abs, antibodies; ACA, anti-centromere antibodies; ANA, Antinuclear antibodies; dSSc, diffuse SSc; GI, Gastrointestinal; ILD, Interstitial Lung Disease; lSSc, limited SSc; NR, Not Reported; PAH, Pulmonary Arterial Hypertension; RNP, RNA polymerase; RP, Raynaud's phenomenon; SSc, Systemic Sclerosis

limited cutaneous SSc (lSSc), whereas two had diffuse (dSSc). Autoantibody data was available for six patients, all of whom were ANA positive. Anti-Scl-70 antibodies were detected in 3/7 patients. A single patient had both anti-centromere and anti-RNP antibodies, while another patient tested positive for anti-Ro antibodies. Nailfold capillaroscopy was performed in 4 patients with active pattern. Specific organ involvement, serological, and capillaroscopic characteristics are summarized in Table 1.

**Conclusion**

Pediatric-onset RP may precede the diagnosis of SSc by several years or even decades. The predominance of vascular and cutaneous manifestations, along with abnormal capillaroscopic findings and autoantibody positivity, supports the concept of an early microangiopathic process preceding overt systemic disease. These findings highlight the need for increased awareness and careful evaluation of RP in pediatric populations, as well as the potential role of early screening strategies to identify patients at risk for developing SSc.

**Disclosure of interest**

None declared.

## P344

### Juvenile systemic sclerosis in two eras: clinical, immunological and genetic insights from a single centre

M. K. Osminina[1], V. A. Podzolkova[1,*], O. V. Lazareva-Batyreva[2], Y. O. Kostina[2], M. Y. Kuzina[2], E. S. Zhuravleva[2], T. V. Belyaeva[3], N. S. Podchernyaeva[1]
[1]Children's Diseases, Sechenov First Moscow State Medical University; [2]Sechenov Center of Maternity and Childhood; [3]Sechenov First Moscow State Medical University, Moscow, Russian Federation
**Correspondence:** V. A. Podzolkova
*Pediatric Rheumatology 2026*, **24(S1):**P344

**Introduction**

Juvenile systemic sclerosis (JSSc) is a rare rheumatologic condition with a poor prognosis, and data on its clinical, immunological and genetic characteristics in paediatric patients remain limited. Well-characterised single-centre studies excluding overlap syndrome can provide valuable insights into the disease spectrum. Further detailed analysis is needed to improve early recognition and management.

**Objectives**

To compare two groups of JSSc patients followed in the same centre in different eras: before the widespread use of immunosuppression (1968–1980) and during the era of active combination therapy (2004–2024), and to characterise their clinical, immunological and genetic features, as well as survival.

**Methods**

A retrospective study was conducted on 78 JSSc patients (PRES/ACR/EULAR criteria) treated at Sechenov University. Group 1 (archival) – 38 children treated in 1968–1980; Group 2 (modern) – 40 children followed in 2004–2024. Patients with overlap syndrome were excluded. Genotyping of MMP1 (rs1799750), MMP9 (rs3918242) and NOS3 (rs1799983) was performed in a prospective subgroup of 21 patients (19 diffuse, 2 limited). Statistical analysis included $\chi^2$ tests, odds ratio calculation and logistic regression.

**Results**

Girls predominated in both groups (1:9 modern, 1:5.3 archival). Mean age at disease onset was 8.3 years. The diffuse cutaneous subset was significantly more common in the modern group (82.5% vs. 50%, $p$=0.002), yet the J4S severity score was lower (median 13.5 vs. 20, $p$=0.001), and new visceral involvement over time was less frequent (5% vs. 21%, $p$=0.034). Gastrointestinal (GI) involvement reached 82.5% in the modern group (strictures in 20%). Interstitial lung disease (ILD) was seen in 35% of the modern group and 28.9% of the archival group; pulmonary arterial hypertension occurred only in the archival group (5.3%). The autoantibody profile was stable: ANA 81%, ATA 31.4%, ACA 8.5%. In the archival group, only 38% received short-course oral corticosteroids and 24% received penicillamine (mean 3 months); 13% received only local therapy. In the modern group, all received combination immunosuppression; 13 received biologics. The J4S score increased after 12 months in the archival group despite therapy. Five-year survival in the modern group was 100% (92% in the archival group over the same period); overall, three deaths (7.5%) occurred in the modern group at late time points (74 and 125 months) due to multiorgan failure secondary to ILD and pancarditis. In the genetic substudy (21 patients), no significant associations were found for MMP1 or MMP9 polymorphisms. A trend toward association with JSSc was observed for the NOS3 (rs1799983) GG genotype (OR=2.61, 95% CI: 0.92–7.37, $p$=0.065), which was also associated with significantly lower serum levels of anti-collagen IV antibodies ($p$=0.039).

**Conclusion**

Our clinical and immunological data largely correlate with multicentre studies. The higher frequency of visceral involvement in the modern group, especially GI (82.5%), likely reflects improved diagnostics, whereas skin, joint and Raynaud's manifestations were similar across eras. Modern immunosuppression achieves 100% 5-year survival even in severe diffuse JSSc, suggesting therapeutic pathomorphosis, although late mortality persists. The NOS3 GG genotype may predispose to JSSc and ILD.

**Disclosure of interest**

None declared.

## P345

### From oral ulcers to complete phenotype - longitudinal evolution of pediatric-onset BehÇet's disease in a Brazilian tertiary center

J. P. Lima[1], V. Curi[1], E. R. Ferreira de Araújo Santos[2], N. Batista[2], A. W. Silva de Souza[1], M. T. Terreri[2], G. Clemente[2,*]
[1]Medicine; [2]Pediatrics, Universidade Federal de São Paulo, Sao Paulo, Brazil
**Correspondence:** G. Clemente
*Pediatric Rheumatology 2026*, **24(S1):**P345

**Introduction**

Pediatric-onset behçet's disease (p-BD) is uncommon, has highly variable clinical manifestations, and may take years to evolve from initial symptoms to a complete phenotype. Together, these features make early recognition challenging and may delay diagnosis. Description of the phenotype evolution of p-BD remains limited, particularly in non-endemic regions such as Latin-America.

**Objectives**

To characterize the phenotype and classification evolution of p-BD in a Brazilian tertiary referral center.

**Methods**
We conducted a retrospective cohort study of patients with p-BD followed over a 25-year period at a pediatric vasculitis clinic of a referral center in Sao Paulo, Brazil. Medical records were reviewed to assess baseline and follow-up features and classification status over time according to the Pediatric Behçet's Disease (PEDBD) criteria [1].
**Results**
A total of 20 patients with p-BD were included, with 70% female. Mean age at symptom onset was 8.9 (±4.4) years, with a mean diagnostic delay of 4.5 (±3.4) years. In 55% of patients, a prior alternative diagnosis had been made before the recognition of BD. At diagnosis, recurrent oral ulcers were present in all patients, and genital ulcers, in 65%. Other disease domains were less frequent at clinical diagnosis, including musculoskeletal, skin, ocular, vascular, and neurological involvement. Only 20% of patients fulfilled complete PEDBD classification criteria at diagnosis, compared with 50% by the last visit. Among those who fulfilled criteria, the mean time from symptom onset to complete classification was 4.92 years. HLA-B51 was detected in 29% of patients, whereas pathergy test positivity occurred in 16.7%.
**Conclusion**
BD in children may take years to evolve from initial symptoms to a more characteristic phenotype, and a substantial proportion of patients do not complete the PEDBD criteria despite follow-up. Careful longitudinal follow-up is essential in children with BD-like manifestations, even before a complete phenotype is established.

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**Disclosure of interest**
None declared.

P346
**Isolated left ventricular cardiomyopathy and acute heart failure: a red flag for pediatric Takayasu arteritis**
I. Awan[1,*], C. Sundaramoorthy [2], A. Sridhar[2], K. Khawaja[2]
[1]Pediatric Rheumatology, Leicester Royal Infirmary Hospital, University Hospital Leicester UHL NHS Trust; [2]Paediatric Rheumatology, Leicester Royal Infirmary, University Hospital Leicester UHL NHS Trust, Leicester, UK
**Correspondence:** I. Awan
*Pediatric Rheumatology 2026*, **24(S1):**P346

**Introduction**
Takayasu Arteritis (TA) is a rare large-vessel vasculitis in children, often presenting non-specifically and leading to severe end-organ damage. Isolated left ventricular cardiomyopathy (LVCM) and acute heart failure (AHF) are rare, life-threatening initial presentations.
**Objectives**
Report a critical case of childhood TA presenting with AHF secondary to isolated LVCM, highlighting the role of timely immunosuppression in cardiac recovery.
**Methods**
Case Description: A 15.5-year-old female with ADHD presented with a 4-month history of breathlessness, orthopnea, and constitutional symptoms (loss of appetite, weight loss, hemoptysis). She was urgently admitted for AHF (pulmonary edema, hypertension, raised ProBNP). Urgent ECHO showed severely impaired left systolic function (LVEF ~20%) and isolated left ventricular dysfunction. Paradoxical hypertension exacerbation by ACE inhibitors and a blood pressure discrepancy were red flags, prompting MDT input. Initial autoimmune screening was normal. MRA confirmed a vasculitic etiology, revealing critical stenosis of the descending aorta, renal, coeliac, and superior mesenteric arteries. The TA diagnosis was formally confirmed using the EULAR/PRINTO/PRES criteria. Her vasculitis activity (PVAS 18) was significantly elevated. She was promptly initiated on pulse methylprednisolone, followed by oral prednisolone and subcutaneous methotrexate. This led to rapid clinical stabilization and significant recovery of myocardial contractility (LVEF 45%), characteristic of an inflammatory etiology. Following an allergic reaction to Rituximab, she transitioned to salvage therapy with Tocilizumab (TCZ) and Methotrexate (MTX). Her PVAS improved to 2, though Pediatric Vasculitis Damage Index (PVDI 3) remained elevated due to persistent renovascular stenosis. She is awaiting interventional radiology assessment; recent DMSA and PET scans are normal. Ongoing management involves complex hypertension control, virtual BP monitoring, and MDT coordination, including a 'Hospital Passport' for her ADHD.
**Conclusion**
Isolated LVCM is a severe, rare red flag for pediatric TA. Rapid LVEF recovery post-immunosuppression confirms the inflammatory nature of the dysfunction. Clinicians should apply standardized tools (PVAS and EULAR criteria) early to prevent permanent damage (PVDI). TCZ is an effective second-line agent for patients intolerant to initial biologics. Complex management requires continuous MDT input for therapeutic transitions and optimizing renovascular hypertension.

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**Disclosure of interest**
None declared.

P347
**Unusual association of subpleural cyst and pulmonary embolism in granulomatosis polyangiitis arteritis**
I. Awan[1,*], C. Sundaramoorthy[1], A. Sridhar[1], K. Khawaja[1]
[1]Paediatric Rheumatology, Leicester Royal Infirmary, University Hospital Leicester UHL NHS Trust, Leicester, UK
**Correspondence:** Irum Awan
*Pediatric Rheumatology 2026*, **24(S1):**P347

**Introduction**
Pediatric ANCA-associated vasculitis (AAV) is a rare condition that often presents non-specifically, delaying diagnosis (1-3). In children, AAV frequently follows an aggressive course, requiring urgent multi-disciplinary intervention (2).
**Objectives**
To report an atypical presentation and management of pediatric AAV with life-threatening pulmonary hemorrhage, focusing on critical subpleural events and thrombotic complications.
**Methods**
A comprehensive retrospective review of the patient's electronic medical records, clinical imaging, laboratory investigations and management was conducted.
**Methods/Results**
A 14-year-old male with neurodiversity presented with cough, hemoptysis, profound weight loss, myalgia, and critical pallor (HB 6.8 g/DL). His condition rapidly progressed to life-threatening pulmonary haemorrhage requiring four weeks of mechanical ventilation and resulted in pulmonary-renal syndrome. Empirical immunosuppression was initiated. Granulomatosis with polyangiitis (GPA) was confirmed by high p-ANCA titre (PR3 523) with PVAS score 19/63 (4). His condition was complicated by secondary macrophage activation syndrome (MAS). Post-ICU, severe chest pain led to a CT confirming tension pneumothorax and pulmonary embolism, managed with drainage and anticoagulation. Recurrent pneumothorax required surgical pleurodesis. CT also revealed an unusual right subpleural cyst. Management involved rituximab, cyclophosphamide 6mo, and anakinra. He stabilised with normalised p-ANCA titres (PVAS 0/63) and was weaned from ventilation. Early Multidisciplinary MDT input, including psychological support, facilitated his transition to rehabilitation, addressing neurodiversity challenges. Currently, he is home-schooled, stable (Pediatric Vasculitis damage Index PVDI 1/72) having regular Rheumatologist & MDT follow up.
**Conclusion**
This case highlights three key points in pediatric GPA: 1) Profound pallor and hemoptysis signal life-threatening pulmonary haemorrhage, requiring urgent empirical immunosuppression.2) Critical GPA requires continuous monitoring for unusual pulmonary complications such as recurrent pneumothorax, pulmonary embolism, and rare subpleural cysts. 3) Early, continuous MDT involvement is essential for holistic re-

covery, particularly when addressing neurodiversity through patient-empowered communication.

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**Disclosure of interest**
None declared.

## P348

### Comparative performance of prognostic scoring systems in pediatric glomerulonephritis with crescents


I. Stojkic[1,*], S. E. Wendefer[2], J. Gaut[3], L. Biederman[4], W. E. Smoyer[1], G. Hidalgo[5]
[1]Nationwide Children's Hosptial, Columbus, USA; [2]University of British Columbia, Vancouver, Canada; [3]Washington University in Saint Louis, Saint Louis; [4]Cincinnati Children's Hosptial, Cincinnati; [5]Hackensack Meridian Health, Edison, USA
**Correspondence:** I. Stojkic

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**Introduction**
Predicting progression to kidney failure in glomerulonephritis with crescents (CGN) is a critical clinical goal. Pediatric CrGN differs from adult disease, yet established prognostic scoring systems have not been well validated in children. We compared the performance of four scoring systems and evaluated the ANCA Kidney Risk Score (AKRiS) in a large, multi-center, heterogeneous, pediatric cohort.
**Objectives**
We compared the performance of four scoring systems and evaluated the ANCA Kidney Risk Score (AKRiS) in a large, multi-center, heterogeneous, pediatric cohort.
**Methods**
We conducted a retrospective study using the Pediatric Nephrology Research Consortium cohort. We included children with GN, ≥2 glomerular crescents on biopsy and ≥12 months of follow-up. The primary outcome was kidney failure at 12 months. Four scoring systems were evaluated: Berden classification, Percentage of ANCA Crescentic Score (PACS), ANCA Renal Risk Score (ARRS), and AKRiS. Discrimination was assessed using area under the receiver operating characteristic curve (AUC). Subgroup analyses examined performance of the ARRS and AKRiS in ANCA-associated/pauci-immune GN and IgA nephropathy.
**Results**
Among 355 patients (median age 14 years, 65% female), etiologies included IgA nephropathy (20%), ANCA-associated GN (14%), and other CGN (67%). Kidney failure occurred in 57 patients (16%) at 12 months. In the overall cohort, ARRS and AKRiS demonstrated the highest discrimination (AUC 0.90 each), followed by PACS (0.89) and Berden (0.86). In ANCA-associated/pauci-immune GN, AKRiS outperformed ARRS (AUC 0.97 vs. 0.91). In IgA nephropathy, performance was lower (AKRiS AUC 0.80; ARRS AUC 0.77).
**Conclusion**
In pediatric GN with crescents, ARRS and AKRiS demonstrate excellent discrimination for predicting 1-year kidney failure and outperform Berden and PACS classifications. AKRiS provides incremental benefit in ANCA-associated/pauci-immune GN cases but not in the overall cohort. The ARRS and AKRiS demonstrated robust performance across ANCA and nonANCA etiologies, supporting its utility for risk stratification across multiple etiologies of CGN.
**Disclosure of interest**
None declared.

## P349

### Changes in nasopharyngeal microbiota composition in children with Kawasaki disease: a case-control study


J. Sánchez-Manubens[1], G. Gonzalez Comino[2], M. Blanco Fuertes[2], J. M. Mosquera Angarita[3], S. Carriquí Arenas[3], N. Sanz Marcos[4], C. Grasa[5], B. Mercader[6], N. Cahís[7], S. Corral Arboledas[8], S. Pie Raventós[9], N. López Segura[10], X. Rodó[11], C. Muñoz-Almagro[2], J. Anton Lopez[3,*]
[1]Pediatric Rheumatology, Parc Taulí Hospital Universitari. Institut d'Investigació i Innovació Parc Taulí (I3PT-CERCA), Sabadell; [2]Infectious Diseases and Microbiome Research Group; [3]Pediatric Rheumatology; [4]Pediatric Emergency, Hospital Universitari Sant Joan de Déu, Barcelona; [5]Pediatrics, Hospital Universitario La Paz, Madrid; [6]Pediatrics, Hospital Virgen de la Arrixaca, Murcia; [7]Pediatric Emergency, Parc Taulí Hospital Universitari. Institut d'Investigació i Innovació Parc Taulí (I3PT-CERCA), Sabadell; [8]Pediatrics, Hospital General de Granollers, Granollers; [9]Pediatrics, Hospital Universitari Josep Trueta, Girona; [10]Pediatrics, Hospital del Mar; [11]ISGlobal, Barcelona, Spain
**Correspondence:** J. Anton Lopez

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**Introduction**
Kawasaki disease (KD) is an acute pediatric vasculitis of unknown etiology, with increasing evidence suggesting host–microbiota interactions may contribute to disease pathogenesis. While gut microbiota alterations have been described, respiratory microbiota involvement remains largely unexplored.
**Objectives**
To characterize nasopharyngeal microbiota composition in children with KD compared with viral respiratory controls and healthy controls, while exploring associated epidemiological, clinical, and microbiological factors.
**Methods**
Nasopharyngeal samples from 233 children (45 KD, 84 viral controls, 104 healthy controls) underwent 16S rRNA gene sequencing targeting the V3–V4 region. Bioinformatic analyses using QIIME2 and R included quality control, taxonomic classification, alpha/beta diversity analyses, differential abundance testing, unsupervised clustering, and real-time PCR for Streptococcus pneumoniae carriage.
**Results**
KD patients demonstrated significantly altered nasopharyngeal microbiota composition compared with both control groups, with greater richness but lower evenness. Beta-diversity analyses confirmed clear compositional separation. KD samples showed increased Corynebacterium and Dolosigranulum abundance and reduced Streptococcus and Moraxella abundance. ASV45 (Corynebacterium propinquum) was strongly associated with KD. Streptococcus pneumoniae carriage was reduced in KD cases. PAM clustering identified seven microbial clusters, including one KD-enriched cluster characterized by high Dolosigranulum/Corynebacterium abundance and lower rates of vaginal delivery.
**Conclusion**
This study provides novel evidence that children with KD exhibit a distinct nasopharyngeal microbiota profile, supporting a potential role of airway microbial dysbiosis in KD pathogenesis. The association with Corynebacterium propinquum and identification of a KD-enriched microbial cluster suggest disease-specific microbial signatures and offer new insights into host–microbiota interactions in KD. Further multicenter studies are needed to validate these findings and clarify underlying mechanisms.

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**Disclosure of interest**
None declared.

## P350

### Diagnostic value of popliteal vein total wall thickness in pediatric Behçet disease

L. Koru[1,*], F. Haslak[1]
[1]Pediatric Rheumatology, Istanbul Medeniyet Univerctiy, Istanbul, Türkiye
**Correspondence:** L. Koru
*Pediatric Rheumatology 2026*, **24(S1):**P350

**Introduction**
Behçet disease (BD) is a systemic inflammatory disorder that shares features with autoinflammatory diseases and vasculitis. It has a broad clinical spectrum and most commonly presents with oral and genital ulcers, skin lesions, and ocular involvement. The characteristic distribution of cutaneous manifestations and the tendency to develop thrombosis in vessels of all sizes provide important diagnostic clues.[1,2] Recent studies have demonstrated increased venous wall thickness in the lower extremity veins of patients with BD, suggesting that this finding may reflect vascular inflammation and support the diagnosis.[3,4].
**Objectives**
In this context, the present study aimed to investigate the diagnostic significance of popliteal vein wall thickness in pediatric BD.
**Methods**
This observational cross-sectional study was conducted at our tertiary referral center between October 2025 and January 2026. Patients diagnosed with BD according to the Pediatric Behçet Disease (PEDBD) classification criteria and age- and sex-matched healthy controls were included. Patients fulfilling at least three PEDBD criteria were classified as complete BD, whereas those fulfilling two criteria were classified as incomplete BD. Popliteal vein total wall thickness was measured ultrasonographically from the posterior wall at the popliteal fossa in all participants. Multiple measurements were obtained for each individual, and mean values were used for analysis.
**Results**
The study included 39 patients with BD (13 complete and 26 incomplete) and 34 healthy controls. Total wall thickness of both popliteal veins was significantly higher in both complete and incomplete BD groups compared to healthy controls, whereas no significant difference was observed between complete and incomplete BD groups ($p$<0.001). Popliteal vein total wall thickness demonstrated good discriminatory performance for distinguishing BD from controls (right AUC=0.852; left AUC=0.871; $p$<0.001). The optimal cut-off value for both popliteal veins was 0.43 mm. At this threshold, sensitivity and specificity were 89.7% and 61.8% for the right popliteal vein, and 87.2% and 55.9% for the left popliteal vein, respectively.
**Conclusion**
Popliteal vein total wall thickness may serve as a supportive ultrasonographic finding in the diagnosis of pediatric BD.

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**Disclosure of interest**
None declared.

## P351

### Real-world urinalysis monitoring practices in childhood IgA vasculitis according to rash presentation: data from the JIR-CLiPS network

M. A. Beldie[1,2,*], M. Ganeva[3], D. Gorczyca[4], T. Giani[5], M. Jelusic[6], N. Kifer[6], J. Marčiulynaitė[7], M. Hofer[8], F. Hofer[8], M. Frank[8], A. Ramanauskienė[9], Y. Uziel[10], T. Kallinich[4,11], A. Šnipaitienė[7] on behalf of JIR-CLiPS network
[1]Pediatrics, "Grigore T. Popa" University of Medicine and Pharmacy, Iasi, Romania, [2]Centre for Translational Medicine, Semmelweis University, Budapest, Hungary, [3]Department of Paediatric Rheumatology, Medical University Sofia, Sofia, Bulgaria, [4]Charité - Universitätsmedizin Berlin, Corporate Member of Freie Universität Berlin and Humboldt-Universität, Berlin, Germany, [5]Department of Paediatrics, AOU Meyer IRCCS, Florence, Italy, [6]Department of Pediatrics, University Hospital Centre, School of Medicine University of Zagreb, Zagreb, Croatia, [7]Department of Paediatrics, Lithuanian University of Health Sciences, Kaunas, Lithuania, [8]Fondation Rhumatismes-Enfants-Suisse, Lausanne, Switzerland, [9]Gijos Clinics, Kaunas, Lithuania, [10]Department of Paediatrics, Meir Medical Centre, Kfar Saba, Gray School of Medicine, Tel Aviv University, Tel Aviv, Israel, [11]Deutsches Rheuma-Forschungszentrum, an institute of the Leibniz Association, Berlin, Germany
**Correspondence:** M. A. Beldie
*Pediatric Rheumatology 2026*, **24(S1):**P351

**Introduction**
Renal involvement is the major determinant of long-term outcome in IgA vasculitis (IgAV). Emerging evidence suggests that severe or atypical skin manifestations may be associated with an increased risk of nephritis development. Although current international guidelines predominantly focus on the management of established nephritis, evidence guiding the duration of urinalysis follow-up in patients without initial renal involvement remains unclear.
**Objectives**
This study aimed to evaluate real-world urinalysis follow-up practices in children with IgAV without initial renal involvement, focusing on the influence of rash phenotype on clinician decision-making, and to explore variations by clinician specialty, years of experience, and country.
**Methods**
An online English-language survey was distributed through the JIR-CliPS (Juvenile Inflammatory Rheumatism; Clinical Practice Strategies) Network, established within COST (European Cooperation in Science and Technology) action CA21168. The survey evaluated urinalysis follow-up duration in children with IgA vasculitis without intitial renal involvementacross four rash presentation scenarios: typical rash, atypical rash, diffusely distributed rash, and recurrent rash. Respondents selected one of five predefined follow-up duration categories (≤2, ≤4, ≤6, ≤12, or >12 months). Differences in follow-up recommendations across rash phenotypes were assessed using the Friedman test with post-hoc pairwise Wilcoxon signed-rank tests and Bonferroni correction. Interclinician variability was assessed using Shannon entropy.
**Results**
Between September 2022 and May 2026, 190 responses were collected from clinicians across 46 countries and 5 continents, predominantly paediatric rheumatologists (56.6%). Recommended urinalysis follow-up duration differed significantly across rash phenotypes (Friedman $\chi^2$=65.13, $p$<0.001). Typical skin rash was associated with the shortest follow-up duration (median 6 months, mean 8.7±4.8 months), whereas atypical, diffusely distributed, and recurrent rashes were each assigned a median follow-up duration of 12 months (means 10.9±5.4, 11.7±5.2, and 12.8±5.3 months, respectively). Post-hoc analysis confirmed significant differences between typical rash and all other rash phenotypes (all $p$<0.001), as well as between atypical and recurrent rash ($p$<0.001). Shannon entropy values were highest for atypical rash (1.980) and diffusely distributed rash (1.826). No significant differences in follow-up practices were observed according to specialty, years of experience, or country.
**Conclusion**
Urinalysis follow-up duration in IgAV patients without initial renal involvement varies significantly in real-world clinical practice, depending on rash type, with a longer follow-up for recurrent and atypical presentations. These findings support the need for studies on urinalysis follow-up by different rash phenotype in IgAV.
**Disclosure of interest**
None declared.

## P352

### Diagnosis, treatment and monitoring of pediatric Behçet's disease: systematic literature review informing the ISSAID/PRES recommendations

M. V. Mastrolia[1,2,*], M. Romano[3,4], D. Piskin[4,5], D. Rigante[6,7], D. Poddighe[8], S. Demir[9], E. Sag[10], N. Zitoun[11], N. Ughi[12], C. Acikel[13], S. Venkateswaran[14], J. Park[3], K. Deiva[15], I. Tugal-Tutkun[16], I. Koné-Paut[17], G. Simonini[1,2], E. Demirkaya[3,4,5]
[1]Meyer Children's Hospital IRCCS, Rheumatology Unit. ERN ReCONNET center, [2]University of Florence, Neurofarba Department, Firenze, Italy, [3]Department of Pediatrics, Schulich School of Medicine & Dentistry, University of Western Ontario, Division of Pediatric Rheumatology, [4]University of Western Ontario, Canadian Behcet's and Autoinflammatory Diseases Center, [5]Schulich School of Medicine & Dentistry, University of Western Ontario, Department of Epidemiology and Biostatistics, London, Canada, [6]Fondazione Policlinico Universitario A. Gemelli IRCCS, Department of Life Sciences and Public Health, [7]Università Cattolica Sacro Cuore, Roma, Italy, [8]VinUniversity, Vinhomes Ocean Park, Gia Lam, College of Health Sciences, Hanoi, Viet Nam, [9]Faculty of Medicine, Eskisehir Osmangazi University, Department of Pediatric Rheumatology, Eskisehir, [10]Faculty of Medicine, Hacettepe University, Department of Pediatric Rheumatology, Ankara, Türkiye, [11]London Health Sciences Centre Research Institute, London, Canada, [12]Multispecialist Medical Department, ASST Grande Ospedale Metropolitano Niguarda, Division of Rheumatology, Milano, Italy, [13]ClinCompetence Cologne GmbH, Head of Division of Biometry, Cologne, Germany, [14]Department of Pediatrics, Schulich School of Medicine & Dentistry, University of Western Ontario, Division of Neurology, London, Canada, [15]National Reference Center for Rare Neuro-Immune diseases of the Brain and Spinal cord (MIRCEM), UMR 1184, Faculty of Medicine Paris Saclay, le Kremlin Bicêtre, France and Institut Universitaire de France, Pediatric Neurology Department, AP-HP, Paris Saclay University Hospitals, Bicêtre Hospital, Paris, France, [16]Eye Hospital, Uveitis Service, Eye Protection Foundation, Bayrampasa, Instanbul, Türkiye, [17]Faculty of Medicine Paris Saclay, le Kremlin Bicêtre, France and FHU Inflamme, Pediatric rheumatology Department, AP-HP, Paris Saclay University Hospitals, Bicêtre Hospital, National Reference Center for autoinflammatory disease and inflammatory amyloidosis (CEREMAIA), Paris, France
**Correspondence:** M. V. Mastrolia
*Pediatric Rheumatology 2026*, **24(S1):**P352

**Introduction**
Pediatric-onset Behçet's disease (BD) accounts for up to 20% of cases and represents a distinct clinical entity characterized by evolving phenotypes and age-specific patterns of organ involvement. Diagnosis in children remains particularly challenging. Pediatric BD often begins with incomplete or atypical manifestations—most commonly recurrent oral ulcers—that may precede multisystem involvement by several years. Given these challenges, systematic evaluation of the available evidence is essential.
**Objectives**
This systematic literature review provides a comprehensive synthesis of current knowledge on the diagnosis, treatment, and monitoring of pediatric BD, aiming to inform forthcoming consensus recommendations by the International Society of Systemic Auto-Inflammatory Diseases (ISSAID) and the Pediatric Rheumatology European Society (PReS).
**Methods**
A systematic search was conducted according to PRISMA guidelines. Observational studies reporting clinical features, diagnostic criteria, management strategies, and outcomes in BD patients diagnosed before the age of 16 years were included. Proportional meta-analysis was performed to give pooled estimates for organ system involvement.
**Results**
Fifty-two studies, encompassing 2,929 patients, met inclusion criteria. Sex distribution was balanced, with a 1:1 male-to-female ratio. The age of onset differed, with 1 year old being the lowest median age of onset and 2 years the lowest median age of diagnosis. The pooled random-effects estimate demonstrated that 50% of patients were HLA-B51 positive (95% CI: 0.5–0.6). Mucocutaneous manifestations were nearly universal (97.8%) and frequently represented the initial feature (80.9%). Musculoskeletal (36%), ocular (35%), neurological (17.9%), gastrointestinal (20%), vascular (15.4%) manifestations showed substantial variability in prevalence. Therapeutic approaches varied widely and were extrapolated from adult practice, with treatment guided by organ involvement and severity.
**Conclusion**
Pediatric BD encompasses a heterogeneous spectrum of phenotypes requiring harmonized diagnostic frameworks, structured phenotypic stratification, and standardized monitoring to improve long-term outcomes. The relative burden and combination of organ manifestations varied across cohorts, reflecting both biological heterogeneity and differences in study design.
**Disclosure of interest**
None declared.

## P353

### When the fever is not long enough: a case report of incomplete Kawasaki disease complicated by giant coronary aneurysm

M. Olszewska[1,*], K. Peterschein[2], A. Wietecha-Pikul[1], P. Kwinta[1], A. Morka[2]
[1]Department of Paediatrics, [2]Department of Paediatric Cardiology, Jagiellonian University Medical College, Cracow, Poland
**Correspondence:** M. Olszewska
*Pediatric Rheumatology 2026*, **24(S1):**P353

**Introduction**
Diagnosis of Kawasaki disease (KD) can be challenging, particularly in infants. In this age group, the clinical picture may differ from the typical presentation, with fewer principal features, corresponding to incomplete KD [1]. However, even in infants, prolonged unexplained fever remains the key diagnostic symptom. Reports of KD with short-lasting, self-limiting fever are extremely rare [2].
**Objectives**
To report a case of incomplete KD with short-lasting fever, which was complicated by a giant coronary artery aneurysm (CAA).
**Methods**
Case report.
**Results**
A four-month-old boy was admitted to the hospital with incomplete KD. About three weeks earlier, he had presented with a two-day, mild fever, rash, erythema and cracking of the lips. Ten days later, he was referred to the local hospital because of abnormal laboratory test results, including C-reactive protein (CRP) of 67 mg/L, leukocytosis of $21\times10^9$/L, and thrombocytosis of $919\times10^9$/L. On admission, desquamation of fingers and toes and erythema of the lips were noted. During hospitalisation, further increases in inflammatory markers and thrombocytosis were observed. Echocardiography (ECHO) performed a week later revealed CAA. Treatment with intravenous immunoglobulin at 2 g/kg, methylprednisolone (mPSL) at 2 mg/kg and acetylsalicylic acid (ASA) at 4 mg/kg was initiated. The infant was referred to our hospital for further evaluation. On admission, physical examination did not reveal any new abnormalities. Laboratory tests showed a decrease in CRP to 10 mg/L, leukocytosis of $23\times10^9$/L and thrombocytosis of $1721\times10^9$/L. The control ECHO revealed a giant aneurysm of the right coronary artery (RCA) (Z score 11.6) and a medium aneurysm of the left main coronary artery (LMCA) (Z score 6.5). In addition to mPSL and ASA, unfractionated heparin was administered. Two weeks later, a gradual decrease in CA size was observed, with Z scores of 5.53 for the RCA and 3.09 for the LMCA. The boy was discharged on ASA and warfarin treatment. After three months of follow-up, normal LMCA and a further decrease in RCA size were found.
**Conclusion**
Patients with incomplete KD, who have short-lasting fever, are at risk of delayed diagnosis and the development of CAA because they do not meet current diagnostic criteria [1]. ECHO should be performed in infants with the cardinal features of KD, even if the fever duration is not typical for KD, as timely treatment reduces the risk of CAA and associated cardiovascular complications in future.

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**Trial registration identifying number**.
Not applicable.
**Disclosure of interest**
None declared.

## P354

### Unusual pathway and systemic presentation in IGA vasculitis in a child with undifferentiated inflammatory Bowel disease

I. Awan[1,*], C. Sundaramoorthy [1], A. Sridhar[1], K. Khawaja[1]
[1]Paediatric Rheumatology, Leicester Royal Infirmary, University Hospital Leicester UHL NHS Trust, Leicester, UK
**Correspondence:** I. Awan
*Pediatric Rheumatology 2026*, **24(S1):**P354

**Introduction**
Pediatric patients with autoimmune conditions (e.g. IBD) and non-specific musculoskeletal (MSK) pain need vigilant follow-up (1). Atypical presentations or drug complications can mask systemic vasculitis. Critical multidisciplinary team (MDT) collaboration is essential for timely, life-saving diagnosis.
**Objectives**
This case details the diagnostic concundrum and MDT management of an undifferentiated IBD and chronic MSK pain. We demonstrate an unusual pulmonary-first sequence/features that masked the correct diagnosis of underlying vasculitis.
**Methods**
Case Presentation: A 14-year-old with undifferentiated IBD and chronic MSK pain was admitted for Infliximab. He developed acute, severe shortness of breath, managed as presumed Infliximab pneumonitis (high-dose IV steroids, antibiotics, respiratory support). Bronchopulmonary lavage and lung biopsy were normal. He was discharged on mask oxygen, and Rheumatology input was initiated. Acute admission workup showed no proteinuria and a mild ANCA PR3 titer of 40 with Pediatric Vasculitis Activity Score (PVAS) 1/63. Three months later, a purpuric rash developed, followed by significant proteinuria (PCR 318.6 mg/ml). This pulmonary-skin-renal sequence was critical. Renal biopsy confirmed aggressive IgA vasculitis nephritis (IgAVN), ISKDC Grade 3 A with 22% crescents, yielding PVAS 15/63. The MDT initiated intensive immunosuppression (oral Prednisone 40 mg daily x 28 days, MMF, ACEi). Infliximab was transitioned to Vedolizumab for IBD. Currently, he has regular MDT follow-up, stable on MMF/Vedolizumab, with normalized creatinine (40 µmol/L) and albumin-to-creatinine ratio (2.9 mg/mmol).
**Conclusion**
This case shows IgAVN can present with an unusual pulmonary-first sequence, creating a major diagnostic conundrum (IgA vasculitis vs. Infliximab pneumonitis). Aggressive renal involvement (22% crescents) highlights the need for diagnostic accuracy. The case underscores the critical necessity of continuous follow-up, serial proteinuria monitoring, and timely MDT involvement to ensure life-preserving therapy.
**Keywords**
IgA vasculitis nephritis (IgAVN), Undifferentiated Inflammatory Bowel Disease (IBD), Infliximab pneumonitis, Multidisciplinary team (MDT), Proteinuria monitoring, Systemic vasculitis.

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**Disclosure of interest**
None declared.

## P355

### Global treatment patterns in IGA vasculitis: data from the JIR-CLIPS network

N. Kifer[1,*], D. Kifer[2], M. Ganeva[3], A. Snipaitiene[4], J. Uziel[5,6], V. Hentgen[7], C. Lanna[8], S. Sahin[9], F. Hofer[10], M. Hofer[11], T. Giani[12], M. Jelusic[1], D. Gorczyca[13], T. Kallinich[13,14] on behalf of JIR-CLiPS network
[1]Department of Pediatrics, University Hospital Centre Zagreb, School of Medicine University of Zagreb, [2]Department of Biophysics, University of Zagreb Faculty of Pharmacy and Biochemistry, Zagreb, Croatia, [3]Department of Pediatric Rheumatology, Medical University Sofia, Sofia, Bulgaria, [4]Department of Pediatrics, Lithuanian University of Health Sciences, Kaunas, Lithuania, [5]Department of Pediatrics, Meir Medical Center, Kfar-Saba, [6]Tel Aviv University School of Medicine, Tel Aviv, Israel, [7]Paediatric Rheumatology, Versailles Hospital, Versailles, France, [8]Department of Locomotor System, School of Medicine, Universidade Federal de Minas Gerais, Belo Horizonte, Brazil, [9]Department of Pediatric Rheumatology, Istanbul University Cerrahpasa, Istanbul, Türkiye, [10]Fondation Rhumatismes-Enfants-Suisse, Lausanne, [11]Department of Pediatrics, Hôpital Riviera-Chablais, Rennaz, Switzerland, [12]Department of Pediatrics, AOU Meyer IRCCS, Florence, Italy, [13]Charité Universitätsmedizin Berlin, [14]Deutsches Rheuma-Forschungszentrum, an institute of the Leibniz Association, Berlin, Germany
**Correspondence:** N. Kifer
*Pediatric Rheumatology 2026*, **24(S1):**P355

**Introduction**
Management of IgA vasculitis (IgAV) is often guided by the predominantly or most severely affected organ system, resulting in variable treatment strategies.
**Objectives**
This study aimed to characterise real-world treatment approaches for different clinical manifestations of IgA vasculitis and to explore geographic variation in therapeutic strategies.
**Methods**
An online English-language survey assessed therapeutic approaches to IgAV across eight clinical scenarios: typical rash (< or > 4 weeks), atypical rash, recurrent rash, severe limitation due to musculoskeletal involvement (< or > 4 weeks), and severe abdominal symptoms (< or > 4 weeks). Disease severity was physician-assessed. Respondents selected up to three treatment options per scenario, including nonsteroidal anti-inflammatory drugs (NSAIDs), disease-modifying antirheumatic drugs (DMARDs), different glucocorticoid (GC) regimen, colchicine, dapsone and others. Responders were grouped according to similarities in treatment selection using hierarchical cluster (Ward's D linkage method, discordant response distances). Clusters were further analysed according to neonatal mortality rate categories (0–5, $n$=132; 5–10, $n$=30; ≥10, $n$=17), by countries with highest participation (Turkey $n$=26, France $n$=20, Brazil $n$=16, Germany $n$=15) and by physicians' experience. The project is funded as a COST (European Cooperation for Science and Technology) action CA21168.
**Results**
Between September 2022 and January 2026, 185 responses from countries across five continents were collected from physicians. Treatment strategies differed geographically in recurrent rash scenario: cluster combining dapsone with low-dose GC included more participants from Germany ($p$=0.025), whereas cluster combining colchicine and low-dose GC included more participants from Turkey ($p$=0.022). Independently of clustering, colchicine use for recurrent skin rash was more frequent in Turkey and less frequent in Germany (both $p$=0.005). Geographic variation was observed for gastrointestinal (GI) treatment strategies, with Brazilian participants more frequently used GC pulses in GI involvement <4 weeks ($p$=0.001), while low-dose GC use in GI involvement >4 weeks was more common in France ($p$=0.019) and Germany ($p$=0.019) and less common in Turkey ($p$=0.003). Treatment of severe musculoskeletal involvement did not differ significantly. Differences in neonatal mortality rates, as a surrogate for healthcare status, were mainly observed in early typical rash.
**Conclusion**
Considerable international heterogeneity exists in IgAV management. The greatest variability was observed in the treatment of recurrent cutaneous and severe GI manifestations, whereas treatment of musculoskeletal involvement was more consistent across geographic regions. This variability likely reflects areas of limited evidence and the complexity of decision-making across different clinical and real-life geographic settings.
**Disclosure of interest**
None declared.

## P357

### Myocarditis and interstitial lung involvement in a 13-year old girl with eosinophilic granulomatosis with polyangiitis - case report

P. Kunstelj[1,*], S. Ilovar Strehar[2], S. Šetina Šmid[3], R. Zbačnik[4], M. Praprotnik[3], N. Toplak[1,5]
[1]Department of Allergology, Rheumatology and Clinical Immunology, [2]Department of Cardiology, [3]Department of Pulmonology, University Children's Hospital, University Medical Centre Ljubljana, [4]Department of Radiology, University Medical Centre Ljubljana, [5]Faculty of Medicine, Faculty of Medicine, University of Ljubljana, Ljubljana, Slovenia
**Correspondence:** P. Kunstelj
*Pediatric Rheumatology 2026*, **24(S1):**P357

**Introduction**
Eosinophilic granulomatosis with polyangiitis (EGPA) is an idiopathic necrotizing small-vessel vasculitis characterized by asthma, marked eosinophilia and eosinophilic inflammation (1,2). It is extremely rare among children. In the French national pediatric database for rare lung diseases, 14 cases were recorded between 1980 and 2012 (3). The clinical presentation may include cardiac, pulmonary, neurological, cutaneous, gastrointestinal and ENT involvement. Cardiac involvement is more common in ANCA negative patients (1,3).
**Objectives**
We aim to report a rare case of myocarditis and interstitial lung involvement in a pediatric patient with severe ANCA-negative EGPA.
**Methods**
Clinical data were obtained from our hospital's electronic medical record system.
**Results**
a 13-year old girl was admitted to the peditric rheumatology ward due to vasculitic rash, joint pain and a history of asthma. Laboratory evaluation revealed moderately elevated inflammatory markers, marked eosinophilia, and substantially elevated troponin levels accompanied by ECG changes. Chest X-ray demonstrated pulmonary infiltrates. Cardiac MR confirmed myocarditis. Cardiac function remained preserved. HRCT revealed interstitial lung involvement. The patient received intensive antiinflammatory therapy with intravenous immunoglobulins, high-dose methylprednisolone, cyclophosphamide and anti-IL5 receptor agent benralizumab, along with supportive care. Eosinophil counts and troponin levels declined early after initiation of intensive antiinflammatory treatment, followed by gradual improvement of symptoms and pulmonary function.
**Conclusions**
in patients with asthma, marked eosinophilia and vasculitic rash, EGPA should be strongly suspected. Evaluation for cardiac and pulmonary involvement and prompt intensive anti-inflammatory therapy are essential (1,2).

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**Disclosure of interest**
None declared.

## P358

### Clinical profile and outcome of patients with kawasaki disease complicated by giant coronary aneurysm: a long term followup study from tertiary care centre in North India

S. Sarkar[1,*], R. K. Pilania[1], D. Desai[1], Y. Sharma[1], P. Vignesh[1], D. Suri[1], M. Singhal[2], A. rawat[1], A. Gupta[1], S. Singh[1]
[1]Paediatrics, [2]Radiodiagnosis and Imaging, PGIMER Chandigarh, Chandigarh, India
**Correspondence:** S. Sarkar
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**Introduction**
Kawasaki Disease–associated giant coronary artery aneurysms (GCAA) represent the most severe form of coronary involvement in children. However, long term follow-up patterns and predictors of persistent aneurysm remain incompletely characterized.
**Objectives**
Clinical profile and demographic characteristics of children with Giant coronary aneurysm in Kawasaki Disease. Predictors of severe coronary involvement in Kawasaki Disease. Follow-up and outcome of the children with giant coronary artery aneurysm in Kawasaki disease. To evaluate coronary artery specific regression and time to regression analysis.
**Methods**
This retrospective cohort included 50 children with giant coronary artery aneurysms (absolute diameter ≥8 mm and/or Z-score ≥10) secondary to Kawasaki Disease managed between 2010 and 2025. Mean follow-up duration was 5.8 years. Echocardiographic and CT coronary angiography data were analyzed to evaluate coronary remodeling, vessel-specific regression kinetics, and predictors of persistent aneurysms using comparative statistics, Kaplan–Meier analysis, odds ratios, and ROC curve analysis.
**Results**
Fifty children ($n$=50) with giant coronary artery aneurysms (GCAA) secondary to Kawasaki Disease were included. Mean age at diagnosis was 27 months (range 2 months–13 years), with $n$=12/50 (24%) aged <1 year; $n$=36/50 (72%) were males. Complete and incomplete KD was seen in $n$=22/50 (44%) and $n$=28/50 (56%) respectively. Median duration to intravenous immunoglobulin (IVIG) administration was 13.5 days, while IVIG resistance occurred in $n$=8/50 (16%). Mean CRP was 46.3 mg/dL (range 8–198), ESR 64.4 mm/hr (range 22–132), platelet count $608\times10^9$/L (range 210–1240), and NT-proBNP 3480 pg/mL (range 420–35,000).Coronary involvement was predominantly multivessel with RCA involvement in $n$=38/50 (76%), LAD in $n$=36/50 (72%), LMCA in $n$=10/50 (20%), and LCX in $n$=2/50 (4%). Bilateral and pan-coronary disease occurred in $n$=31/50 (62%) and $n$=11/50 (22%) respectively. Regression occurred in $n$=28/50 (56%), while persistent giant aneurysms/non-regression occurred in $n$=20/50 (40%). Mean time to regression was 302.6 days. LAD aneurysms regressed earlier than RCA aneurysms [248 vs. 412 days; $p$=0.021], with lower cumulative regression probability in RCA aneurysms (log-rank $p$=0.034). RCA involvement predicted persistent aneurysms (OR 2.84, $p$=0.047). RCA diameter >10.2 mm and Z-score >18.4 best predicted persistence (AUC 0.78 and 0.81 respectively). Thrombus, calcification, myocardial ischemia, and stenotic lesions occurred in $n$=7/50 (14%), $n$=5/50 (10%), $n$=3/50 (6%), and $n$=2/50 (4%) respectively.
**Conclusion**
RCA aneurysms exhibit delayed regression and increased risk of persistent aneurysm. GCAA are an important complication of KD and should be managed aggressively to prevent complications.
**Disclosure of interest**
None declared.

P359

**Predictors of coronary artery abnormalities in children with Kawasaki disease: a single-center retrospective study**

S. Kurbanova[1,*], S. Valieva[1]
[1]Morozov City Children's Clinical Hospital, Moscow, Russian Federation
**Correspondence:** S. Kurbanova
*Pediatric Rheumatology 2026*, **24(S1):**P359

**Introduction**
Coronary artery abnormalities (CAA) remain the most severe complication of Kawasaki disease (KD). Early identification of patients at risk for coronary involvement is essential for timely intensification of anti-inflammatory therapy.
**Objectives**
To analyze clinical and laboratory predictors of coronary artery abnormalities in children with Kawasaki disease.
**Methods**
A retrospective single-center study included 400 children diagnosed with Kawasaki disease between 2014 and 2024. Coronary artery abnormalities included transient coronary dilatation and coronary aneurysms detected during the acute phase according to echocardiographic assessment. Morozov Children's City Clinical Hospital is a tertiary referral center receiving severe and diagnostically challenging Kawasaki disease cases from different regions of the Russian Federation. Statistical analysis was performed using StatTech v.4.12.7. Quantitative variables were compared using Mann–Whitney U test, categorical variables using Fisher's exact test. $P<0.05$ was considered statistically significant.
**Results**
Coronary artery abnormalities were observed in 196 patients (49.0%). Patients with coronary involvement were diagnosed significantly later compared to children without coronary abnormalities (median 9.5 [IQR 7.0–11.8] vs. 7.0 [6.0–9.0] days, $p<0.001$). Accordingly, intravenous immunoglobulin (IVIG) administration was initiated later in the coronary group (median 9.0 [7.0–12.0] vs. 7.0 [6.0–9.0] days, $p<0.001$). Patients with coronary abnormalities tended to be younger, although the difference did not reach statistical significance (median age 22.5 [10.0–40.8] vs. 27.0 [12.0–44.0] months, $p=0.215$). Incomplete Kawasaki disease was not associated with increased coronary risk (28.6% vs. 27.5%, $p=0.607$). Among clinical manifestations, oral mucosal changes were significantly less frequent in patients with coronary involvement (75.8% vs. 87.6%, $p=0.003$). Patients with coronary abnormalities demonstrated significantly higher platelet counts during disease course (median $677\times10^9$/L [552–819] vs. $615\times10^9$/L [509–751], $p=0.005$), while CRP, ferritin, hemoglobin and albumin levels did not differ significantly between groups ($p>0.05$). IVIG resistance was markedly more frequent among patients with coronary abnormalities (35.9% vs. 5.9%, $p<0.001$). Consequently, these patients more often required intensified anti-inflammatory treatment, including repeated IVIG infusions and biologic therapy (12.0% vs. 0.5%, $p<0.001$).
**Conclusion**
Delayed diagnosis and delayed IVIG administration were strongly associated with coronary artery abnormalities in Kawasaki disease. IVIG resistance and thrombocytosis were significantly more frequent in patients with coronary involvement. Absence of prominent mucosal manifestations may contribute to delayed recognition of Kawasaki disease and increased coronary risk. The high proportion of coronary abnormalities in our cohort may additionally reflect referral bias due to the tertiary-center setting and admission of severe Kawasaki disease cases from multiple regions of the country.
**Disclosure of interest**
None declared.

P360

**Functional evidence of circulating endotheliotoxicity in refractory Kawasaki disease complicated by giant coronary aneurysms and myocardial infarction**

S. Kurbanova[1,*], A. Khapchaev[2], V. Shirinsky[2], S. Valieva[1]
[1]Morozov City Children's Clinical Hospital, [2]National Medical Research Center of Cardiology Named after Academician E.I. Chazov, Moscow, Russian Federation
**Correspondence:** S. Kurbanova
*Pediatric Rheumatology 2026*, **24(S1):**P360

**Introduction**
Kawasaki disease is a systemic vasculitis of childhood predominantly affecting the coronary arteries. Despite significant advances in understanding the disease, the mechanisms underlying severe endothelial injury and the formation of giant coronary aneurysms remain insufficiently understood.
**Objectives**
To evaluate the effects of patient plasma on the morphofunctional state of the endothelial monolayer in vitro in refractory Kawasaki disease complicated by giant coronary aneurysms, intracoronary thrombosis, and myocardial infarction, as well as to assess the association between endothelial toxic activity and the severity and clinical phase of the disease.
**Methods**
A retrospective analysis of clinical, laboratory, and instrumental data was performed in a 2-year-old patient with severe Kawasaki disease. Endothelial barrier function was assessed using cultured human umbilical vein endothelial cells (HUVECs) by measuring monolayer electrical impedance with the ECIS-z system (Applied BioPhysics, USA). Morphological assessment of the endothelial monolayer was performed using phase-contrast video microscopy.Patient serum samples obtained at different stages of the disease (days 34, 50, and 62 of illness) were analyzed, along with plasma samples from patients with Kawasaki disease without coronary artery involvement and from patients with febrile infectious illnesses.
**Results**
The diagnosis of Kawasaki disease was established only on day 24 of illness after detection of coronary artery involvement. Despite treatment with intravenous immunoglobulin, glucocorticosteroids, and etanercept, Infliximab progression of coronary vasculitis was observed, leading to the formation of a giant aneurysm of the left anterior descending coronary artery up to 18 mm, right coronary artery aneurysm, and intracoronary thrombosis. **On day 50 of illness, the patient developed myocardial infarction associated with acute coronary syndrome, elevated cardiac biomarkers, and ischemic ECG changes.** Addition of 10% patient serum to HUVEC culture induced pronounced endothelial injury characterized by progressive disruption of endothelial barrier function (baseline impedance 15,000 Ohm vs. 6,000 Ohm after 69 h, $p<0.05$), loss of intercellular contacts, and endothelial cell death **(initially 100% confluent HUVEC monolayer, decreasing to 40% confluence after 72 h)**. Maximal endotheliotoxic activity was observed in serum obtained during the myocardial infarction phase and temporally coincided with the most severe coronary artery involvement. Removal of patient serum from the culture medium before the development of massive cell death resulted in restoration of endothelial barrier integrity **(12,000 Ohm at 62 h after plasma removal)**, suggesting potentially reversible early endothelial injury. During clinical improvement and stabilization of the patient's condition, a marked reduction in serum endotheliotoxicity was observed. Sera from other patients with Kawasaki disease without coronary aneurysm formation and from patients with febrile infectious illnesses did not demonstrate damaging effects on HUVECs.
**Conclusion**
.This clinical case demonstrates marked endotheliotoxic activity of patient plasma in refractory Kawasaki disease associated with the development of catastrophic coronary complications. Functional endothelial models may represent a promising tool for investigating the pathogenesis of severe forms of Kawasaki disease, identifying biomarkers of unfavorable disease course, and developing novel targeted therapeutic strategies.
**Disclosure of interest**
None declared.

P361

**Tuberculosis-associated Erythema induratum of Bazin coexisting with cutaneous polyarteritis nodosa: a case report**

S. Turkucar[1,*], S. N. Tiğrak[1], G. Oguz[1], E. Comut[2], N. Calli Demirkan[2]
[1]Pediatric Rheumatology; [2]Pathology, Pamukkale University, Denizli, Türkiye
**Correspondence:** S. Turkucar
*Pediatric Rheumatology 2026*, **24(S1):**P361

**Introduction**
Erythema induratum of Bazin (EIB) is a rare tuberculid presenting as chronic lobular panniculitis with necrotizing vasculitis, predominantly

affecting young women and adolescents. It results from a delayed-type hypersensitivity reaction to Mycobacterium tuberculosis antigens rather than from direct cutaneous invasion [1]. EIB overlaps histopathologically with systemic vasculitides — particularly cutaneous polyarteritis nodosa (cPAN) — making diagnosis challenging when both coexist [2, 3].

**Objectives**

We present a 14-year-old girl with EIB coexisting with cPAN, complicated by antituberculous drug hypersensitivity, to highlight the diagnostic and therapeutic challenges of this rare constellation and raise awareness of EIB in the pediatric differential diagnosis of chronic lower-extremity ulcers.

**Methods**

A 14-year-old girl with no prior medical history or TB contact presented with a one-month history of painful, ulcerated lesions on the left leg and papular lesions on the right, unresponsive to antibiotics. QuantiFERON-TB Gold was positive; PPD induration was 20 mm. Chest CT excluded active pulmonary TB. Autoimmune serology (ANA, ANCA, anti-dsDNA, antiphospholipid antibodies) was negative. Left-leg biopsy showed granulomatous panniculitis consistent with EIB; right-leg biopsy revealed necrotizing panniculitis with transmural vessel wall infiltration, consistent with cPAN. [3,4] Antituberculous therapy (rifampicin, isoniazid, pyrazinamide) was suspended on day 8 due to maculopapular rash and fever. Sequential drug desensitization was applied successfully. Prednisolone, colchicine, and azathioprine were added for cPAN. Fever resolved and lesions regressed fully within six weeks. EIB can coexist with cutaneous necrotizing vasculitis and may mimic pyoderma gangrenosum or a primary bacterial infection, leading to diagnostic delay [1, 2]. Biopsying multiple anatomically distinct sites is essential, as different lesions may display divergent pathological patterns within the same patient. [3,4] Anti-TB drug hypersensitivity complicates management; desensitization protocols enable safe reintroduction of first-line agents. EIB must be considered early in young patients with chronic, antibiotic-resistant lower-extremity ulcers. PPD/QuantiFERON testing and deep skin biopsy are essential for timely diagnosis.

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**Disclosure of interest**

None declared.

## P362

### The difficulties of the diagnostic stage of verifying the diagnosis of ANCA-associated vasculitis in a teenage girl


Y. M. Spivakovskiy[1,*], M. D. Bezmenova[2], O. V. Volkova[2], A. Y. Spivakovskaya[3]
[1]Department of Faculty Pediatrics, State Medical University n.a. V.I. Razumovsky, [2]S.R.Mirotvortsev University Hospital, [3]Department of Hospital Pediatrics, State Medical University n.a. V.I. Razumovsky, Saratov, Russian Federation
**Correspondence:** Y. M. Spivakovskiy

*Pediatric Rheumatology 2026*, **24(S1):**P362

**Introduction**

ANCA-associated vasculitis (AAV) is a group of autoimmune diseases that mainly affect small/medium sized vessels. They are characterized by chronic immune inflammation of small vessels, a polymorphic clinical picture with frequent involvement of the upper respiratory tract and the presence of circulating autoantibodies to the cytoplasm of neutrophils. In childhood, AAV is extremely rare and therefore difficult to diagnose in a timely manner, which can lead to disability of patients and an unfavorable outcome, more often due to damage to the lungs and kidneys.

**Objectives**

The purpose of presenting a clinical case of AAV in a teenage patient is to analyze the difficulties and identify errors that occurred at the diagnostic stage, which significantly delayed the start of adequate program therapy for the disease.

**Methods**

The analysis of the clinical picture of the disease and the stages of the diagnostic process was carried out by examining the data of the medical documentation of a 16-year-old patient who was treated in the pediatric department of the University Hospital.

**Results**

The first symptoms of the disease appeared at the age of 15, when, after repeated episodes of acute respiratory infections, shortness of breath and bouts of dry, persistent cough were noted. With these complaints, the patient was observed by a pulmonologist at the regional hospital, where he was diagnosed with bronchial asthma (BA) and prescribed appropriate therapy. However, even against the background of increased doses of basic therapy, frequent severe seizures persisted, which led to monthly hospitalizations, in which systemic steroids (sCS) were invariably used, which gave only a short-term (no more than 2-3 weeks) improvement, followed by a recurrence of severe respiratory disorders. Similar episodes were recorded throughout the year. The issue of prescribing dupilumab was considered as in a patient with uncontrolled BA. In this regard, an allergist's consultation was conducted, followed by an examination by an otorhinolaryngologist and a rheumatologist at the University Clinic, who consistently expressed doubts about the correctness of the diagnosis and recommended a second examination. A comprehensive allergological examination revealed no signs of sensitization, while a CT- scan of the larynx with phonation revealed a narrowing of the glottis to critical values, and shortness of breath was inspiratory. From that moment on, the treatment takes place at the University Clinic. Antibodies to neutrophil elastase and lactoferrin were detected. A diagnosis of AAV with damage to the respiratory system was established, however, a morphological examination was required to clarify the variant. Unfortunately, the severity of the condition and the degree of respiratory failure required urgent medical measures: pulse therapy with the use of sCS and cytostatics, against which the girl's condition improved significantly. Subsequent follow-up showed that the clinical effect achieved with long-term therapy persists.

**Conclusion**

The presented clinical case indicates the lack of vigilance of the medical community in relation to AAV.

**Disclosure of interest**

None declared.

## P363

### Clinical manifestations of pediatric IGA vasculitis: a 5-year single-center study


Z. Stoyanova[1,*], K. Temelkova[1,2], T. Vasilev[1,2], M. Ganeva[1,2], A. Dasheva-Dimitrova[1,2], D. Kibarova-Hristova[1,2], S. Stefanov[1,2]
[1]Rheumatology, Children's University Hospital "Prof. Ivan Mitev", [2]Department of Pediatrics, Medical University, Sofia, Bulgaria
**Correspondence:** Z. Stoyanova

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**Introduction**

IgA vasculitis (IgAV) is the most common systemic vasculitis in childhood and is characterized by IgA-dominant immune deposits affecting small vessels of the skin, gastrointestinal tract, joints, and kidneys. Clinical manifestations and disease courses may vary according to geographic and ethnic background, while data from Eastern European pediatric populations remain limited. This study aimed to evaluate the demographic characteristics, clinical manifestations, laboratory findings, treatment strategies, and outcomes of children diagnosed with IgAV at a single tertiary center in Bulgaria.

**Objectives**

To evaluate the demographic characteristics, clinical manifestations, laboratory findings, treatment approaches, and prognostic features of children diagnosed with IgA vasculitis at a single tertiary center in Bulgaria.

**Methods**

Medical records of children hospitalized with IgAV at the Children's University Hospital in Sofia, Bulgaria, between January 2020 and December 2025 were retrospectively reviewed. Demographic, clinical, laboratory, imaging, and treatment data were collected and analyzed.

**Results**
A total of 125 patients were included (mean age 6.4 years; male-to-female ratio 1.27:1), with hospital admissions occurring predominantly during autumn and winter. A suspected infectious trigger was identified in 46% of children, while SARS-CoV-2 IgM positivity was detected in 25 patients (20%). Joint, gastrointestinal, and renal involvement were observed in 110 (88%), 59 (47%), and 14 (11%) patients, respectively; testicular involvement occurred in 5 patients. Joint manifestations preceding purpura onset were observed in 26 (21%) patients, while gastrointestinal symptoms before rash appearance occurred in 30 (24%) patients. Severe extra-renal complications occurred in 3 patients, including intestinal obstruction ($n$=1), ascites ($n$=1), and posterior reversible encephalopathy syndrome associated with epididymitis ($n$=1). No significant differences in inflammatory markers or D-dimer levels were observed between patients with and without abdominal or renal involvement. Corticosteroid treatment was started in 104 (83%) patients due to severe rash and edema, gastrointestinal or renal involvement, or persistent disease.
**Conclusion**
IgAV occurred predominantly in young children with slight male predominance and seasonal clustering, supporting a possible infectious trigger. Joint involvement was the most frequent clinical manifestation, followed by gastrointestinal and renal involvement. Extra-articular symptoms often preceded purpura onset, potentially contributing to delayed diagnosis. Although overall prognosis was favorable, severe gastrointestinal, neurological, and renal complications occurred in a minority of patients. Early recognition of atypical presentations may facilitate timely diagnosis and management.
**Disclosure of interest**
None declared.

## P364

### Biologic therapy in childhoodonset Takayasu arteritis: a Russian multicenter cohort study

V. A. Podzolkova[1,*], S. I. Valieva[2], M. M. Kostik[3], A. A. Glazyrina[4], Y. O. Kostina[5], S. K. Kurbanova[2], S. S. Vakhlyarskaya[6], V. A. Seraya[5], E. P. Turginekova[3], V. T. Yudina[2], O. V. Shpitonkova[1], M. K. Osminina[1], I. S. Avrusin[3], E. A. Lagutina[6], N. A. Geppe[1]
[1]Children's Diseases, Sechenov First Moscow State Medical University, [2]Morozov Children's Moscow City Clinical Hospital, Moscow, [3]Hospital Pediatrics, Saint Petersburg State Pediatric Medical University, Saint Petersburg, [4]Pirogov Russian National Research Medical University, [5]Pediatric Rheumatology, Sechenov Center of Maternity and Childhood, [6]Russian Children's Clinical Hospital of Pirogov Russian National Research Medical University, Moscow, Russian Federation
**Correspondence:** V. A. Podzolkova
*Pediatric Rheumatology 2026,* **24(S1):**P364

**Introduction**
Biologic diseasemodifying antirheumatic drugs (bDMARDs), especially tocilizumab (antiIL6) and TNFα inhibitors (TNFi), are being used more often in paediatric Takayasu arteritis (TA). Nevertheless, realworld evidence on their effectiveness in children remains scarce. Furthermore, which presenting features may predict later biologic switching, and how the two drug classes compare in everyday practice, are still open questions.
**Objectives**
To characterise the use of bDMARDs in a Russian paediatric TA cohort; to determine whether any initial clinical or laboratory findings are associated with a higher number of sequential bDMARD lines (reflecting treatment switches); and to compare disease activity and laboratory parameters at the final followup between patients who were receiving tocilizumab versus those receiving TNF inhibitors as their current biologic therapy.
**Methods**
Retrospective multicentre study of 79 children fulfilling EULAR/PRINTO/PRES criteria for TA. Associations between presenting features and the count of bDMARD lines were assessed with Spearman's correlation and linear regression. Patients were divided according to the biologic agent used at the last followup (tocilizumab, $n$=28; TNFi, $n$=10). Group comparisons were performed using the MannWhitney U test and Fisher's exact test. Statistical significance was set at $p<0.05$.
**Results**
bDMARDs were given to 40/79 patients (50.6%). Comparing those who ever received bDMARDs ($n$=40) with those who did not ($n$=39) revealed several significant differences at disease onset: the bDMARDtreated group had higher median CRP (48.0 vs. 18.0 mg/L, $p$=0.012), higher mean ESR (54.7 vs. 38.7 mm/h, $p$=0.003), lower mean haemoglobin (102.7 vs. 110.4 g/L, $p$=0.020), and a greater median number of affected vessels (6.0 vs. 4.0, $p$=0.018). They also experienced longer followup (median 30.5 vs. 15.0 months, $p<0.001$), and at the last evaluation they more often had involvement of the ascending aorta (31.6% vs. 10.3%, $p$=0.045) and the aortic arch (36.8% vs. 10.3%, $p$=0.022). Firstline bDMARD was tocilizumab in 28 patients (70%) and TNFi in 12 (30%). Tocilizumab served as the first and sole bDMARD in 24/40 cases (60%). Among all initial markers examined, only an elevated white blood cell count (WBC) correlated significantly with a greater number of subsequent bDMARD lines (after correcting the regression direction: $\rho = +0.472$, $p$=0.003). At the final followup, patients on tocilizumab ($n$=28) had less active disease (ITAS.A>0: 25.9% vs. 66.7%, $p$=0.046), lower ESR (5.0 vs. 23.0 mm/h, $p$=0.016) and lower WBC (6.5 vs. $9.44\times10^9$/L, $p$=0.034) than those on TNFi ($n$=10), whereas CRP and the total number of bDMARD lines did not differ. Children who received tocilizumab as firstline monotherapy tended to be older at disease onset (13.0 vs. 10.5 years, $p$=0.052).
**Conclusion**
The need for bDMARDs was associated with higher baseline inflammatory markers, more extensive vascular damage, and longer followup, reinforcing that more severe disease drives biologic use. Leukocytosis at diagnosis predicted more complex biologic therapy, and tocilizumab was associated with better disease control. Prospective studies are needed to confirm these pilot findings.
**Disclosure of interest**
None declared.

## P365

### The effect of gender on functional status, pain, quality of life, and fatigue in children with IGA vasculitis

Z. Tatar[1,*], S. S. Gümüş[2], E. Gür Kabul[3], B. Başakcı Çalık[2], S. N. Tığrak[4], S. Türkuçar[4]
[1]Home Care Services Program, Department of Health Care Services, Şırnak University, Şırnak, [2]Faculty of Physiotherapy and Rehabilitation, Pamukkale University, Denizli, [3]Department of Physiotherapy and Rehabilitation, Faculty of Health Sciences, Uşak University, Uşak, [4]School of Medicine, Pediatric Rheumatology, Pamukkale University, Denizli, Türkiye
**Correspondence:** Z. Tatar
*Pediatric Rheumatology 2026,* **24(S1):**P365

**Introduction**
IgA vasculitis (IgAV) is more common in men, and sex-related differences have been reported in terms of some organ involvement and prognosis (1). However, it has been noted that these differences are not robust or consistent (2). This suggests that the effect of sex on various parameters may not be definitive.
**Objectives**
The aim of this study is to investigate the effect of gender on functionality, pain, quality of life, and fatigue in children with IgAV.
**Methods**
The study included 11 girls (mean age 10±2.64 years) and 10 boys (mean age 9.66±2.08). After collecting demographic and disease-related data, participants' pain levels were assessed using the Visual Analog Scale (VAS). Quality of life was assessed using the Pediatric Quality of Life Inventory Rheumatology Module (PedsQL), and fatigue was assessed using the PedsQL Multidimensional Fatigue Scale (PedsQL-Fatigue) child self-report forms and parent-proxy-report. Functional status and the level of limitation in daily living activities were assessed using the Childhood Health Assessment Questionnaire (CHAQ). The Mann–Whitney U test was used to compare differences between the two groups. $p<0.05$ was considered statistically significant.
**Results**
There were no differences between girls and boys in terms of age ($p$=0.816), height ($p$=0.694), weight ($p$=0.902), and duration of illness ($p$=0.737). When comparing the data of girls and boys, there was no difference between the two groups in terms of pain intensity ($p$=0.66),

but there were significant differences in favor of girls in terms of CHAQ-general well-being ($p$=0.009), PedsQL-worry ($p$=0.029), PedsQL-fatigue total ($p$=0.001), PedsQL-fatigue general tiredness ($p$=0.001), PedsQL-fatigue fatigue during sleep and rest ($p$=0.003), and PedsQL-fatigue cognitive fatigue ($p$=0.024). According to the parent proxy-report results, there was a significant difference in favor of parents of girls in terms of PedsQL-total ($p$=0.016), PedsQL-fatigue total ($p$=0.005), and PedsQL-fatigue general fatigue ($p$=0.005).

**Conclusion**

The results of this study suggest that in children with IgAV, male gender may have a worse impact on overall well-being, quality of life, and fatigue. This could indicate that boys are more affected by the disease than girls. Furthermore, parents of boys may have a worse perception of their children's quality of life and fatigue.

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**Disclosure of interest**

None declared.

## P366

### Educational videos in pediatric rheumatology

T. Hospach[1,*], F. Weller[2], H. Girschick[3], J.-P. Haas[4], A. Heiligenhaus[5], J. Roth[6], A. Raab[7], F. Dressler[7], K. Minden[8], D. Windschall[9]
[1]Center for Pediatric Rheumatology, Stuttgart, [2]Prof-Hess Children's Hospital, Bremen, [3]Children's Hospital Friedrichshain, Berlin, [4]German center for pediatric rheumatology, Garmisch-Partenkirchen, [5]Eye Center St Franziskus Hospital, Münster, Germany, [6]Pediatric Rheumatology, Lucerne, Switzerland, [7]University, Children's Hospital, Hannover, [8]German Research Center for Rheumatology, Berlin, [9]Center for Rheumatology Münsterland, Sendenhorst, Germany
**Correspondence:** T. Hospach
*Pediatric Rheumatology 2026*, **24(S1):**P366

**Introduction**

In clinical practice, patients with rheumatic diseases are often treated by physicians who are not yet experienced in rheumatology or by general pediatricians. Individual knowledge transfer is time- and resource-intensive, and relevant pathological findings are often not available in the learning setting. Learning through printed materials is no longer considered up to date. As a result, both patient care and training are frequently suboptimal. Concise, practice-oriented videos have the potential to provide targeted and time-efficient mini-training. Such videos can be accessed via streaming, both during rheumatology-specific activities (e.g., performing a joint examination or ultrasound) and for preparation. Against this background, four educational videos were produced, targeting pediatric residents without rheumatology expertise and aspiring rheumatologists.

**Objectives**

The following objectives were defined.
1. Improvement of the quality of care for patients with rheumatic diseases.
2. Enhancement of training for residents.
3. Promotion and recruitment of young professionals for the field of pediatric rheumatology.
4. Reduction of resource demands for educators.

**Methods**

Within a professional recording setting, several educational videos were produced.

**Results**

Videos are available for the following topics.
1. Learning the musculoskeletal examination.
2. Demonstration of joint pathological findings.
3. Interdisciplinary approach to uveitis.
4. Musculoskeletal ultrasound.
5. Polyarthritis (in progress).

The videos can be accessed via the QR code shown.

**Conclusion**

Concise and practice-oriented educational videos can help to improve training in rheumatology. enhance the quality of patient care. increase the attractiveness of the field of rheumatology. An evaluation is planned.

**Disclosure of interest**

None declared.

## P367

### Clinical spectrum and management of cardiac manifestations in pediatric rheumatic diseases

A. Hercberg Moreno[1], R. Alcobendas Rueda[1], A. Remesal Camba[1], C. Udaondo Gascón[1,*], L. Martínez Marcelo[1], V. Courel del Río[2]
[1]Hospital Universitario La Paz, Madrid, [2]Hospital Valle del Nalón, Oviedo, Spain
**Correspondence:** C. Udaondo Gascón
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**Introduction**

Cardiac involvement in pediatric rheumatic diseases (PRD) represents a critical clinical challenge, ranging from subclinical findings to life-threatening emergencies such as severe myocarditis, acute heart failure, and arrhythmias. Chronic inflammation and autoimmunity not only damage cardiac structures acutely but also accelerate premature atherosclerosis, increasing long-term morbidity and mortality. Early detection is vital, as cardiac injury can be the primary determinant of initial prognosis.

**Objectives**

To describe the frequency, spectrum of cardiac manifestations and chronology of onset (at disease presentation vs. during follow-up) in a pediatric cohort with rheumatic diseases, and to evaluate their therapeutic response.

**Methods**

Clinical chart review.

**Results**

We analyzed a cohort of pediatric patients diagnosed with Systemic Lupus Erythematosus (SLE), Takayasu Arteritis and autoinflammatory syndromes. A multimodal assessment was performed using electrocardiography (ECG), echocardiography, and CT angiography (CTA). Inflammatory biomarkers—including C-reactive protein (CRP) and Erythrocyte Sedimentation Rate (ESR)—were recorded. The timing of cardiac manifestations was classified as occurring either prior to or after the diagnosis of the underlying rheumatic disease. In the analyzed cohort ($n$=9; 2 SLE, 2 Takayasu Arteritis and 5 recurrent or chronic pericarditis), cardiac involvement was predominantly identified as an early manifestation. Specifically, 87.5% of patients (7 out of 8 recorded cases) presented with cardiac clinical features prior to the diagnosis of the underlying rheumatic disease. This trend is consistent with recent literature indicating that in juvenile SLE, 84% of patients exhibit cardiac abnormalities at diagnosis, with 19% presenting even before the diagnosis is established. Clinical findings in this cohort included lupus myocarditis complicated by cardiorespiratory arrest, dilated cardiomyopathy, and coronary aneurysms. Pericardial effusion was the most frequent finding, primarily detected via echocardiography. Regarding management, 33% of the cohort required Anakinra, while the remaining patients were managed by optimizing baseline immunosuppressive therapy. Acute cardiac episodes strongly correlated with significantly elevated acute-phase reactants (CRP >150 mg/L).

**Conclusion**

Cardiac manifestations in PRD are not merely late-stage complications but frequently constitute the initial presentation or "debut" of the disease. Consequently, cardiovascular screening with ECG and echocardiography should be mandatory from the first clinical suspicion of a rheumatic condition. Aggressive control of systemic inflammation from onset is imperative to mitigate the risk of cumulative damage and premature cardiovascular disease in adulthood.

**Disclosure of interest**

None declared.

## P368

### Unexplained recurrent myopericardial inflammation in adolescents: unmasking underlying celiac disease

C. Udaondo[1,*], N. C. Fuertes[2], R. Alcobendas Rueda[1], G. Moral Garcia[3], A. Hercberg Moreno[1], A. Remesal Camba[1], L. Martinez Marcelo[1], S. Barreiro Perez[1], C. Lopez Garcia[4]
[1]Pediatric Rheumatology, Hospital La Paz, Madrid, [2]Pediatric, Hospital Puerta del Mar, Cadiz, [3]Rheumatology, Hospital Universitario Virgen Macarena, sevilla, [4]Pediatric gastroenterology, Hospital La Paz, Madrid, Spain
**Correspondence:** C. Udaondo
*Pediatric Rheumatology 2026*, **24(S1):**P368

**Introduction**
Recurrent or otherwise unexplained pericarditis, myocarditis, or dilated cardiomyopathy should prompt evaluation not only for infections, classic autoimmune diseases, and metabolic causes, but also for underlying celiac disease1. Cardiac involvement may precede, accompany, or follow the diagnosis of celiac disease, and may occur without gastrointestinal symptoms2,3.
**Objectives**
To present two adolescent cases of recurrent myocarditis and pericarditis as initial manifestation of celiac disease.
**Methods**
Two case reports are described. Data were extracted from medical records and complemented by a literature review.
**References** Case 1: A 12yearold male was admitted with acute chest pain and bloody diarrhea. He had a history of two prior episodes of myocarditis in the context of acute gastroenteritis. Complementary tests showed elevated troponin, with echocardiography and ECG findings compatible with myocarditis. Stool PCR for *Campylobacter* was positive. NSAIDs and colchicine were started. Extensive autoimmune and genetic workup for cardiomyopathy and connective tissue disease was performed with negative results. Serology showed markedly elevated IgA antitissue transglutaminase and positive antiendomysial antibodies, with a moderaterisk HLA haplotype. A glutenfree diet was started without prior biopsy. The patient remains asymptomatic. Case 2: A 14yearold male presented idiopathic pericarditis with large pericardial effusion and serositis requiring ICU admission and drainage, with elevated inflammatory markers, negative autoimmune studies and rapid response to anakinra plus antiinflammatory drugs. 3 months after voluntary discontinuation of anakinra, he developed a new episode of pericarditis. Study revealed markedly elevated IgA antitissue transglutaminase and positive antiendomysial antibodies, leading to the diagnosis of celiac disease. He remains asymptomatic on anakinra, colchicine and a glutenfree diet.
**Conclusion**
Unexplained or recurrent pericarditis or myocarditis should prompt a broad systemic search, in which celiac disease is a prevalent and potentially treatable consideration. Recognizing this heart–gut link can allow timely diagnosis and gluten-free treatment that may reverse or stabilize cardiac damage in selected patients.

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**Disclosure of interest**
None declared.

## P369

### Primary hypertrophic osteoarthropathy due to *HPGD* and *SLCO2A1* variants in four Slovak families

D. Mišíková[1,*], B. Balažiová[1], E. Vrtíková[2], Z. Seligova[3], K. Skalická[1], G. Hrčková[1], O. Hamidová[1], T. Dallos[1]
[1]Paediatrics, National Institute of Children's Diseases, Bratislava, [2]National Institute of Rheumatic Diseases, Piešťany, [3]Paediatric rheumatology outpatient clinic, Kežmarok, Slovakia
**Correspondence:** D. Mišíková
*Pediatric Rheumatology 2026*, **24(S1):**P369

**Introduction**
Primary hypertrophic osteoarthropathy (PHO)/pachydermoperiostosis (Touraine-Solente-Golé syndrome) is a very rare disorder of impaired prostaglandin (PG) catabolism/distribution due to variants in *HPGD* (15-hydroxyprostaglandin dehydrogenase) or *SLCO2A1* (PG transporter) genes. Persistently elevated PGE2 causes digital clubbing, skeletal abnormalities (bone hypertrophy, periostosis and acroosteolysis), skin changes (pachydermia, seborrhoea, acne and hyperhidrosis), chronic enteropathy and anaemia.
**Objectives**
To characterize the clinical presentation, genotype-phenotype correlations and inheritance patterns in four PHO patients and their pedigrees, diagnosed in Slovakia between 1999 and 2026.
**Methods**
Description of clinical, laboratory, molecular genetics and imaging findings in affected children and their families.
**Results**
Four Slovak children with PHO and their families were investigated. Among 2 female and 2 male patients, the median age of symptom onset was 10.5 years (range 2-17), but due to a significant diagnostic delay of 3.1 years (range 0.08-17), the median age at diagnosis was 17 years (range 12-18). Clinical symptoms included digital clubbing and arthralgia/arthritis in all patients ($n$=4), followed by hyperhidrosis ($n$=3) and diarrhoea ($n$=1). Imaging studies revealed periostosis in 3 patients. Molecular genetic testing confirmed pathogenic variants in the *HPGD* gene ($n$=1 homozygous c.175_176delCT and $n$=1 compound heterozygous c.120delA/c.446 C> T) in 2 female patients, and pathogenic variants in the *SLCO2A1* gene ($n$=1 homozygous c.302T> C and $n$=1 heterozygous c.1660G> A) were found in 2 male patients. We identified autosomal recessive inheritance in three families and autosomal dominant inheritance in one family. Management included long-term NSAIDs (preferably selective COX-2 inhibitor celecoxib). One patient with refractory progressive disease was treated with intravenous bisphosphonates, intra-articular and systemic corticosteroids, with limited effect; new therapeutic options are being considered.
**Conclusion**
Our small cohort of PHO patients illustrates a significant genotype–phenotype variability linked to *HPGD* and *SLCO2A1* gene variants in all clinically suspected patients. NSAID usually lead to symptomatic relief, but escalation of treatment to csDMARDS or corticosteroids may be required in severe cases.
Written informed consent for publication of all concerned individuals was obtained.
**Disclosure of interest**
None declared.

## P370

### Scurvy in the modern era: mimicking rheumatic disease

D. Lages de Miranda[1,*], R. A. Santos[1], J. Baptista de Lima[2], I. Couto Guerra[3], S. Alves[2], C. Zilhão[2]
[1]Pediatrics, [2]Pediatric Rheumatology Unit, [3]Pediatric Hematology Unit, Centro Materno-Infantil do Norte, Unidade Local de Saúde de Santo António, Porto, Portugal
**Correspondence:** D. Lages de Miranda
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**Introduction**
Scurvy is a rare disease in high-income countries but remains an important differential diagnosis in children with musculoskeletal complaints and restrictive eating behaviours.
**Objectives**
To highlight scurvy as a diagnostic pitfall in pediatric patients presenting with musculoskeletal symptoms.
**Methods**
A 5-year-old male with non-verbal autism spectrum disorder and severe food selectivity was admitted for investigation of progressive and severe musculoskeletal pain with refusal to walk. He had a one-month history of lower limb pain, claudication progressing to inability to bear weight and swelling of wrists, knees and ankles. Petechiae over the lower limbs and gingival bleeding were also reported. Parents referred a similar presentation the year before, associated with dental caries, for which anti-inflammatory drugs, oral corticosteroids and antibiotics were tried with limited improvement. On examination, he appeared pale, with follicular hyperkeratosis in the upper and lower limbs and petechiae on both legs and feet. Gingivitis was noted. He presented with bilateral wrist swelling and limited dorsiflexion, bilateral knee swelling with preferred flexion, and swelling of the ankles and dorsum of the feet. The swelling extended beyond the articular margins, without associated warmth or erythema. Initial laboratory evaluation showed mild microcytic anemia with otherwise unremarkable autoimmune and infectious work-up and no indicators of bleeding dyscrasia. Lower limb imaging demonstrated diffuse osteopenia, dense metaphyseal bands consistent with Frankel lines, associated growth arrest lines and metaphyseal irregularities, with a possible epiphyseal ring sign at the proximal fibula. Bone marrow examination and scintigraphy excluded malignant etiologies. Given the history of extreme dietary restriction, with exclusive consumption of pasta, cake and yoghurt for 3 years, the cutaneous findings, gingival bleeding and radiological abnormalities, scurvy was suspected. Seric ascorbic acid was measured and found to be <0.10 mg/dL. Oral vitamin C supplementation was initiated (100 mg 12/12 h), leading to clinical improvement, by progressive recovery of walking and resolution of gingivitis and cutaneous lesions. Scurvy should be considered in children with restrictive diets presenting with musculoskeletal symptoms, particularly when accompanied by hemorrhagic or dermatological signs. Recognition of this condition allows prompt treatment and prevents unnecessary diagnostic and therapeutic interventions.
**Disclosure of interest**
None declared.

## P372

### The forgotten side of pediatric primary Raynaud's phenomenon: non-pharmacological management and lifestyle factors

E. E. Firat[1,*], T. Güzel[1], M. T. Karadogan[1], B. Cinpolat[1], Z. Haziroglu Okmen[1], F. Cakmak[1]
[1]Peidatric Rheumatology, Basaksehır Cam ve Sakura City Hospital, Istanbul, Türkiye
**Correspondence:** E. E. Firat
*Pediatric Rheumatology 2026*, **24(S1):**P372

**Introduction**
Primary Raynaud Phenomenon (pRP) is characterized by episodic vasospasm of peripheral vessels without an underlying connective tissue disease. In many patients, symptoms can be adequately controlled with non-pharmacological strategies such as maintaining warmth, engaging in regular physical activity, and maintaining healthy nutritional habits, thereby reducing the need for pharmacological treatment. However, lifestyle characteristics and supportive management behaviors in pediatric pRP have been insufficiently investigated.
**Objectives**
This study evaluated lifestyle-related supportive factors (Mediterranean diet adherence, physical activity, vitamin status, and complementary nutritional approaches) in pediatric pRP patients, and examined their relationships with disease severity and capillaroscopic findings.
**Methods**
This cross-sectional study included 20 pediatric pRP patients. Mediterranean diet adherence was assessed via the KIDMED index, and physical activity levels through age-appropriate questionnaires. Omega-3 supplementation and beetroot consumption were recorded as supportive practices. Serum ferritin, vitamin B12, folate, and 25-OH vitamin D levels were retrospectively reviewed. Disease severity was evaluated using the Raynaud Condition Score (RCS), and microvascular involvement by nailfold videocapillaroscopy (NVC). Statistical analyses examined associations between lifestyle variables, RCS, and NVC findings.
**Results**
Of the 20 patients, 55% were female, with a mean age of 15.38 ± 1.39 years. The mean KIDMED score was 4.1 ± 2.86, indicating predominantly low-to-moderate adherence to the Mediterranean diet. Physical activity levels were generally low, with 70% of patients classified as low-active. Vitamin D deficiency was present in 60% of patients, whereas no vitamin B12 deficiency was detected. No significant associations were observed between RCS and Mediterranean diet adherence, physical activity, vitamin levels, omega-3 supplementation, or beetroot consumption. However, patients with nonspecific capillaroscopic abnormalities had significantly higher RCS values compared to those with normal capillaroscopic findings (7.15 ± 2.79 vs. 4.60 ± 2.01, $p$=0.023).
**Conclusion**
Pediatric pRP patients demonstrated suboptimal lifestyle profiles, particularly regarding physical activity and Mediterranean diet adherence. Although no direct associations with disease severity were identified, likely due to the limited sample size, these findings highlight the potential importance of supportive lifestyle measures in the management of pediatric pRP. The association between capillaroscopic abnormalities and higher symptom burden further supports the clinical relevance of microvascular assessment. Larger prospective studies are needed to determine whether lifestyle optimization may contribute to symptom control and vascular protection in pediatric pRP.
**Disclosure of interest**
None declared.

## P373

### Recurrent pericarditis in Sotos syndrome: an emerging epigenetic-inflammatory phenotype

G. Inguscio[1,2,*], M. De Angelis[1,2], M. Sanz-Cuesta[3], D. Ferri-Rufete[4], A. Insalaco[5], C. Celani[5], A. Aquilani[5], E. Hoppenreijs[6], Y. Herman[7], C. Hinze[8], G. Tiller[9], K. Rücklová[10,11], Á. Pérez-Casares[4], A. F. Martínez Monseny[12], J. Calzada-Hernández[13], N. P. Anand[14], M. Serrano Gimare[15], E. Boesaard[6], I. Pagnini[2], I. Maccora[2], M. V. Mastrolia[2], L. Gatti[2], E. Marrani[2], S. Romano[1,2], G. Simonini[1,2], T. Giani[2]
[1]Paediatric Department, University of Florence, [2]Rheumatology Unit, AOU Meyer Children's Hospital IRCCS, Florence, Italy, [3]Department of Pediatrics, Hospital General, Parc Sanitari Sant Joan de Déu, [4]Department of Pediatric Cardiology, Hospital Sant Joan de Déu, Barcelona, Spain, [5]Division of Rheumatology, Ospedale Pediatrico Bambino Gesù IRCCS, Rome, Italy, [6]Department of Pediatric Rheumatology, Amalia Children's Hospital, Radboud University Medical Center, Nijmegen, Netherlands, [7]Royal Children's Hospital, Melbourne, Australia, [8]Department of Pediatric Rheumatology and Immunology, Münster University Hospital, Münster, Germany, [9]Department of Rheumatology, The Royal Children's Hospital, Melbourne, Australia, [10]Klinikum Stuttgart, Stuttgart, Germany, [11]Third Faculty of Medicine, Charles University, Prague, Czech Republic, [12]Department of Genetics, Hospital Sant Joan de Déu, [13]Department of Pediatric Rheumatology, Hospital Sant Joan de Déu, Barcelona, Spain, [14]Division of Rheumatology, Manipal Hospital, Bangalore, India, [15]Department of Pediatric Neurology, Hospital Sant Joan de Déu, Barcelona, Spain
**Correspondence:** G. Inguscio
*Pediatric Rheumatology 2026*, **24(S1):**P373

**Introduction**
Sotos syndrome (SS), a rare overgrowth disorder caused by pathogenic NSD1 variants, is associated with neurodevelopmental impairment and congenital heart disease. Pericardial inflammatory disease has rarely been reported in SS, and its clinical spectrum remains poorly characterized.
**Objectives**
To describe the clinical phenotype, disease course, treatment response, and inflammatory features of pediatric patients with SS-associated pericarditis.
**Methods**
A retrospective, international multicenter study including patients with molecularly confirmed SS and pericarditis from 5 referral centres was

conducted (2010–2025). Severe pericarditis was defined by cardiac tamponade and/or the need for invasive pleural or pericardial management.
**Results**
13 patients (6 males) developed acute pericarditis at a median age of 12 years (range 1.4–17). Eight patients carried high-impact NSD1 variants (5 nonsense, 2 deletions, 1 splicing), whereas 5/13 carried missense variants. Chest pain was the most common presenting symptom (69%), frequently associated with fever and dyspnea. Pericardial effusion was present in all patients, including cardiac tamponade in 2 (15%). Severe pericarditis and pleuro-pulmonary involvement each occurred in 9/13 patients (69%). ECG abnormalities were documented in 9/13 (69%), mainly non-specific repolarization abnormalities and T-wave changes. CRP elevation was observed in 11/13 patients (85%), leukocytosis in 7/13 (54%), and mild troponin elevation in one patient (8%). Additional variants potentially related to inflammatory or cardiovascular susceptibility were identified in 4/13 patients (31%) (MEFV $n$=2, DNMT3A $n$=1, MYBPC3 $n$=1), all associated with severe disease requiring invasive management. Recurrent pericarditis occurred in 10/13 patients (77%), with up to 4 relapses in one patient. Pleural and/or pericardial drainage procedures were required in 9/13 (69%), and one patient underwent pericardiectomy. Corticosteroids and colchicine were each required in 11/13 patients (85%), while anakinra was administered in 9/13 (69%), mainly in recurrent or treatment-refractory disease with clinical remission outcome. All patients carrying additional inflammatory-related variants required IL-1 blockade.
**Conclusion**
Pericarditis in SS shows an aggressive relapsing inflammatory phenotype characterized by large effusions, frequent pleuro-pulmonary involvement, substantial recurrence burden, and common need for IL-1 blockade. These findings expand the inflammatory spectrum of SS and suggest a possible modulatory role of additional inflammatory-related variants beyond the NSD1 defect itself.
**Disclosure of interest**
None declared.

## P374

### A comparative analysis of pediatric- and adult-onset primary Sjogren syndrome in a Brazilian tertiary center

V. Balbi[1], N. E. Aikawa[2], K. T. Kozu[1], S. Pasoto[2], A. Franco[2], E. Bonfa[2], C. A. A. Silva[1], L. M. A. Campos[1], A. M. Elias[1,*]
[1]Pediatric Rheumatology Unit, Children's Institute, Hospital das Clínicas, Faculdade de Medicina, Universidade de São Paulo, [2]Division of Rheumatology, Hospital das Clínicas, Faculdade de Medicina, Universidade de São Paulo, São Paulo, Brazil
**Correspondence:** A. M. Elias
*Pediatric Rheumatology 2026*, **24(S1):**P374

**Introduction**
Sjogren syndrome (SS) is a rare inflammatory and autoimmune condition in childhood, with limited pediatric studies conducted to date. Few studies have directly compared clinical and laboratory features between adult SS(aSS) and pediatric SS(pSS) patients.
**Objectives**
To evaluate demographic, clinical, laboratory, histopathological, and treatment data in primary pSS compared to adult population.
**Methods**
Medical records of 152 patients [10pSS (≤18 years-old) and 142aSS] followed at our tertiary center were reviewed to compare clinical and therapeutic manifestations. pSS and aSS were classified according to 2002 American-European Consensus Group(AECG) and 2016 ACR-EULAR Classification Criteria for SS.
**Results**
Median age at diagnosis was 13(2.75-16) years in pSS and 50(20-70) years in aSS. Median disease duration was 5.5(0.25-11.9) years in pSS and 9(0.66-36) years in aSS. Female sex were similar in both groups (80%vs.93%, $p$=0.180, respectively). 2002 AECG classification criteria were fulfilled by 30% of pSS and 96.4% of aSS patients ($p$<0.001). Considering 2016 ACR-EULAR classification criteria, 30% of pSS and 87.3% of aSS met these criteria ($p$<0.001). Presence of dry eyes (50%vs.95%,$p$<0.001), dry mounth (40%vs.95%, $p$<0.001), and arthralgia (20%vs.59%, $p$=0.021) were more frequent in aSS. Other manifestations such as glandular enlargement, skin involvement, arthritis, cytopenias, and renal and neurological involvement were similar in both groups ($p$>0.05). Laboratory anormalities, including Schirmer's test, sialography, anti-Ro/SSA, anti-La/SSB, RF, and low levels of C3 and C4 did not differ between the groups, except by the signicant higher median of IgG (1890vs.2476,$p$=0.005) and presence of ANA (70%vs.97.9%,$p$=0.004) in adult patients. Regarding treatment, hydroxychloroquine (50%vs.33%,$p$=0.023) was used more frequently in pediatric population, while the use of oral corticosteroids, methylprednisolone pulses, methotrexate, azathioprine, mycophenolate mofetil, cyclophosphamide, and Rituximab was similar in pSS and aSS. Lymphoma(2.8%) and death(9.9%) were only observed in adult cohort.
**Conclusion**
Primary pSS was rarely observed in our terciary center, potentially due to its under recognition, since children often do not fulfil the classification criteria developed for adult populations. The presence of recurrent parotid, seen in 70% of paediatric patients in the presente study, should alert to the possibility of pSS.
**Disclosure of interest**
None declared.

## P375

### Pediatric synovial chondromatosis: a comprehensive literature review

M. Fastiggi[1,*], L. Barchi[1], B. Fioravanti[2], G. Martini[3], A. De Fanti[4], M. Cappella[1]
[1]Pediatric Rheumatology Unit, Arcispedale Santa Maria Nuova, Reggio Emilia, [2]Pediatric Unit, University of Modena and Reggio Emilia, Modena, [3]Pediatric Division, University Hospital S Maria della Misericordia, Udine, [4]Pediatric Unit, Arcispedale Santa Maria Nuova, Reggio Emilia, Italy
**Correspondence:** M. Fastiggi
*Pediatric Rheumatology 2026*, **24(S1):**P375

**Introduction**
Synovial chondromatosis (SC) is a rare benign condition caused by synovial metaplasia with formation of intra-articular osteocartilaginous bodies[1]. SC typically affects adults and large joints, while pediatric cases are uncommon and may mimic rheumatic disease leading to delayed diagnosis[2]. To fill this knowledge gap, we report a comprehensive literature review of SC in children.
**Objectives**
Objective of the study was to systematically review the available pediatric literature on SC in order to characterize its clinical presentation, diagnostic pathway, therapeutic management and outcomes.
**Methods**
A comprehensive literature search was conducted using PubMed, Google Scholar and Embase databases. The following search terms were used: "synovial chondromatosis" OR "synovial osteochondromatosis", filtered for pediatric age. Clinical, instrumental and management data were collected. English full-text studies with patients aged ≤18 years of age with histopathologically confirmed diagnosis of synovial chondromatosis were included for the study. Secondary pediatric synovial chondromatosis cases with underlying joint disease were excluded.
**Results**
39 publications were included in the final analysis. Mean age at diagnosis was 10 years (range 2–18) with a slight male predominance (63%). The interval between symptom onset and diagnosis was 12 months (range 1–72), typically characterized by localized pain (92%) and functional limitation (75%). The knee was the most frequently involved joint (38%), followed by the shoulder (17%). All patients underwent surgical treatment, with open surgery performed in 63% of cases. Arthroscopic procedures were associated with a significantly higher recurrence rate compared to open procedure (OR 14.7; $p$ = 0.017). The overall recurrence rate in pediatric patients was 12%, comparable to adult populations. Long-term outcomes were generally favorable, although 20% of patients experienced postoperative sequelae.
**Conclusion**
SC should be considered in children with persistent monoarticular symptoms. Open surgical excision provides excellent outcomes in pediatric patients, with low recurrence rates and favorable prognosis compared with arthroscopy. Multidisciplinary collaboration and long-term follow-up are crucial to optimize outcomes and monitor for recurrence.

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**Disclosure of interest**
None declared.

## P376

### Heterogenous clinical presentation of juvenile Sjögren syndrome: experience from two spanish tertiary centers

A. Villarejo Perez[1,*], A. Capilla Miranda[2], R. Gonzalez Preciado[1], E. Enriquez Merayo[1], L. Fernandez Silveira[2], M. Camacho Lovillo[2], J. De Inocencio[1]
[1]Pediatric Rheumatology Unit, University Hospital 12 De Octubre, Madrid, [2]Pediatric Infectious Diseases, Rheumatology And Immunology Unit, University Hospital Virgen Del Rocio, Sevilla, Spain
**Correspondence:** A. Villarejo Perez
*Pediatric Rheumatology 2026*, **24(S1):**P376

**Introduction**
Juvenile Sjögren syndrome (jSS) is a chronic systemic autoimmune disorder characterized by lymphocytic infiltration and progressive dysfunction of the exocrine glands, accounting for approximately 1-1.3% of all cases of primary Sjögren syndrome. Although adults typically present with sicca symptoms, pediatric patients exhibit a broader and more heterogeneous clinical spectrum. In addition, the limited performance of the 2016 ACR/EULAR classification criteria in pediatric populations further complicates the diagnosis of jSS.
**Objectives**
To describe clinical presentation and laboratory findings of patients diagnosed with jSS.
**Methods**
A retrospective study including cases diagnosed since 2012 was conducted at two tertiary centers in Spain. Data regarding symptoms at diagnosis, laboratory findings, and diagnostic test results were collected.
**Results**
Fourteen patients (12/14 female) were diagnosed with jSS, with a median age at symptom onset of 13.2 years and a median age at diagnosis of 13.6 years, corresponding to a median diagnostic delay of 4.8 months. The most frequently reported symptom were xerophthalmia, recurrent parotitis and xerostomia (35.7%), followed by arthralgia, asthenia, and fever (21.4%), and arthritis and recurrent oral aphtosis (7.1%). One patient was asymptomatic at diagnosis. Two patients presented with cytopenias at disease onset, one of whom secondary to thrombotic thrombocytopenic purpura. Anti-Ro/SSA antibodies were detected in 71.4% of patients, while anti-La/SSB antibodies were present in 50%. Antinuclear antibodies were observed in 64.3% of patients. Rheumatoid factor positivity was observed in 76.9% of cases. Median ESR, CRP, and IgG levels at diagnosis were 15 mm/h (IQR 7.5-34.5), 0.11 mg/dL (IQR 0.03-0.7), and 1,625 mg/dL (IQR 1,363-2,081), respectively. Salivary gland ultrasound abnormalities, according to SGUS scoring, were identified in 64.3% of patients. Keratoconjunctivitis affecting at least one eye was observed in 42.8% of patients; 28.6% had an abnormal Schirmer test, and 14.2% had an Ocular Staining Score ≥5. Labial salivary gland biopsy was performed in 13 of 14 patients and demonstrated focal lymphocytic sialadenitis with a focus score ≥1 in 84.6% of cases.
**Conclusion**
jSS presents with heterogeneous clinical manifestations, frequently including sicca symptoms, recurrent parotitis, and systemic involvement. SGUS abnormalities and labial salivary gland biopsy were commonly observed and may support early diagnosis in pediatric patients.

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**Disclosure of interest**
None declared.

## P377

### Scurvy mimicking autoimmune diseases in pediatric patients with neurodevelopmental disorder

Y. Stepaniuk[1,2,*], O. Oshlianska[2]
[1]National Specialized Children's Hospital «Okhmatdyt» of the Ministry of the Health of Ukraine, [2]Shupyk National Healthcare University of Ukraine, Kyiv, Ukraine
**Correspondence:** Y. Stepaniuk
*Pediatric Rheumatology 2026*, **24(S1):**P377

**Introduction**
Nowadays, scurvy remains clinically relevant in specific patient groups and may closely mimic autoimmune diseases in clinical practice. Due to its heterogeneous presentation, including musculoskeletal manifestations (arthralgia, hemarthrosis, muscle pain, gait disturbances) and mucocutaneous involvement, scurvy may resemble juvenile idiopathic arthritis (JIA), chronic recurrent multifocal osteomyelitis (CRMO), systemic lupus erythematosus (SLE), dermatomyositis, and other rheumatologic conditions. Although uncommon in high-income countries, vitamin C deficiency continues to occur in children with neurodevelopmental disorders, restrictive eating behavior, psychiatric conditions or malabsorption syndromes.
**Objectives**
To highlight diagnostic challenges of pediatric scurvy initially misinterpreted as autoimmune disease.
**Methods**
Retrospective case series of two children diagnosed with scurvy based on clinical, laboratory, and imaging findings.
**Case 1**
A 5-year-old girl with autism spectrum disorder (ASD) developed acute hip pain and limping, initially interpreted as reactive arthritis. Over months, she developed progressive soft tissue swelling, severe pain, inability to walk, and subfebrile fever. Laboratory tests showed elevated ESR, CRP, anemia, thrombocytosis, increased creatine kinase and lactate dehydrogenase, as well as deficiencies of vitamin D, folate, and vitamin B12. Differential diagnosis included dermatomyositis, thrombosis, neurofibromatosis, and malignancy. Myositis profile was negative, and muscle biopsy was nonspecific. MRI and CT revealed extensive bilateral subperiosteal hematomas of the femora and tibiae. Coagulopathies were excluded, and genetic testing for neurofibromatosis was negative. Given severe food selectivity, vitamin C deficiency was suspected and confirmed (<0.4 mg/L).
**Case 2**
A 7-year-old boy with severe ASD presented with purpuric rash of the lower extremities, followed by progressive musculoskeletal pain, morning stiffness, and refusal to walk. Laboratory tests showed mildly elevated ESR and CRP, anemia and multiple deficiencies, including iron, vitamin D, folate, and vitamin B12. Rheumatologic evaluation excluded vasculitis, SLE, JIA and inflammatory myopathies. MRI showed multifocal bone marrow edema of the pelvis, femora, sacroiliac joints, and spine, suggestive of inflammatory bone disease. CRMO was considered. However, a detailed dietary history revealed restrictive eating and severe vitamin C deficiency (<0.4 mg/L) was confirmed. In both cases, rapid improvement occured after vitamin C supplementation, with pain reduction and restored ambulation. These cases highlight that scurvy can mimic autoimmune disease clinically and radiologically, leading to delayed diagnosis and extensive unnecessary investigations. Awareness of nutritional risk factors and early consideration of vitamin C deficiency are essential for timely diagnosis and treatment.
**Disclosure of interest**
None declared.

## P378

### Safety profile of long-term antibiotic prophylaxis in patients with rheumatic fever: a population-based study

S. Handelsman[1], G. Amarilyo[2], L. Goldberg[1], Y. Levinsky[2,*]
[1]Pediatric B; [2]Pediatric rheumatology, Schneider children's medical center of Israel, Petah Tikva, Israel
**Correspondence:** Y. Levinsky
*Pediatric Rheumatology 2026*, **24(S1):**P378

**Introduction**
Rheumatic fever (RF) is a severe inflammatory condition that develops as a delayed immune response to untreated streptococcal pharyngitis. To prevent recurrent disease and long-term valvular heart damage, prolonged antibiotic prophylaxis is routinely administered. While this strategy has been shown to effectively prevent RF recurrences and chronic cardiac complications, data regarding the long-term adverse effects of extended antibiotic exposure are limited. Concerns have been raised regarding a potential increased risk of autoimmune diseases, allergic conditions, and antibiotic resistance.
**Objectives**
To evaluate the prevalence of adverse events associated with long-term antibiotic prophylaxis in patients with rheumatic fever.
**Methods**
Using the database of Clalit Health Services, the largest healthcare provider in Israel covering more than 4.5 million individuals, we conducted a retrospective cohort study of patients diagnosed with RF. Adverse events were identified and analyzed. Treatment adherence was assessed using the mean possession ratio (MPR), with MPR >80% defined as high adherence. Inclusion required more than one year of antibiotic exposure. Three groups were constructed: RF patients with high adherence, RF patients with low adherence, and a healthy control group. Univariate and multivariable analyses were performed to compare the incidence of predefined outcomes between groups.
**Results**
A total of 4,886 individuals were included: 1,277 in the high-MPR RF group, 1,633 in the low-MPR RF group, and 1,976 healthy controls. Compared with healthy controls, RF patients with high adherence did not demonstrate an increased risk of autoimmune diseases, including immune thrombocytopenic purpura (ITP), juvenile idiopathic arthritis (JIA), inflammatory bowel disease (IBD), systemic lupus erythematosus (SLE), or autoimmune thyroiditis. Regarding allergic conditions, the high-MPR group exhibited a higher prevalence of asthma [232 (18.2%) vs. 307 (15.5%), $P$=0.049] and food allergy [86 (6.7%) vs. 86 (4.4%), $P$=0.003]. No significant differences were observed in rates of drug allergy, anaphylaxis, or atopic dermatitis. Notably, when comparing RF patients with high versus low adherence, these differences were not observed.
**Conclusion**
Overall, long-term antibiotic prophylaxis for rheumatic fever appears safe with respect to the development of autoimmune diseases and allergic conditions. These results indicate the safety of the treatment and support current recommendations for prolonged antibiotic prophylaxis in RF. Given the generally low adherence to prophylactic treatment, the findings of this study may help improve patient and caregiver confidence and enhance long-term adherence.
**Disclosure of interest**
None declared.

## P379

### Describing central capabilities in caregivers of children with rheumatic diseases

F. A. Vasquez Triminio[1,*], Z. I. García Murillo[1], N. E. Rubio Perez[1], F. García Rodriguez[1], A. V. Villareal Treviño[1], I. P. Ballestas[2], A. M. De Lara[2], R. A. Falcón Zúñiga[1], C. E. López Cardona[1]
[1]Pediatria, Hospital Universitario, Nuevo Leon; [2]Rheumatology, Rheumatology Unit, Hospital General de México "Dr. Eduardo Liceaga", Ciudad Mexico, Mexico
**Correspondence:** F. A. Vasquez Triminio
*Pediatric Rheumatology 2026*, **24(S1):**P379

**Introduction**
Adequate control of pediatric rheumatic diseases largely depends on the primary caregiver, who assumes a central role in treatment administration, symptom monitoring, clinical decision-making, and coordination of medical follow-up. Human well-being has been conceptualized from multiple perspectives. The human capabilities approach proposed by Martha Nussbaum provides a robust theoretical framework that understands well-being as the real opportunity to achieve essential functions for a dignified life, beyond the mere possession of goods or subjective adaptation to adverse conditions. According to Nussbaum, a just society must guarantee a minimum threshold of ten central capabilities that enable individuals to live with dignity. These capabilities are not internal states, but real opportunities for action and choice, shaped by personal, social, economic, and cultural factors.
**Objectives**
To analyze the impact on Nussbaum's central capabilities in primary caregivers of children with rheumatic diseases.
**Methods**
A qualitative study with an interpretative approach was conducted. In-depth, semi-structured individual interviews were designed based on Nussbaum's ten central capabilities, adapted to the context of pediatric rheumatologic care. Primary caregivers of pediatric patients diagnosed with rheumatic diseases (juvenile idiopathic arthritis, systemic lupus erythematosus, and juvenile dermatomyositis), aged 18 to 80 years, attending the Pediatric Rheumatology Clinic at the University Hospital "Dr. José Eleuterio González," were included.
**Results**
A significant impact on caregivers' central capabilities was identified. The most affected domains were emotional well-being, social affiliation, and control over the environment, manifested as anxiety, social isolation, and difficulties maintaining employment and financial stability. Physical health was also affected, with reports of fatigue, sleep disturbances, and reduced self-care. A higher caregiving burden was associated with broader and more severe restrictions across multiple capabilities, particularly among those with limited support networks and greater time dedicated to caregiving.
**Conclusion**
The findings show that the role of primary caregiver of children with rheumatic diseases is associated with significant limitations in multiple central capabilities, particularly in emotional well-being, social interaction, autonomy, and control over the environment. These results highlight the importance of implementing a comprehensive care approach centered on the child–caregiver dyad.

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**Disclosure of interest**
None declared.

## P380

### Physical fitness and disease burden in juvenile fibromyalgia: toward objective tools for personalized exercise therapy

A. B. Ronchetti[1,*], C. Gattorno[2], C. Ferrari[2], M. Sormani[3], P. Cardellicchio[2], C. Tacchino[2], A. Aiello[2], M. Bertamino[2], A. Calcagno[2], L. Doglio[2], M. Mordeglia[4], M. Mazzoni[5], N. Maronese[5], C. Ferrari[2], M. Mino Marotta[6], M. Ayoubi Khajekini[7], C. Paolo[8], C. Proietti Ciolli[9], B. Ceriana[1], S. Cencioni[1], M. Gattorno[5], C. Malattia[1,5]
[1]Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genova; [2]Physical Medicine and Rehabilitation Unit, IRCCS Istituto Giannina Gaslini; [3]Department of Health Sciences, University of Genova; [4]University of Genova, Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI),, [5]UOC Reumatologia e Malattie Autoinfiammatorie, IRCCS Istituto Giannina Gaslini, Genova; [6]Physical medicine and rehabilitation section, University of Foggia, Foggia; [7]Department of Surgical Sciences, University of Turin; [8]Department of Surgical Sciences, University of Torino, Torino; [9]Pediatrics, University of Ferrara, Ferrara, Italy
**Correspondence:** A. B. Ronchetti
*Pediatric Rheumatology 2026*, **24(S1):**P380

**Introduction**
Juvenile fibromyalgia (JFM) is a chronic condition associated with significant functional impairment and reduced quality of life. Exercise is key in treatment; however, objective tools for guidance and monitoring are lacking.
**Objectives**
To identify performance-based physical fitness tests that best reflect overall fitness and disease burden in JFM.
**Methods**
Physical fitness was assessed using the 6-Minute Walk Test (6MWT), Standing Broad Jump, 30-Second Sit-to-Stand Test (STS), Shuttle Run, Flamingo Balance Test, 30-Second Walking Test (30sWT), and Medicine Ball Throw. Results were compared with age- and sex-matched healthy controls. Associations between fitness measures and disease severity (pain intensity, fatigue [PedsQL], patient global assessment [PGA], physician global assessment [PhGA], Functional Disability Inventory [FDI], and WeeFIM) were evaluated using correlation analyses and multivariate linear regression.
**Results**
44 JFM patients (39 females; mean age 16.4 years) were included. Compared with controls, patients showed significantly reduced performance across multiple fitness domains. Shuttle Run demonstrated the strongest correlations with disease burden. WeeFIM ($r = -0.70$, $p < 0.001$), FDI ($r = 0.50$, $p = 0.002$), pain ($r = 0.42$, $p = 0.005$), fatigue ($r = -0.42$, $p = 0.006$), PhGA ($r = 0.43$, $p = 0.005$), and PGA ($r = 0.40$, $p = 0.01$). The 6MWT correlated with WeeFIM ($r = -0.51$, $p < 0.001$), FDI ($r = -0.40$, $p = 0.002$), and PGA ($r = -0.40$, $p = 0.03$), while Standing Broad Jump was associated with fatigue ($r = -0.53$, $p < 0.001$), FDI ($r = -0.51$, $p = 0.001$), PhGA ($r = -0.43$, $p = 0.01$), and PGA ($r = -0.40$, $p = 0.01$). In multivariate analyses, Shuttle Run remained independently associated with pain intensity and PhGA ($\beta = 0.20$, SE = 0.08, $p = 0.02$;$\beta = 0.16$, SE = 0.08, $p = 0.04$, respectively).
**Conclusion**
JFM exhibit marked impairment in physical fitness compared with healthy peers. Shuttle Run, 6MWT, and Standing Broad Jump showed the strongest associations with disease severity, functional disability, and reduced autonomy. Notably, Shuttle Run independently correlated with PGA and PhGA disease severity.These findings support the use of standardized fitness assessments to better characterize patient status and guide personalized exercise-based interventions. Longitudinal studies are needed to assess adherence and treatment response.

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**Disclosure of interest**
None declared.

## P381

**Psychological state of children with rheumatic diseases in Ukraine**
O. A. Oshlianska[1,2,*], K. A. Yats [2], E. V. Zbrozhyk[2], V. Lunov[3]
[1]Department of Pediatrics, Neonatology, Childhood Infectious Diseases, Immunology and Allergology, Platon L. Shupyk National Healthcare University of Ukraine; [2]Pediatric, State Institution State Institution "Ukrainian Center of Maternity and Childhood of the National Academy of Medical Sciences of Ukraine"; [3]Department of General and Medical Psychology, Bogomolets National Medical University, Kyiv, Ukraine
**Correspondence:** O. A. Oshlianska
*Pediatric Rheumatology 2026*, **24(S1):**P381

**Introduction**
Stress potentially affects disease activity, the frequency of relapses of rheumatic diseases (RD), and the scope of required treatment.
**Objectives**
To assess the impact of individual war-related stressors on the severity of anxiety in pediatric patients with RD in Ukraine.
**Methods**
Ninety patients with RD (64 JIA, 5 SLE, 2 jSSc, 7 JDM, 12 — other) and their parents were interviewed and assessed by a psychologist. The mean disease duration was 4.02 years (range 0–16). Forty-two patients were assigned to a high-stressor-exposure subgroup on the basis of one or more of the following criteria: residence in frontline regions ($n$=7); a parent currently serving in the Armed Forces ($n$=38; 4 of whom had been killed in action); or close relatives remaining in areas of active hostilities ($n$=13). A comparative analysis was performed between objective data from medical records on the course of RD, parental subjective assessments of the child's health status, and the results of psychological testing.
**Results**
Of the surveyed parents, 11% believed that the war had contributed to the onset or exacerbation of the disease in their child; 17% reported increased difficulty in accessing medical facilities, and 35% reported difficulties in obtaining medications. According to unified disease-activity scales, high disease activity was registered in 5 patients, moderate — in 11, and minimal — in 28; 46 patients were examined during clinical remission. The mean CHAQ score was 0.92±0.8 and did not correlate with parental perception of deterioration ($r$=0.17). The mean anxiety severity on the Beck Anxiety Inventory (BAI) in children with RD was 11.05±2.3, corresponding to a moderate level and exceeding that of the historical control cohort (analogous assessment in 2000–2004: 7.8±3.6). The highest score was recorded in patients from frontline regions (13.57±1.9). The proportion of patients with moderate-to-high anxiety in the high-stressor subgroup reached 20%, compared with 12.49% in patients without additional stressors. Spielberger–Khanin Inventory scores (40.62±5.1) did not differ significantly between subgroups; nevertheless, the high-stressor subgroup demonstrated a greater proportion of elevated values, predominantly attributable to items reflecting trait anxiety. The SCARED screening revealed that 9 (21.4%) patients with high stressor exposure were at high risk of emotional disorders (total score >30), compared with 4 (8.3%) in the comparison subgroup; differentiation by subscale revealed a predominance of generalized persistent anxiety. Beck Depression Inventory (BDI) scores averaged 8.95±6.1; signs of depression of varying severity were identified in 16.5% of the lower-stressor subgroup and in 24% of the high-stressor subgroup.
**Conclusion**
Parental anxiety regarding the child's health was associated with the level of anxiety in the child. Anxiety in children with RD is significantly elevated — to a greater extent in the presence of additional stressors — and does not correlate with the objective clinical assessment of disease status. There is a critical need for routine mental-health screening and individualized psychological support for children with RD in Ukraine.

**References**

1. To the resilient little patients and their parents.

**Disclosure of interest**
None declared.

## P382

**What really matters to parents of children with juvenile idiopathic arthritis: pilot online survey of parents from Russia and CIS countries**
O. Loskutova (Konopelko)[1,*], N. Tsurikova[2], E. Zholobova[3]
[1]Fantasy Clinic, Moscow; [2]PLUS Clinic, Rostov-on-Don; [3]Sechenov University, Moscow, Russian Federation
**Correspondence:** O. Loskutova (Konopelko)
*Pediatric Rheumatology 2026*, **24(S1):**P382

**Introduction**
Adherence to therapy in juvenile idiopathic arthritis (JIA) is influenced not only by drugs but also by parental understanding, communication with clinicians and psychosocial support. Existing data show low adherence and high emotional burden among parents, while information on their specific needs in Eastern Europe and CIS countries remains limited.

**Objectives**
To conduct a pilot assessment of informational needs, psychosocial requirements, and use of psychological support and complementary and alternative medicine (CAM) among parents of children with confirmed JIA in Russia and CIS countries.
**Methods**
We conducted a cross-sectional anonymous online survey of parents of children with a confirmed JIA diagnosis under the care of a paediatric rheumatologist. Participants were recruited on 10–11 November 2019 via invitations posted on the professional social media accounts of a single paediatric rheumatologist (convenience sample). In total, 38 completed questionnaires from respondents in 18 cities were analysed. The questionnaire comprised 10 items on the diagnostic journey, time to acceptance of the disease, sources of information, awareness of patient organizations, experience with psychological counselling, use of CAM, participation in patient education programmes, expectations from rheumatology services and current psychological state. Clinical characteristics of the children (JIA subtype, disease duration, disease activity and antirheumatic treatment) were not collected.
**Results**
The majority of respondents (73.7%; 95% CI 58.0–85.0; *n*=28) reported that they had accepted the diagnosis; the mean time to acceptance was 10.2 ± 8.5 months from diagnosis. In 26.3% (95% CI 15.0–42.0; *n*=10), acceptance had not occurred. Psychological support had been sought by 34.2% (95% CI 21.2–50.1; *n*=13); among them, 5.3% (95% CI 1.5–17.3; *n*=2) reported a negative experience. CAM was used by 26.3% (95% CI 15.0–42.0; *n*=10); an additional 2.6% (*n*=1) had not yet used CAM but expressed interest, whereas 15.8% (95% CI 7.4–30.4; *n*=6) were strongly opposed. Osteopathy was most frequently mentioned (80% of CAM users), followed by homeopathy, acupuncture, phytotherapy and other non–evidence-based modalities. Overall, 36.8% (95% CI 23.4–52.7; *n*=14) rated their psychological state as stable or positive, 34.2% (95% CI 21.2–50.1; *n*=13) as unstable, and 28.9% were unable to provide a clear self-assessment.
**Conclusion**
In this pilot survey (*n*=38), one in four parents reported using CAM, one in three had sought psychological support and the mean time to acceptance of the diagnosis exceeded 10 months. These findings indicate a substantial unmet need for structured psychosocial support and support the development of paediatric rheumatology education focused on everyday-life concerns and early psychosocial care.
**Disclosure of interest**
None declared.

## P383

### Beyond the patient: psychosocial wellbeing and quality of life in caregivers of children with pediatric rheumatic diseases

K. Tansuhaj[1,*], S. Kanchanasapha[1], S. Wongjitjanyong[1], S. Swatesuthisan[1], N. Siripensopa[1], T. Lerthattasilp[1], S. Nanthapisal[1]
[1]Faculty of Medicine, Thammasat University, Pathumthani, Thailand
**Correspondence:** K. Tansuhaj
*Pediatric Rheumatology 2026*, **24(S1):**P383

**Introduction**
Paediatric rheumatic diseases (PRDs) are chronic, multisystem conditions that substantially impair the physical functioning and wellbeing of affected children. In managing these conditions, caregivers assume considerable responsibility, navigating complex therapeutic regimens, frequent clinical follow-up, and child resistance to treatment. Such demands place caregiver psychosocial wellbeing at risk, with potential consequences for treatment adherence and patient outcomes. Nevertheless, caregiver-centred psychosocial wellbeing in paediatric rheumatology remains largely unexplored.
**Objectives**
To evaluate mental health status and health-related quality of life (HRQoL) among caregivers of children with PRDs, and identify independent determinants of poorer psychosocial outcomes.
**Methods**
A cross-sectional study was conducted via structured telephone interviews with caregivers of paediatric patients with PRDs receiving care at Thammasat University Hospital, Thailand over a 10-year period. Caregivers' Demographic characteristics, HRQoL (WHOQOL-BREF-THAI), and mental health status (Depression Anxiety Stress Scale-21; DASS-21) were assessed. Univariate and multivariable linear regression analyses were performed to identify factors independently associated with QoL and depression scores.
**Results**
A total of 105 patient–caregiver dyads were enrolled. The majority of patients had inflammatory rheumatic diseases, predominantly systemic lupus erythematosus and juvenile idiopathic arthritis. Caregivers had a mean age of 46.1 ± 8.9 years; most were mothers (77.1%), female (88.6%), married (84.8%), and employed (80.0%). Despite only 22.9% of patients having active disease, caregivers reported high perceived disease severity (median visual analog scale 8.0, IQR 6.0–9.0). Overall caregivers' QoL was moderate-to-good; however, the social relationships domain was most adversely affected, with 10.5% of caregivers reporting poor QoL in this domain. Normal depression scores were observed in 76.2% of caregivers, whereas above-threshold anxiety and stress symptoms were present in 41.0% and 29.5%, respectively. On multivariable analysis, lower perceived income sufficiency (□□ = −6.30, 95% CI −8.92 to −3.68, $p < 0.001$) and greater follow-up adherence burden (□□ = −6.17, 95% CI −10.86 to −1.48, $p = 0.010$) were independently associated with lower overall QoL. Younger caregiver age was independently associated with higher depression scores (□□ = −0.027, 95% CI −0.053 to −0.000, $p = 0.049$).
**Conclusion**
Although caregivers of children with PRDs demonstrated broadly moderate-to-good overall QoL, significant psychosocial vulnerability persisted, particularly with respect to anxiety, stress, and social relationships. Paediatric rheumatology rehabilitation should extend beyond disease control to encompass systematic caregiver-centred psychosocial screening and targeted support, with priority given to younger caregivers, those with lower perceived income sufficiency, and those experiencing follow-up adherence burden. Caregiver psychosocial needs warrant integration into long-term, family-centred paediatric rheumatology care.
**Disclosure of interest**
None declared.

## P386

### Description of central capabilities in children with rheumatologic diseases

Z. I. García Murillo[1,*], F. A. V. Triminio[1], F. G. Rodríguez[1], A. V. V. Treviño[1], N. E. R. Pérez[1], I. P. Ballestas[2], A. M. De Lara[2], R. A. F. Zúñiga[1], C. E. L. Cardona[1]
[1]Pediatría, Hospital Universitario "Dr. José Eleuterio González", Monterrey, Nuevo León; [2]Reumatología, Hospital General de México "Dr. Eduardo Liceaga", Ciudad de México, Mexico
**Correspondence:** Z. I. García Murillo
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**Introduction**
Pediatric rheumatologic diseases affect not only physical health, but also the emotional, social, and moral development of children and adolescents. Martha Nussbaum's central capabilities approach provides a framework to analyze how chronic diseases modify patients' real opportunities to develop and exercise essential human functions.
**Objectives**
To analyze the impact of pediatric rheumatologic diseases on Martha Nussbaum's central capabilities in children and adolescents.
**Methods**
An exploratory–interpretative qualitative study was conducted in patients aged 6 to 16 years with juvenile idiopathic arthritis, systemic lupus erythematosus, juvenile dermatomyositis, juvenile scleroderma, and systemic vasculitis, treated at a referral center in northeastern Mexico. The first phase consisted of semi-structured interviews aimed at exploring experiences related to bodily health, emotions, affiliation, play, autonomy, and participation in decision-making. Interviews were audio-recorded, transcribed verbatim, and analyzed using thematic content analysis. The second phase employed Q methodology to identify shared patterns of subjectivity regarding well-being, dignity, autonomy, and caregiving relationships.

**Results**
Significant impairments were identified in capabilities related to bodily health, play, emotions, affiliation, and control over one's environment. Participants described physical limitations, school absenteeism, reduced recreational activities, anxiety, frustration, and experiences of social exclusion or overprotection. Furthermore, the impact on capabilities was found to depend not only on disease activity, but also on familial, educational, social, and structural factors. Some patients described strengthened capabilities related to resilience, reflection about the future, and participation in decisions regarding their care.
**Conclusion**
This study provides a bioethical and patient-centered perspective on the experience of living with pediatric rheumatologic diseases, promoting models of care that are more sensitive to the progressive autonomy, dignity, and comprehensive well-being of children and adolescents.

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**Disclosure of interest**
None declared.

P387
**Exercise studies in children and adolescents with juvenile idiopathic arthritis: a bibliometric analysis**
Z. Tatar[1,*], E. Gür Kabul[2], B. Başakcı Çalık[3]
[1]Home Care Services Program, Department of Health Care Services, Şırnak University, Şırnak; [2]Department of Physiotherapy and Rehabilitation, Faculty of Health Sciences, Uşak University, Uşak; [3]Faculty of Physiotherapy and Rehabilitation, Pamukkale Üniversity, Denizli, Türkiye
**Correspondence:** Z. Tatar
*Pediatric Rheumatology 2026*, **24(S1):**P387

**Introduction**
Exercise in children with Juvenile Idiopathic Arthritis (JIA) facilitates daily living activities by increasing muscle strength, bone health, and exercise capacity; it improves quality of life and reduces long-term health risks (1). A bibliometric analysis of the exercise field in JIA can identify research trends, effective studies, and knowledge gaps, thereby guiding future research and clinical practice.
**Objectives**
The aim of this study is to examine research on exercise in JIA using bibliometric analysis methods.
**Methods**
The data for this study consisted of articles published in the Web of Science database between 1992 and 2025. Bibliometric analysis was used to investigate the productivity, publication performance, high-impact journals and articles, country-specific productivity, and trending topics in exercise research among children and adolescents with JIA.
**Results**
A total of 135 studies were included in the analyses. An examination of the Web of Science index records for these studies revealed that 128 were indexed in the Science Citation Index Expanded, 17 in the Social Sciences Citation Index, 5 in the Emerging Sources Citation Index, and 28 in the Conference Proceedings Citation Index-Science. Publication productivity was found to fluctuate over the years. The highest publication productivity was in 2018, with 15 publications. The country with the highest publication productivity was the United States, with 30 publications. "Arthritis and Rheumatism" was the journal with the highest publication productivity, with 12 publications. The trending topic was "physical activity." Regarding author productivity, authors with one publication in the field accounted for 78% of all authors, those with two publications accounted for 10%, and those with three publications accounted for 4%. The study with the most global citations was the work by van Brussel et al. (2011) (2).
**Conclusion**
Scientific output in exercise research involving children and adolescents with JIA was found to be relatively low and to fluctuate. However, the growing emphasis on physical activity trends may indicate that exercise therapy is not limited to structured programs implemented in clinical settings, but that approaches aimed at increasing overall physical activity levels in daily life are also gaining importance.

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**Disclosure of interest**
None declared.

P388
**Paired-pulse TMS reveals a distinct pattern of intracortical modulation in juvenile fibromyalgia: a pilot study**
P. Cardellicchio [1], A. B. Ronchetti[2,*], C. Tacchino [1], M. Mazzoni[3], N. Maronese[3], C. Proietti Ciolli[4], B. Ceriana[2], A. Aiello[1], M. Bertamina[1], A. Calcagno[1], L. Doglio[1], M. Severina[5], M. Mordeglia[2], M. Pavanello[6], L. Nobili [2,7], A. Ravelli[2,8], M. Gattorno[3], C. Malattia[2,3]
[1]Physical Medicine and Rehabilitation Unit, IRCCS Istituto Giannina Gaslini; [2]Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genova; [3]UOC Reumatologia e Malattie Autoinfiammatorie, IRCCS Istituto Giannina Gaslini, Genova; [4]Pediatrics, University of Ferrara, Ferrara; [5]Neuroradiology Unit,; [6]Neurosurgery Unit; [7]Neuropsychiatry Unit; [8]Direzione Scientifica, IRCCS Istituto Giannina Gaslini, Genova, Italy
**Correspondence:** A. B. Ronchetti
*Pediatric Rheumatology 2026*, **24(S1):**P388

**Introduction**
Juvenile fibromyalgia syndrome (JFS) is a heterogeneous condition characterized by widespread musculoskeletal pain, fatigue, non-refreshed sleep, and mood disturbances. Impairment in physical, social, and emotional functioning is common and affects the patient's ability to perform the activities of daily living. Transcranial magnetic stimulation (TMS) has emerged as a potential neurophysiological biomarker in adult fibromyalgia (FM), revealing reduced intracortical inhibition and increased motor thresholds, which correlate with pain, fatigue, and cognitive dysfunction.
**Objectives**
To investigate intracortical inhibitory and facilitatory circuits in adolescents with primary juvenile fibromyalgia syndrome (JFS) using paired-pulse TMS, and to compare these findings with age-matched healthy controls.
**Methods**
Fifteen adolescents with primary JFS (12 females; median age at diagnosis: 13.6 years; median disease duration: 2.3 years) were enrolled. Exclusion criteria included the use of centrally acting medications and contraindications to TMS. Paired-pulse TMS paradigms were used to assess short-interval intracortical inhibition (sICI; GABA-A–mediated), long-interval intracortical inhibition (LICI; GABA-B–mediated), and intracortical facilitation (ICF; glutamatergic-mediated). Single-pulse TMS was used to evaluate corticospinal excitability and cortical silent period (cSP). Group comparisons between JFS patients and healthy controls were performed using unpaired t-tests with Welch's correction ($p < 0.05$).
**Results**
No significant differences were observed between groups in resting motor threshold or cSP duration. In contrast, JFS patients showed significantly reduced sICI ratios ($p = 0.04$) and significantly increased log10-transformed LICI ($p = 0.01$) and ICF ($p = 0.006$) compared with controls.
**Conclusion**
This is the first study to demonstrate measurable abnormalities in intracortical excitatory and inhibitory circuits in adolescents with JFS using paired-pulse TMS. The observed pattern suggests reduced inhibitory

and enhanced facilitatory intracortical drive. These findings support the potential of TMS-derived metrics as objective biomarkers for JFS and provide a rationale for the development of age-specific neuromodulatory interventions.

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**Disclosure of interest**
None declared.

## P389

**Current evidence on non-pharmacological interventions in juvenile fibromyalgia**


A. Gkouvi[1], A. Gkoutzourelas[1,*], E. C. Pardali[1], D. P. Bogdanos[1], M. G. Grammatikopoulou[1]
[1]Department of Rheumatology and Clinical Immunology, University General Hospital of Larissa, Faculty of Medicine, School of Health Sciences, University of Thessaly, Larissa, Greece
**Correspondence:** A. Gkoutzourelas

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**Introduction**
Juvenile fibromyalgia (JFM) is a poorly understood chronic condition characterized by diffuse musculoskeletal pain, fatigue, sleep disturbance and mood disorders. Management remains challenging due to the disease's multifactorial nature and the limited availability of pediatric evidence.
**Objectives**
The purpose of this review is to summarize current evidence on non-pharmacological treatments for JFM.
**Methods**
Three databases (PubMed, Cochrane, clinicaltrials.gov) were systematically searched by two independent reviewers. The search identified 229 records. After duplicate removal, followed by title and abstract screening and full text assessment, five studies were included.
**Results**
Cognitive behavioral therapy (CBT) was the most frequently studied intervention. It improved functional disability and depressive symptoms compared with education-based interventions, although its effect on pain seems limited. The Fibromyalgia Integrative Training for Teens programme (FIT), that combines CBT with structured neuromuscular exercise, showed greater reductions in pain compared to CBT alone. When biomechanical outcomes were analyzed, FIT improved hip abduction strength and external hip rotation, suggesting better stability during functional movements. In a larger multicenter trial, FIT was not superior to CBT or graded aerobic exercise (GAE), however all treatments reduced pain and disability. Approximately 1 in 4 patients had noteworthy improvement and pain intensity reduced gradually, while results were maintained after 12 months.
**Conclusion**
Overall, non-pharmacological interventions can improve pain and functioning in patients with JFM, particularly CBT and structured exercise. CBT consistently reduced disability but effects on pain were more limited. Integrated programs including CBT and neuromuscular approaches are feasible and may reduce pain and enhance functional movement outcomes. Treatment should be individualized according to symptoms, functional limitations, exercise tolerance and patient preferences. Future research should compare these interventions with pharmacological treatment and examine whether combining behavioral, exercise-based and medication approaches lead to further improvements.

**Disclosure of interest**
None declared.

## P390

**Association of medication adherence with illness perception and quality of life in pediatric Familial Mediterranean Fever: preliminary study**


A. Doğru Kılınç[1,*], B. Başer Taşkın[1], Ö. Akgün[1], N. Aktay Ayaz[1]
[1]Pediatric Rheumatology, Istanbul University Faculty of Medicine, Istanbul, Türkiye
**Correspondence:** A. Doğru Kılınç

*Pediatric Rheumatology 2026*, **24(S1):**P390

**Introduction**
Familial Mediterranean fever (FMF), a childhood autoinflammatory disease, requires long-term treatment adherence to prevent recurrent attacks, persistent inflammation, and disease-related complications. Psychosocial factors such as disease perception and health-related quality of life can affect medication adherence and thus influence disease outcomes in childhood FMF patients.
**Objectives**
This study aimed to investigate whether illness perception and health-related quality of life are associated with medication adherence in pediatric FMF patients.
**Methods**
This cross-sectional study included pediatric patients aged 8–18 years diagnosed with FMF. Medication adherence, illness perception, and health-related quality of life were assessed using the Medication Adherence Scale in FMF (MASIF), the Brief Illness Perception Questionnaire (B-IPQ), and the Pediatric Quality of Life Inventory (PedsQL), respectively, based on patient self-report. Patients were classified as having poor adherence (MASIF <60) or good adherence (MASIF ≥60) according to MASIF scores.
**Results**
A total of 81 pediatric patients with FMF were classified into poor adherence (Group 1, $n$=35) and good adherence (Group 2, $n$=46) groups. Demographic and disease-related characteristics, including clinical findings, annual attack frequency, biologic agent use, and disease activity scores, were similar between the groups (all $p$>0.05). Regarding the PedsQL, Group 2 demonstrated higher overall total scores (OTS) and physical health total scores (PHTS) ($p$ = 0.012 and $p$ < 0.001, respectively). In the B-IPQ evaluation, Group 1 had higher scores on the consequences and emotional response dimensions (both $p$ < 0.001), whereas Group 2 had higher scores on treatment control ($p$ = 0.017). Correlation analysis demonstrated positive correlations between MASIF total scores and PedsQL OTS (rho=0.415, $p$<0.001), PHTS (rho=0.484, $p$<0.001), emotional health total score (rho=0.319, $p$=0.004), and psychosocial health total score (rho=0.294, $p$=0.008). Regarding B-IPQ subscales, MASIF total scores demonstrated negative correlations with consequences (rho = −0.376, $p$ < 0.001), concern (rho = −0.261, $p$ = 0.018), and emotional response (rho = −0.243, $p$ = 0.029), while positive correlations were observed with personal control (rho=0.288, $p$=0.009) and treatment control (rho=0.406, $p$<0.001).
**Conclusion**
Good medication adherence was associated with higher quality of life, more favorable illness perception, and stronger treatment control perception in pediatric FMF patients, highlighting the importance of psychosocial factors in disease management.
**Disclosure of interest**
None declared.

## P391

**Perspectives of patients and caregivers on disease activity assessment in pediatric Familial Mediterranean Fever**


B. Küçükali[1,*], F. Açıkgöz[2], Ç. Yıldız[1], B. Acun[1], N. Belder[1], M. Kutlar[1], K. Ahmadova[1], Ö. Özenli Yağcı[1], K. Deveci Şeker[1], D. Gezgin Yıldırım[1], S. A. Bakkaloğlu[1]
[1]Pediatric Rheumatology; [2]Faculty of Medicine, Gazi University, Ankara, Türkiye
**Correspondence:** Batuhan Küçükali

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**Introduction**
Composite disease activity instruments incorporating patient-reported outcomes are well established across rheumatology and are central to treat-to-target strategies. Yet, FMF remains a notable exception, where the only validated disease activity tool, AIDAI, primarily reflects symptom presence and duration and does not incorporate patients' perception of symptom severity or functional impact. This limitation may lead to incomplete assessment of disease burden and potential underestimation of clinically relevant activity.
**Objectives**
To investigate patients' and caregivers' perspectives on disease activity in pediatric FMF and to explore the relative importance of symptom severity, duration, and functional impact.
**Methods**
In this cross-sectional pilot study, 95 FMF patients and caregivers were interviewed using a semi-structured questionnaire between March–September 2025. Symptom burden was assessed using Likert scales and a weighted burden score. A priority index (PI) identified symptoms requiring immediate treatment. Ordinal logistic regression explored factors associated with perceived burden. Test-retest reliability was assessed in 20 patients using intraclass correlation coefficient (ICC).
**Results**
Abdominal pain and fatigue showed highest perceived burden, whereas fever was most frequently prioritized for immediate treatment (PI 0.42). Fatigue was rarely prioritized (PI 0.02). Most respondents (87.4%) considered symptom severity at least as important as duration in disease activity; 84% emphasized functional impact, inability to perform daily activities. Physical activity avoidance was reported by 35.8%. Longer follow-up was associated with lower fever burden (OR 0.98; $p$=0.026), while absence of recent attack was associated with lower abdominal pain burden (OR 0.25; $p$=0.028). The questionnaire demonstrated excellent reliability (ICC=0.87).
**Conclusion**
FMF disease activity is perceived as multidimensional, extending beyond symptom duration to include severity and functional impact not captured by current tools such as AIDAI. Fever phobia and fatigue normalization represent actionable targets, and until validated composite tools become available, clinicians should routinely inquire about school absenteeism and activity restriction while providing targeted patient education. Incorporation of patient-reported outcomes into composite disease activity measures may enable more comprehensive, patient-centered treat-to-target management.

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**Disclosure of interest**
None declared.

## P394

### Validity and reliability of the international fitness scale and the self-perceived health-related physical fitness questionnaire in children and adolescents with juvenile idiopathic arthritis and familial Mediterranean fever

B. N. Yumak[1,2,*], A. Yekdaneh[3], A. Albayrak[1,4], A. N. Şeker[1,5], I. Donmez[6], A. Yılmaz[7], F. G. Demirkan[8], F. Çakmak[9], Ö. Akgün[10], N. Aktay Ayaz[10], N. Arman[11]
[1]Institute of Graduate Studies, Physiotherapy and Rehabilitation Doctorate Program, Istanbul University-Cerrahpasa; [2]Faculty of Health Sciences, Physiotherapy and Rehabilitation (English) Program, Istanbul Atlas University; [3]Vocational School of Health Services, Physiotherapy English Program, Fenerbahce University; [4]Faculty of Health Sciences, Physiotherapy and Rehabilitation Program, Istanbul Kent University; [5]Vocational School of Health Services, Physiotherapy Program, Bahcesehir University; [6]Physiotherapist, Private Health Services Unit; [7]Institute of Graduate Studies, Physiotherapy and Rehabilitation Master Program, Istanbul University-Cerrahpasa; [8]Department of Pediatric Rheumatolog, Kanuni Sultan Suleyman Training and Research Hospital; [9]Department of Pediatric Rheumatology, Basaksehir Cam ve Sakura Hospital; [10]Department of Pediatric Rheumatology, Istanbul University Istanbul Faculty of Medicine; [11]Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Istanbul University-Cerrahpasa, Istanbul, Türkiye
**Correspondence:** B. N. Yumak
*Pediatric Rheumatology 2026*, **24(S1):**P394

**Introduction**
Practical barriers related to equipment, space, and time, together with the limited use of standardized self-report tools, challenge the routine assessment of physical fitness (PF) in Juvenile Idiopathic Arthritis (JIA) and Familial Mediterranean Fever (FMF).
**Objectives**
This study aimed to investigate the construct validity, internal consistency, and test–retest reliability of the International Fitness Scale (IFIS) and the Self-Perceived Health-Related Physical Fitness Questionnaire for Children (SPHQ-C) in children and adolescents with JIA and FMF.
**Methods**
This cross-sectional study included 151 patients with a mean age of 13.8±2.5 years, comprising 86 with JIA (4 with concomitant FMF) and 65 with FMF. PF was assessed via the FitnessGram Test Battery (curl-up, push-up, trunk lift, back-saver sit-and-reach, and PACER). Functional performance was evaluated via the 6-Minute Walk-6MWT, 10-Stair Climb-10SCT, 30-Second Sit-to-Stand-30STS, and 60-Second Sit-to-Stand-60STS tests. Perceived PF was assessed online via IFIS and SPHQ-C. Construct validity (Pearson correlation), internal consistency (Cronbach's alpha), and test–retest reliability (intraclass correlation coefficients) were analyzed.
**Results**
In the total sample, Cronbach's alpha was 0.79 for IFIS and 0.77 for SPHQ-C, while test–retest ICC values were 0.72 and 0.80, respectively. In the JIA group, alpha and ICC were 0.81/0.76 for IFIS and 0.78/0.77 for SPHQ-C. In the FMF group, alpha was 0.75 for both, with ICCs of 0.66 and 0.83, respectively. Flexibility had the highest reliability for IFIS in JIA (ICC=0.75) and for SPHQ-C overall (JIA: ICC=0.79; FMF: ICC=0.71). In JIA, SPHQ-C showed significant correlations with 10SCT ($r$=0.467), push-up ($r$=-0.450), 30STS ($r$=-0.432), 60STS ($r$=-0.368), curl-up ($r$=-0.355), PACER ($r$=-0.317), 6MWT ($r$=-0.282), while IFIS correlated with push-up ($r$=-0.360), 30STS ($r$=-0.303), 60STS ($r$=-0.275). Correlation directions aligned with perceived PF scoring. Most JIA correlations remained significant after adjusting for age, sex, and BMI. In FMF, initial SPHQ-C correlations with PACER ($r$=-0.318), push-up ($r$=-0.316), and 10SCT ($r$=0.260) became non-significant after adjustment.
**Conclusion**
IFIS and SPHQ-C demonstrated acceptable internal consistency and test–retest reliability in children and adolescents with JIA and FMF. SPHQ-C showed greater temporal stability, especially in FMF, while both scales showed stronger construct validity in JIA than in FMF. These findings suggest that self-reported PF measures may reflect objective PF and functional performance more consistently in JIA, whereas their interpretation in FMF may be more influenced by age, sex, and BMI. IFIS and SPHQ-C may therefore serve as complementary, practical tools for perceived PF assessment in pediatric rheumatic diseases.
**Disclosure of interest**
None declared.

## P395

### Sleep disturbance and fatigue in pediatric chronic nonbacterial osteomyelitis: a burden beyond disease activity

E. Cingoz[1,*], V. Cam[2], E. Sag[2], T. Kantemir[3], M. O. Erkan[2], D. Unal[2], H. Ercan Emreol[2], E. Yalcin[3], O. Basaran[2], D. Dogru Ersoz[3], N. Emiralioglu Ordukaya[3], S. Ozen[2], Y. Bilginer[2], H. U. Ozcelik[3]
[1]Pediatrics; [2]Pediatric Rheumatology; [3]Pediatric Pulmonology, Hacettepe University, Ankara, Türkiye
**Correspondence:** E. Cingoz
*Pediatric Rheumatology 2026*, **24(S1):**P395

**Table 1 (Abstract P395)** Sleep and fatigue scores in CNO patients and controls

| | CNO Active (*n*=41) | CNO Inactive (*n*=50) | Control (*n*=50) | *p* |
|---|---|---|---|---|
| CSHQ | 51(47–58) | 44(39–52) | 39(37–40) | <0.001*; <0.001** |
| SSR | 39(35–42) | 34(29–39) | 29(28–31) | <0.001*; <0.001** |
| PedsQL-MFS | 57(49–64) | 65(51–75) | 75(72–79) | 0.006*; <0.001** |

Values are median (IQR). p values represent *active vs. inactive CNO; **inactive CNO vs. control (Mann–Whitney U)

Higher CSHQ/SSR = worse sleep; lower PedsQL-MFS = more fatigue

**Introduction**
Chronic nonbacterial osteomyelitis (CNO) is an autoinflammatory bone disease with nocturnally exacerbated bone pain; cytokine activity and night-time pain are two potential mechanisms for sleep disruption, yet objective sleep findings by polysomnography (PSG) have not been characterised and no controlled study has assessed sleep and fatigue in CNO.
**Objectives**
To compare sleep quality and fatigue between children with CNO and healthy controls, to examine their relationship with disease activity and to characterise polysomnographic findings in patients with elevated sleep complaints.
**Methods**
This single-centre cross-sectional study included 91 children with CNO and 50 healthy controls (siblings or peers) similar in age and sex. Patients were defined as active or inactive based on the Clinical Disease Activity Score II. Sleep was assessed by the parent-reported Children's Sleep Habits Questionnaire (CSHQ) and child-reported Sleep Self-Report (SSR); fatigue by the PedsQL Multidimensional Fatigue Scale (PedsQL-MFS). Patients with CSHQ ≥41 (cut-off) were offered PSG.
**Results**
Children with CNO had worse scores than controls on all parameters of CSHQ, SSR, and PedsQL-MFS. The difference persisted in inactive patients vs. controls. Furthermore, active patients had worse scores than inactive (Table 1). In 33 PSG recordings, total sleep time was 275±88 min, sleep efficiency (SE) 62.7% (49.1–73.8), REM 9.6% (5.6–14.3) and apnoea–hypopnoea index 0.6/h (0.2–1.8); no obstructive events. SE and REM were below paediatric normative ranges (85–91% and 19–20%, respectively). Patients with nocturnal pain-related awakenings ≥2 nights/week (*n*=26) had worse scores than those with rare awakenings (*n*=65): CSHQ 55 vs. 47, SSR 39 vs. 35, PedsQL-MFS 52.1 vs. 63.9; all $p \leq 0.007$. Notably, such awakenings persisted in 12% of inactive patients.
**Conclusion**
Sleep disturbance and fatigue were more frequent and severe in children with CNO than in healthy peers, more pronounced in active disease, yet persisting in clinically inactive patients. PSG revealed shortened, inefficient sleep with REM suppression and sleep fragmentation, without obstructive events. Children with frequent pain-related night-time awakenings had worse sleep and fatigue scores, including in inactive patients. Routine sleep and fatigue assessment should be part of CNO follow-up. Screening may help identify both patients with pain-related sleep disruption and those with sleep burden irrespective of disease activity.
**Disclosure of interest**
None declared.

## P397

### The experience of acupuncture: patient reported outcome benefits of acupuncture treatment alongside standard non-pharmacological treatment in the management of paediatric chronic pain. A preliminary qualitative study

J. Riemer[1,*], J. Wang[2]
[1]Paediatric Rheumatology, Royal Manchester Children's Hospital; [2]Psychology, University of Manchester, Manchester, UK
**Correspondence:** J. Riemer
*Pediatric Rheumatology 2026,* **24(S1):**P397

**Introduction**
Chronic pain affects up to 30% of children and adolescents and is associated with significant physical, emotional, and social impairments. While multidisciplinary non-pharmacological interventions remain the cornerstone of treatment, acupuncture is increasingly used as an adjunctive therapy. However, there is limited evidence exploring patient-reported experiences and perceived benefits of acupuncture within paediatric chronic pain services. This study aimed to investigate the lived experiences, acceptability, and perceived outcomes of acupuncture alongside standard care in children and adolescents with chronic pain.
**Objectives**
To explore how children and adolescents describe their experiences of acupuncture within a multidisciplinary chronic pain service. To identify patient-reported physical, emotional, and functional outcomes associated with acupuncture treatment. To examine how acupuncture complements other non-pharmacological interventions. To generate thematic insights into the perceived value, mechanisms, and acceptability of acupuncture from the patient perspective.
**Methods**
A qualitative service evaluation was conducted within a UK NHS specialist paediatric chronic pain service. Seven participants aged 10–18 years with primary chronic pain who had received acupuncture were purposively recruited. Semi-structured interviews were conducted immediately post-treatment, focusing on treatment experiences, symptom changes, acceptability, and integration with wider care. Interviews were audio-recorded, transcribed verbatim, and analysed using reflexive thematic analysis following Braun and Clarke's six-phase framework.
**Results**
Participants described acupuncture as a calming and acceptable intervention that supported emotional regulation, improved sleep, and provided variable symptom-specific pain relief. Seven themes emerged: relaxation, sleep improvement, differentiated pain relief, tolerable sensory experiences, practitioner communication, flexible parental involvement, and treatment environment. Communication, predictability, and supportive practitioner behaviour were central to positive experiences. Environmental factors and service logistics also influenced engagement and perceived benefit.
**Conclusion**
Paediatric acupuncture may provide meaningful emotional, functional, and symptom-related benefits beyond pain reduction alone. Outcomes appear shaped by both physiological and psychosocial factors, including practitioner interaction, sensory experience, parental support, and treatment context. These findings support acupuncture as a valuable adjunct in multidisciplinary paediatric chronic pain management and highlight the importance of patient-centred, relational, and environmental considerations in service design.

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**Disclosure of interest**
None declared.

## P398

### Physical activity in children with JIA: influence of age, disease characteristics and treatment

K. Petrovic[1,*], D. Novakovic[1], M. Grujic[1], S. Lukovic[1], M. Vorkapic[1]
[1]Institute of Rheumatology, Belgrade, Serbia
**Correspondence:** K. Petrovic
*Pediatric Rheumatology 2026,* **24(S1):**P395

**Introduction**
It is known that children with juvenile idiopathic arthritis (JIA) have lower level of physical activity than healthy children, however in era of biologic drugs when low disease activity or remission is achievable, physical activity still remains important tool for sustaining good quality of life.
**Objectives**
The aim of this study was to assess physical activity levels in children with JIA receiving biological therapy, as well as to identify other associated factors that can have impact on physical activity.
**Methods**
This cross-sectional study included 30 children with JIA receiving biological therapy who were in remission and whose parents or legal guardians provided written informed consent prior to participation. Physical activity was assessed using the Physical Activity Questionnaire for Children (PAQ-C), while functional status of patients was evaluated using the Childhood Health Assessment Questionnaire (CHAQ). Clinical and treatment-related data, including disease duration, subtype, pain intensity, psychological coping, and therapy history, were obtained from electronic medical records.
**Results**
The study included 8 boys (26.7%) and 22 girls (73.3%), with a mean age of 11.6 ± 2.4 years. According to physical activity level, 9 children (30.0%) were classified as having low physical activity, while 21 children (70.0%) had moderate physical activity levels. Children with low physical activity levels tended to be older compared with those with moderate physical activity levels (14 0.0 vs. 11.0 years, $p = 0.046$). No significant differences were observed between the groups regarding sex distribution, BMI values (20.7 vs. 19.7 kg/m$^2$, $p = 0.756$), disease duration (7.8 vs. 6.7 years, $p = 0.568$), polyarticular disease frequency (77.8% vs. 76.2%, $p = 1.00$), pain intensity (3.3 vs. 3.0, $p = 0.534$), or psychological coping with the disease (13.9 vs. 4.0, $p = 0.402$). Regarding treatment characteristics, biologic therapy duration was comparable between the groups (54.6 vs. 48.8 months, $p = 0.946$). Biologic therapy switching was more frequent among children with low physical activity levels (44.4% vs. 23.8%, $p = 0.389$), although the difference was not statistically significant. No significant differences were found regarding corticosteroid use, methotrexate use, or ANA positivity between the groups.
**Conclusion**
Disease characteristics did not appear to significantly affect physical activity levels in children with JIA receiving biological therapy, while older age seemed to be associated with lower activity. Larger prospective studies are needed to confirm these results. These findings highlight the importance of regular monitoring, education, and targeted interventions to promote physical activity in children with JIA.

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**Disclosure of interest**
None declared.

## P401

**Assessing the participant experience with fibrodysplasia ossificans progressiva: qualitative interviews from the optima trial**

J. Gu[1], C. Hartford[1], S. Martin[2], N. Harris[2], R. Papa[3,*], P. Delai[4], R. Keen[5], K. Mishima[6], K. Dahir[7], N. Uemura[8], T. Fujiwara[9], S. Rhee[1], R. Pordy[1], R. J. Sanchez[1]
[1]Regeneron Pharmaceuticals, Inc., Tarrytown, NY; [2]RTI Health Solutions, Durham, NC, USA; [3]IRCCS Istituto Giannina Gaslini, Genoa, Italy; [4]Hospital Israelita Albert Einstein, São Paulo, Brazil; [5]Royal National Orthopedic Hospital NHS Trust, Middlesex, UK; [6]Nagoya University Hospital, Nagoya, Japan; [7]Vanderbilt University, Nashville, TN, USA; [8]Oita University, Oita; [9]Kyushu University Hospital, Fukuoka, Japan
**Correspondence:** R. Papa
*Pediatric Rheumatology 2026*, **24(S1):**P401

**Introduction**
Garetosmab (GAR), a fully human monoclonal anti-activin A antibody, was investigated in adults with fibrodysplasia ossificans progressiva (FOP) in the phase 3 OPTIMA trial and demonstrated an acceptable safety profile and reduced new heterotopic bone lesions. In OPTIMA, qualitative interviews were performed in a subset of participants (pts) to further characterize GAR's safety and efficacy.
**Objectives**
Pts were asked to describe FOP symptoms and their impacts before the start of OPTIMA, changes experienced during the trial, and the meaningfulness of the changes.
**Methods**
Sixty-minute semi-structured interviews were conducted with pts using open-ended questions. Pts and moderators were blinded to treatment assignment. Interviews took place at clinical sites in 6 countries (USA, Brazil, Chile, Italy, UK, and Japan) within 14 days following the Week 52 visit or early termination visit. Interview transcripts were coded using ATLAS.ti 9 software, and thematic analysis methods were used to analyze the data. Trends were identified in each interview to generate themes or patterns in pts' experiences with FOP and the study treatment, and to identify any changes seen during the study. As this was a qualitative study, formal hypotheses were not tested.
**Results**
Thirty of 63 (47.6%) OPTIMA pts who completed their Week 52 visit were interviewed. Mean age was 25.5 years (range 18–39); 17 (57%) pts were female. Pretrial, pts reported decreased movement, impaired physical functioning (both 100%), and pain and swelling (both 90%) that affected their ability to perform routine activities. After treatment, most pts (83%) reported ≥1 improvement in a symptom or issue; physical functioning (17/25), movement (16/25), and frequency of flare-ups (14/25) were the most common. Increased movement and decreased pain, respectively, were reported by more pts from the GAR groups (13/20 and 8/20) than placebo (3/10 and 1/10). Pts highlighted the meaningfulness of the improvements, noting greater independence and participation in daily activities.
**Conclusion**
These data show a significant pretreatment burden of disease in pts with FOP and provide insights into the benefits to pts of GAR in OPTIMA.
**Trial registration identifying number**
ClinicalTrials.gov, NCT05394116.
**Disclosure of interest**
J. Gu Shareholder with: Regeneron Pharmaceuticals, Inc., Employee with: Regeneron Pharmaceuticals, Inc., C. Hartford Shareholder with: Regeneron Pharmaceuticals, Inc., Employee with: Regeneron Pharmaceuticals, Inc., S. Martin Consultant with: Regeneron Pharmaceuticals, Inc., Employee with: RTI Health Solutions, N. Harris Consultant with: Regeneron Pharmaceuticals, Inc., Employee with: RTI Health Solutions, R. Papa Grant / Research Support with: PI of the OPTIMA trial, funded by Regeneron Pharmaceuticals, Inc., Consultant with: Regeneron Pharmaceuticals, Inc., P. Delai: None declared, R. Keen Grant / Research Support with: PI of the LUMINA-1 and OPTIMA trials, funded by Regeneron Pharmaceuticals, Inc., K. Mishima: None declared, K. Dahir Grant / Research Support with: PI of the LUMINA-1 and OPTIMA trials, funded by Regeneron Pharmaceuticals, Inc.; reseach support from Alexion, AstraZeneca, Ultragenyx, Kyowa Kirin, Consultant with: Alexion, AstraZeneca, Ultragenyx, Kyowa Kirin, N. Uemura Grant / Research Support with: Regeneron Pharmaceuticals, Inc., Eli Lilly and Company, Taisho Pharmaceutical Co., Ltd., Eisai Co., Ltd., Chugai Pharmaceutical Co., Ltd., and Ono Pharmaceutical Co., Ltd., Consultant with: MSD KK, Ono Pharmaceutical Co., Ltd., Sato Pharmaceutical Co., Ltd., Seren Pharma Inc., Swedish Orphan Biovitrum AB (Sobi), SFG Sciences, Inc., and Juro Sciences, Inc., Paid Instructor with: Mochida Pharmaceutical Co., Ltd., Meiji Seika Pharma Co., Ltd., and MSD KK, T. Fujiwara: None declared, S. Rhee Shareholder with: Regeneron Pharmaceuticals, Inc., Employee with: Regeneron Pharmaceuticals, Inc., R. Pordy Shareholder with: Regeneron Pharmaceuticals, Inc., Employee with: Regeneron Pharmaceuticals, Inc., R. Sanchez Shareholder with: Regeneron Pharmaceuticals, Inc., Employee with: Regeneron Pharmaceuticals, Inc.

P402

**Knee pain in children and adolescents in a tertiary pediatric orthopedic center: a five year electronic health record study**

R. Raupov[1,*], S. Vissarionov[1], G. Babaeva[1], Y. Noyanova[1], M. Kostik[2]
[1]H. Turner National Medical Research Center for Children's Orthopedics and Trauma Surgery; [2]Saint Petersburg State Pediatric Medical University, Saint Petersburg, Russian Federation
**Correspondence:** R. Raupov
*Pediatric Rheumatology 2026*, **24(S1):**P402

**Introduction**
Knee pain is one of the most frequent complaints in children and adolescents. It can arise from a wide spectrum of traumatic, overuse, developmental, inflammatory, and tumorrelated conditions. Most of the existing epidemiological data come from primary care, and they do not fully show which diagnoses are actually seen in children referred to specialists.
**Objectives**
To describe the spectrum of diagnoses underlying knee pain in children and adolescents seen in a tertiary pediatric orthopedic center.
**Methods**
We conducted a retrospective electronic health record–based study of all outpatient visits with knee pain in patients aged 0–17 years over the last five years (2020–2025) at a tertiary pediatric orthopedic center. Demographic data, ICD10 codes, freetext clinical diagnoses, and treating physician were extracted and grouped into clinical categories: trauma/internal derangements, deformities/axis disorders/dysplasia, inflammatory/rheumatologic, overuse/apophysitis, tumor/tumorlike lesions, arthralgia/nonspecific joint pain, and other/unspecified diagnoses. Age was stratified into four groups (0–4, 5–9, 10–13, 14–17 years).
**Results**
We analyzed 21 083 kneerelated outpatient visits in 13 298 unique patients (mean age 13,0 years). Females accounted for 11 877 visits (56,3%) and males for 9 081 (43,1%). The most frequent ICD10 diagnosis codes were M25.5 "pain in joint" (*n*=7 090; 33,6%), M95.8 "other specified acquired deformities of the musculoskeletal system" (*n*=2 558; 12,1%), M23.8 "other internal derangements of knee" (*n*=1 947; 9,2%), M24.1 "other specific joint derangements" (*n*=1 571; 7,4%), and M92.5 "apophysitis of tibial tuberosity" (*n*=1 042; 4,9%). Deformities and inflammatory conditions were relatively more frequent in younger children, whereas internal derangements, overuse conditions, and nonspecific arthralgia predominated in adolescents. Overall, 4 346 patients (32,7%) had ≥2 kneerelated visits and 1 764 (13,3%) had ≥3 visits. Within visits coded as M25.5/M25.8, freetext diagnoses revealed patellofemoral pain, apophysitis, meniscal and ligamentous lesions. Orthopedists managed 17 686 visits (83,9%), rheumatologists 2 189 (10,4%), rehabilitation physicians 658 (3,1%), and neurologists 550 (2,6%), illustrating routine multidisciplinary involvement.
**Conclusion**
Pediatric and adolescent knee complaints in this large ICD10–based outpatient cohort were predominantly noninflammatory. Agedependent patterns, frequent repeat visits, and consistent multidisciplinary involvement underscore the need for structured, agespecific care pathways to improve early recognition and management of mechanical and inflammatory knee disorders.
**Disclosure of interest**
None declared.

P404

**Organization of the work of the family patient school in cooperation with the patient organization of parents of children suffering from juvenile arthritis**

Y. M. Spivakovskiy[1,*], A. Y. Spivakovskaya[2]
[1]Department of Faculty Pediatrics; [2]Department of Hospital Pediatrics, State Medical University n.a. V.I. Razumovsky, Saratov, Russian Federation
**Correspondence:** Y. M. Spivakovskiy
*Pediatric Rheumatology 2026*, **24(S1):**P404

**Introduction**
Involving the medical community in closer cooperation with patient organizations can significantly increase patients' awareness of their disease, the risks of complications, the need for therapy, its types and safety limits, as well as a number of other issues requiring special knowledge.
**Objectives**
The aim of the project, implemented within the region, was to create and implement a set of measures that could significantly increase the awareness of patients with juvenile arthritis (JIA) and their parents about the features of their disease, the need for regular therapy and affect the level of adherence to medical recommendations.
**Methods**
To implement the action plan, an interdisciplinary team of doctors was created, which included employees of the university, the university clinic and student volunteers. The main events of the developed program were the holding of theatrical performances dedicated to World Arthritis Day and World Young Rheumatic Diseases Day, the creation of a series of informational videos dedicated to various specialized issues of rheumatology.
**Results**
The theatrical events included information content on aspects of rehabilitation of patients with JIA. During their performance, patients with JIA became mandatory participants in the performance. This made it possible to demonstrate the integration of patients into society and the possibilities for achieving the desired goals with regular therapy. At least 13 videos were created in which doctors explained various aspects of pediatric rheumatology. Among the topics: historical aspects of rheumatology, issues of the pathogenetic aspects of JIA, treatment approaches, the specifics of the use of various drugs, the need for regular monitoring at the outpatient stage, possible complications of JIA and therapy. The issues of ophthalmological disorders were considered separately with the involvement of an ophthalmologist. Several videos were devoted to the issues of non-drug correction and rehabilitation of patients with JIA. The created video content was posted in the media communities of the patient organization, as well as on the medical websites of the University Clinic. The number of views and reactions gave a good understanding of the high demand for this information resource. Additionally, based on the results of familiarization of the patient community with these materials, answers were given to questions that arose (the most frequent among which were questions about the prospects in "adult" life, social issues of adaptation).
**Conclusion**
The established system of the family patient school has demonstrated the need for additional contacts with the patient community. The results of its implementation have increased the level of awareness of patients about the disease, which has improved compliance and will allow achieving better results in the treatment and rehabilitation of patients with JIA. The established family patient education system demonstrated the need for additional interaction with patients at the community level. The results of this implementation significantly increased patients' awareness of the specifics of their condition, which, in turn, improved compliance with the requirements and improved the results of treatment and rehabilitation of patients with JIA.
**Disclosure of interest**
None declared.

P405

**Clinico-epidemiological profile of pediatric rheumatology diseases in Tripoli, Libya**

A. A. Abushhaiwia[1,*], Y. Elfawires[1], L. O. Mendonca[2]
[1]Pediatric Rheumatology, Tripoli children's Teaching Hospital, faculty of Medicine, university of Tripoli, Tripoli, Libya; [2]discipline of clinical immunology and allergy, faculdade de medicina a, universidade de são Paulo, São Paulo, Brazil
**Correspondence:** A. A. Abushhaiwia
*Pediatric Rheumatology 2026*, **24(S1):**P405

**Introduction**
Pediatric rheumatology encompasses a diverse spectrum of inflammatory, autoimmune, and autoinflammatory conditions. Data on the epidemiology and clinical characteristics of these diseases in North Africa remain scarce, limiting regional understanding and resource allocation.
**Objectives**
To describe the clinico-epidemiological profile, diagnostic spectrum, treatment patterns, and clinical outcomes of pediatric rheumatology patients followed at a tertiary center in Tripoli, Libya.

**Methods**
A cross-sectional descriptive study of 763 patients registered at the Pediatric Rheumatology Unit, Tripoli Children's Teaching Hospital. Data on demographics, diagnoses, treatments, and outcomes were extracted from the clinical registry and analyzed descriptively.
**Results**
Of 763 patients (53.1% female), the most prevalent diagnoses were miscellaneous/unclassified conditions (26.7%), IgA vasculitis/Henoch-Schonlein purpura (19.5%), juvenile idiopathic arthritis (17.6%), Kawasaki disease (12.2%), and autoinflammatory diseases (9.0%). Mean age of symptom onset was 6.5 years (SD 3.7), with a diagnostic delay of approximately 8 months. NSAIDs and corticosteroids were used in 55.6% and 52.8% of patients, respectively. Methotrexate was the most common conventional DMARD (24.4%). Biologic agents were used in approximately 13.6% of patients. Among those with documented outcomes, 49.2% improved and 46.8% remained stable; overall mortality was 0.5%.
**Conclusion**
This registry-based study provides the first large-scale description of pediatric rheumatology diseases in Libya. The disease spectrum mirrors patterns reported from neighboring Arab countries, with vasculitis and JIA predominating. The high rate of missed follow-up and the limited access to biologic therapy highlight areas requiring health system reinforcement.
**Keywords**
Pediatric rheumatology; Juvenile idiopathic arthritis; IgA vasculitis; Kawasaki diseaseLibya; North Africa; Epidemiology
**Disclosure of interest**
None declared.

P406

## Care of juvenile idiopathic arthritis in the 21st century in Europe

L. De Somer[1,2,*], S. Pazmino[3], J. Anton Lopez[4], B. Bader-Meunier[5], M. Bonde Glerup[6], T. Dallos[7], M. Jelušić[8], O. Killen[9], L. Lijnen[10], K. Minden[11], C. Normand[12], J. Swart[13], L. Van Goidsenhoven[14], J. Willen[15], J. Wójtowicz[16], C. Wouters[1,17] on behalf of PAVE consortium
[1]Department of Pediatrics, University Hospitals Leuven; [2]Department of Microbiology and Immunology; [3]Department of Chronic Diseases and Metabolism, KU Leuven, Leuven, Belgium; [4]Hospital Sant Joan de Déu, Universitat de Barcelona, Barcelona, Spain; [5]Necker Hospital, Assistance Publique-Hôpitaux de Paris, Paris, France; [6]Aarhus University, Aarhus, Denmark; [7]Second Department of Pediatrics, Comenius University Medical School, Bratislava, Slovakia; [8]University Hospital Centre Zagreb, Zagreb, Croatia; [9]Our Lady's Children's Hospital Crumlin, Crumlin, Ireland; [10]Department of Medicine and Life Sciences, Hasselt University, Diepenbeek, Belgium; [11]Charité University Hospital, Berlin, Germany; [12]MCI Secretariat, European Network for Children with Arthritis (ENCA), Geneva, Switzerland; [13]Department of Pediatric Rheumatology and Immunology, University Medical Center Utrecht, Utrecht, Netherlands; [14]Department of Philosophy, University of Antwerp, Antwerp; [15]Child Neurology, University Hospitals Leuven, Leuven, Belgium; [16]REUMAPEDIC Centre and Pediatric Rheumatology Ambulatory, National Institute of Geriatrics, Rheumatology and Rehabilitation, Warsaw, Poland; [17]Department of Microbiology, Immunology and Transplantation, KU Leuven, Leuven, Belgium
**Correspondence:** L. De Somer
*Pediatric Rheumatology 2026*, **24(S1):**P406

**Introduction**
The management and treatment of juvenile idiopathic arthritis (JIA) have greatly improved over recent decades; however, its socioeconomic burden remains considerable and may vary across national healthcare systems.
**Objectives**
To identify barriers to health equity in children with JIA across Europe, including timely access to diagnosis, appropriate treatment and follow-up.
**Methods**
Pseudonymized questionnaires on disease activity, treatment and health-related quality of life (PedsQL) were completed by parents/caregivers of children diagnosed with JIA -all subtypes-, actively receiving treatment, with or without active disease. Data analysis was descriptive, reporting continuous variables as means with standard deviations or medians with interquartile ranges -depending on distribution- and categorical variables as percentages.
**Results**
Across 10 European centres (Belgium, Croatia, Denmark, France, Germany, Ireland, the Netherlands, Poland, Slovakia, and Spain), 442 questionnaires were filled-in of which 351 questionnaires had a completeness over 70% and were used for analysis. The respondents were in its majority (98.6%) the biological mother (77%) or father of the child living with JIA, with a mean age of 42.7 years ± 6.8 (SD), married or living together (85%). Most respondents had a high level of education (69% -University/Academy/Graduate School-), mostly (68%) working full-time, and more than half (56%) are families constituted with two children. The children with JIA, were on average 11.1 years old ± 4.6 (SD), mostly females (67%), with oligoarthritis (46.2%) and polyarthritis (30.9%) as the most frequent subtypes. Overall disease activity was reported to be low with a large majortiy (>85%) mentioning rare or no joint pain nor stiffness. The reported influence of JIA on the school trajectory was low (median 1, IQR 5) on a 0-10 scale with lower values representing less impact. There was almost no impact (≈90% never or almost never) on daily activities and 79% reported no change to their extracurricular activities due to JIA. Furthermore, there was a high level of satisfaction in terms of communication of diagnosis and treatment (median 9, IQR 2) and of accessibility to healthcare (median 8, IQR 4) on a 0-10 scale with higher values representing more satisfaction. Parents reported a low level of worries about the financial consequences due to JIA with a median of 1 (IQR 5) but moderate level of worries about the extent of JIA impact on the child's future (median 5, IQR 6). Moreover, 26% mentioned worrying about the illness and 21% worry about medication side-effects often or almost always. Of note, awareness of national patient associations was low -less than 4 out of 10 patients' families reported to be informed- and less than a quarter of the families informed were involved in a patient organisation. There were no major differences in findings among the different European centres, a detailed subgroup analysis is ongoing.
**Conclusion**
Overall, patients with JIA across Europe appear to have timely access to appropriated health care, well controlled disease, and low impact on daily life in terms of school, activities, and financial consequences. Despite this, parental worrying is a concern which could be addressed with the help of patient organisations.
**Trial registration identifying number**
S66869.
**Disclosure of interest**
None declared.

P407

## Multidisciplinary team online meetings (MDT) for the care and management of patients with type I interferonopathies: a four-year French experience

S. Plassart[1], I. Melki[2], A. Belot[1], O. Boepsflug-Tanguy[3], F. Chasset[4], C. David[5], I. Desguerre[6], A. Hadchouel[6], E. Jeziorski[7], N. Nathan[8], B. Neven[6], F. Robin-Renaldo[8], A. Roubertie[7], C. Sevin[9], M. Tusseau[10], A. Welfringer[6], M. Hully[6], M. L. Frémond[11,*] on behalf of AGS & Interferonopathy French MDT Network
[1]CRMR RAISE, Hospices Civils de Lyon, LYON; [2]Service de Pédiatrie Générale, Hôpital Trousseau, APHP; [3]Hôpital Robert Debré, APHP; [4]Hôpital Tenon, APHP; [5]Hôpital Bichat, APHP; [6]Hôpital Necker-Enfants-Malades, APHP, PARIS; [7]CHU de MONTPELLIER, MONTPELLIER; [8]Hôpital Trousseau, APHP; [9]Hôpital Bicêtre, PARIS; [10]Genetics Department, Hospices Civils de Lyon, LYON; [11]CRMR RAISE, Hôpital Necker-Enfants-Malades, APHP, PARIS, France
**Correspondence:** M. L. Frémond
*Pediatric Rheumatology 2026*, **24(S1):**P407

**Introduction**
Type I interferonopathies (IFNp-1) are a group of rare genetic autoinflammatory disorders characterised by chronic activation of type I interferon (IFN-I) signalling. They encompass a broad clinical spectrum, including neurological, cutaneous, articular, and pulmonary manifestations, and therefore require multidisciplinary evaluation and management. These disorders are often severe, associated with high morbidity and mortality, and may be refractory to conventional immunosuppressive therapies. Moreover, patients are frequently treated with off-label therapies targeting IFN-I signalling, requiring close monitoring and

dedicated follow-up. In France, a national expert multidisciplinary team (MDT) was established in March 2022 to support personalised diagnostic and therapeutic strategies for patients with Aicardi-Goutières syndrome (AGS) or other IFNp-1 through monthly online meetings.

**Objectives**

Here, we report a four-year experience of multidisciplinary online MDT meetings involving French reference centres across all clinical specialties caring for patients with IFNp-1.

**Methods**

The MDT comprises a medical coordinator and a multidisciplinary panel including paediatric and adult specialists in neurology, rheumatology, immunology, pulmonology, dermatology, as well as other subspecialists when required, together with geneticists and scientists. The referring physician submits a form summarising clinical and genetic data, prior management, and the clinical question. Cases are discussed online, with recommendations provided in a written report. All exchanges are secure, with informed consent obtained from patients or their parents. We conducted a retrospective analysis of real-world data from online MDT meetings for AGS & IFNp-1 between March 2022 and March 2026.

**Results**

Over the 48-month study period, 39 MDT sessions were conducted, during which 153 cases were discussed, with a median of 4 cases per session and 86% involving paediatric cases. A total of 115 individual patients were discussed. The majority of requests involved AGS patients (54%, *n*=83). The remaining cases concerned patients with other monogenic IFNp-1 (e.g. SAVI, COPA), as well as patients with suspected genetic interferonopathies prompting genetic analysis and detailed clinical investigation. Among AGS patients, nearly all discussion requests related to therapeutic management, particularly the use of steroids and targeted therapies (JAK inhibitors and anifrolumab). The MDT recommended a targeted treatment in 64% of cases.

**Conclusion**

Given the complexity and multisystem involvement of IFNp-1, MDT meetings provide access to organ-specific expertise, support diagnostic refinement through additional (including genetic) investigations, and enable personalised, guideline-based treatment, and access to innovative therapies. This approach ensures optimal patient care through national expertise and provides a model for the multidisciplinary management of rare disorders.

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**Trial registration identifying number**

**Disclosure of interest**

None declared.

P408

**Paediatric centre care reshapes resource use in PFAPA and JIA: contrasting trajectories from Czech hospitals**

G. Donin[1,2], I. Šarkanová[3], V. Šimka[4], P. Doležalová[4,*]
[1]Department of Biomedical Technology, Czech Technical University, Kladno; [2]Institute of Health Information and Statistics of the Czech Republic; [3]Department of Biomedical Technology, Czech Technical University; [4]Department of Paediatrics and Inherited Metabolic Disorders, General University Hospital in Prague, Prague, Czech Republic
**Correspondence:** P. Doležalová
*Pediatric Rheumatology 2026*, **24(S1):**P408

**Introduction**

Children with chronic or recurrent rheumatic and autoinflammatory complaints, notably periodic fever, aphthous stomatitis, pharyngitis, adenitis (PFAPA) syndrome and juvenile idiopathic arthritis (JIA), often receive fragmented, non-specific care before reaching a paediatric rheumatology centre, where diagnosis and disease-specific management consolidate. The economic and resource-use consequences of this transition are poorly quantified.

**Objectives**

To compare patterns of care, resource use and costs before vs. after a child entered care at a paediatric rheumatology centre, separately for PFAPA and JIA.

**Table 1 (Abstract P408)** Key resource-use and cost changes after rheumatology centre entry

| Outcome | PFAPA (*n*=253) | JIA (*n*=306) | *p* |
|---|---|---|---|
| Total annual cost | **– 6%** | **+ 262%** | <0.001 |
| Hospitalisation rate | **– 60%** | + 1% | <0.001 / NS |
| Inpatient days | **– 46%** | - 15% | <0.001 / NS |
| Outpatient cost (excl. drugs) | **+ 16%** | **+ 151%** | <0.001 |
| Drug cost (among users) | **– 29%** | **+ 1159%** | <0.001 |

NS non significant

**Methods**

A retrospective pre/post cohort study was conducted using the Czech National Registry of Reimbursed Health Services (NRHZS). Children <18y with PFAPA or JIA entering care at one of the two largest Czech paediatric rheumatology centres between 2019 and 2023 were included, with ≥2y follow-up before and after the index visit (first complex rheumatology consultation). Within-patient pre/post changes were estimated using adjusted mixed-effects models with patient-level random intercepts, adjusting for age, sex, calendar year and centre.

**Results**

253 children with PFAPA (median age 5y, 59% boys) and 306 with JIA (10y, 68% girls) entered centre care with sharply divergent trajectories (Table 1). In PFAPA, the overall cost decreased modestly (~6%), yet the care mix reorganised substantially: hospitalisations fell while outpatient management rose, consistent with diagnostic resolution replacing reactive admissions. In JIA, total cost rose ~3.6-fold, driven by biologic initiation in ~40% of patients (vs. <5% pre-index), with drug cost rising >20-fold among recipients. All adjusted contrasts $p<0.001$ unless noted.

**Conclusion**

Centre-based paediatric rheumatology care reshapes healthcare trajectories in disease-specific ways: in PFAPA, fever-related admissions are replaced by scheduled outpatient management without raising overall cost; in JIA, disease-modifying therapy is initiated at substantial but expected incremental cost. Disease-specific economic accounting better supports referral, budget and care-pathway decisions than pooling these conditions.

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**Disclosure of interest**

None declared.

P412

**Safety and tolerability of intranasal live attenuated influenza vaccine in children with juvenile idiopathic arthritis receiving TNFI**

A. Neophytou[1], T. Plessa[1], M. Kardani[1], D. Maritsi[1,*]
[1]Second Department of Pediatrics, "P & A Kyriakou" Children's Hospital, NKUA, ATHENS, Greece
**Correspondence:** D. Maritsi
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**Introduction**

Children with juvenile idiopathic arthritis (JIA) receiving biologic therapies, are at increased risk of infections. Annual influenza vaccination is recommended; however, data on the safety of live attenuated intranasal influenza vaccines (LAIV) in this population remain limited, especially in patients with stable disease on biologics.

**Objectives**

To assess the safety of intranasal LAIV in children with JIA treated with TNFi, by evaluating adverse events, post-vaccination influenza infection, and disease flare occurrence.

**Methods**

We conducted a prospective observational study including children with JIA receiving TNFi. All participants were in clinical remission for at least three months. All subjects had received seasonal influenza vac-

cination in previous years. During the 2025–2026 influenza season, patients received the quadrivalent LAIV at the beginning of the season. Demographics, JIA subtype, disease duration, current and previous treatments, vaccination history and other comorbidities were recorded. Patients were followed for three months post-vaccination. Outcomes included vaccine-related adverse events, occurrence of clinically suspected or laboratory-confirmed influenza, and disease flares requiring treatment escalation. Disease activity was assessed using the Juvenile Arthritis Disease Activity Score (JADAS) at 0 and 3 months. The study was approved by the Hospital's Research and Ethics Committee (Approval number 17/1832/cd110314). Informed consent was obtained. Analyses were conducted using STATA(version 23.0).

**Results**

Thirteen patients were included (9 females, 4 males) with a mean age of 4.5 (+/- 1.2) years. Eight patients had pJIA, three psJIA and the remaining were ext-oligoJIA. Six(45%) were on etanercept and the remaining were on adalimumab. The vaccine was well tolerated, with no immediate or delayed serious adverse events reported. Mild transient vaccine-related reactions such as blocked nose (30%), fatigue (23%) decreased appetite (23%) and myalgia (15%) were observed, but resolved within 5 days. Within the first two weeks following vaccination, no patients developed clinically suspected or confirmed influenza. Two patients were infected by influenza type B approximately two months post-vaccination. Importantly, no patients experienced a flare of their underlying disease or required escalation of treatment during the follow-up period. Subgroup analysis did not detect any differences in terms of side effects amongst the various JIA groups or in between the two treatment arms (ADAvsETN).

**Conclusion**

In this small cohort of children with JIA on TNFi and stable disease, the LAIV demonstrated a favorable safety profile, with no associated disease flares or serious adverse events. These findings suggest that LAIV may be a safe option in carefully selected patients under close monitoring, although larger studies are needed to confirm these findings.

**Disclosure of interest**

None declared.

## P413

### Activator protein-1 (AP-1) as a common driver of ectopic ossification, fibrosis, and inflammation: shared therapeutic targets in FOP and rheumatic diseases

E. D. Kopylov[1,*], P. A. Golubinskaya[2], A. I. Kadykova[3], N. S. Gladyshev[1], A. V. Eremeev[2], R. V. Deev[1]

[1]Avtsyn Research Institute of Human Morphology of Federal state budgetary scientific institution "Petrovsky National Research Centre of Surgery"; [2]Lopukhin Federal Research and Clinical Center of Physical-Chemical Medicine; [3]NCSM FMBA, Moscow, Russian Federation

**Correspondence:** E. D. Kopylov

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**Introduction**

Activator protein 1 (AP-1) is a transcriptional regulator involved in rheumatoid arthritis, psoriasis, and bone-immune crosstalk (osteoimmunology) [1]. Heterotopic ossification (HO), the hallmark of fibrodysplasia ossificans progressiva (FOP), is driven by *ACVR1* mutation. Beyond aberrant Smad signaling, inflammation contributes to disease progression. AP-1 factors are enriched in HO lesions, and their pharmacological inhibition reduces osteochondrogenic differentiation [2].

**Objectives**

This study aimed to non-invasively generate and characterize a patient-derived cellular model of FOP to investigate AP-1 as a shared pathogenic node across FOP and rheumatic diseases.

**Methods**

Dental pulp cells were isolated via explant culture from deciduous teeth of a FOP patient (*ACVR1* R206H) and a healthy control (SHED). Immunophenotyping was performed by flow cytometry. Multi-omics included RNA-seq (Illumina, paired-end 100 bp, STAR alignment to GRCh38) and DIA-LC-MS/MS (Orbitrap Tribrid Lumos, DIA-NN search). Differential expression was defined as |log2FC|>1. Network analysis used STRING v12.0 (confidence >0.7, MCL clustering).

**Table 1 (Abstract P413)** Concordant multi-omic changes in FOP MSCs vs. SHED

| Gene/Protein | log2FC (RNA) | log2FC (Protein) | Function / Relevance |
|---|---|---|---|
| FOS | +9.16 | - | AP-1 subunit; master regulator |
| FOSB | +6.70 | - | AP-1 subunit; osteogenic |
| JUNB | +2.19 | +1.06 | AP-1 subunit; inflammatory |
| ATF3 | +1.83 | - | AP-1 family; stress response |
| IL6 | +1.45 | - | Pro-inflammatory cytokine |
| COL11A1 | +2.10 | +6.54 | Fibrosis/cartilage matrix |
| INHBA | -0.79 | +1,14 | Activin A subunit |
| FST | +1.64 | +3.01 | Activin inhibitor (compensatory) |

**Results**

The FOP MSC line retained fibroblast-like morphology, stable growth, and confirmed the heterozygous *ACVR1* R206H mutation (NM_001111067.4:c.617G> A). Flow cytometry confirmed MSC identity (CD90+, CD73+, CD105+, CD34–, CD45–). Multi-omics revealed massive, concordant AP-1 complex activation (Table 1). Protein-protein interaction networks showed functional links between AP-1 components and inflammatory/fibrotic mediators. GSEA confirmed activation of osteoblast proliferation, cartilage development, and negative regulation of inflammatory response pathways. Interferon-stimulated genes (*OAS1*, *MX1*) and the retroviral capsid-like *PNMA2* were also upregulated, suggesting sterile inflammation.

**Conclusion**

We established and characterized a non-invasively derived, patient-specific FOP cell model that faithfully retains the *ACVR1* R206H driver. Multi-omics consistently identifies extreme AP-1 complex overexpression as a concordant transcriptional signature in FOP. AP-1 links canonical BMP/Smad dysregulation to downstream inflammation and matrix remodeling, mirroring its pathogenic role in common rheumatic diseases. Cellular model provides a human-relevant platform for studying AP-1-driven mechanisms and testing shared AP-1-targeted therapies for FOP and rheumatic conditions featuring ectopic bone, fibrosis, and synovial inflammation.

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**Trial registration identifying number**

**Disclosure of interest**

None declared.

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