English Products

Web of Science™ Search Sign In Register

Search > Results for sarper N (Author) > Identification of the molecular etiology in rare congenital hemolytic anemi…

MENU

Full text at publisher Export Add To Marked List

3 of 102

Identification of the molecular etiology in rare congenital hemolytic anemias using next-generation sequencing with exome-based copy number variant analysis

Are you this author?

By Isik, E (Isik, Esra) [1] , [2] ; Aydinok, Y (Aydinok, Yesim) [3] ; Albayrak, C (Albayrak, Canan) [4] ; Durmus, B (Durmus, Basak) [2] ; Karakas, Z (Karakas, Zeynep) [5] ; Orhan, MF (Orhan, Mehmet Fatih) [6] ; Sarper, N (Sarper, Nazan) [7] ; Aydin, S (Aydin, Sultan) [8] ; Unal, S (Unal, Selma) [9] ; Oymak, Y (Oymak, Yesim) [10] ; Karadas, N (Karadas, Nihal) [2] ; Turedi, A (Turedi, Aysen) [11] ; Albayrak, D (Albayrak, Davut) [12] ; Tayfun, F (Tayfun, Funda) [13] ; Tugcu, D (Tugcu, Deniz) [5] ; Karaman, S (Karaman, Serap) [5] ; Tobu, M (Tobu, Mahmut) [14] ; Unal, E (Unal, Ekrem) [15] ; Ozcan, A (Ozcan, Alper) [15] ; Unal, S (Unal, Sule) [16] ; Aksu, T (Aksu, Tekin) [16] ; Unuvar, A (Unuvar, Aysegul) [5] ; Bilici, M (Bilici, Mustafa) [5] ; Azik, F (Azik, Fatih) [17] ; Ay, Y (Ay, Yilmaz) [18] ; Gelen, SA (Gelen, Sema Aylan) [7] ; Zengin, E (Zengin, Emine) [7] ; Albudak, E (Albudak, Esin) [19] ; Eker, I (Eker, Ibrahim) [20] , [21] ; Karakaya, T (Karakaya, Taner) [22] ; Cogulu, O (Cogulu, Ozgur) [2] ; Ozkinay, F (Ozkinay, Ferda) [2] ; Atik, T (Atik, Tahir) [2] ...Less

View Web of Science ResearcherID and ORCID (provided by Clarivate)

Source EUROPEAN JOURNAL OF HAEMATOLOGY

Volume: 113 Issue: 1 Page: 82-89

DOI: 10.1111/ejh.14194

Published JUL 2024

Early Access MAR 2024

Indexed 2024-04-04

Document Type Article

Jump to ↓ Enriched Cited References

Abstract ObjectivesIn congenital hemolytic anemias (CHA), it is not always possible to determine the specific diagnosis by evaluating clinical findings and conventional laboratory tests. The aim of this study is to evaluate the utility of next-generation sequencing (NGS) and clinical-exome-based copy number variant (CNV) analysis in patients with CHA.MethodsOne hundred and forty-three CHA cases from 115 unrelated families referred for molecular analysis were enrolled in the study. Molecular analysis was performed using two different clinical exome panels in 130 patients, and whole-exome sequencing in nine patients. Exome-based CNV calling was incorporated into the traditional single-nucleotide variant and small insertion/deletion analysis pipeline for NGS data in 92 cases. In four patients from the same family, the PK Gypsy variant was investigated using long-range polymerase chain reaction.ResultsMolecular diagnosis was established in 86% of the study group. The most frequently mutated genes were SPTB (31.7%) and PKLR (28.5%). CNV analysis of 92 cases revealed that three patients had different sizes of large deletions in the SPTB and six patients had a deletion in the PKLR.ConclusionsIn this study, NGS provided a high molecular diagnostic rate in cases with rare CHA. Analysis of the CNVs contributed to the diagnostic success.

Keywords **Author Keywords:** copy number variation; hemolytic anemia; next-generation sequencing; PKLR; SPTB

Keywords Plus: PYRUVATE-KINASE DEFICIENCY; HB BROCKTON; DIAGNOSIS; MUTATIONS

Author Information

Corresponding Address: Isik, Esra (corresponding author)

- Ege Univ, Sch Med, Dept Pediat, Pediat Genet Subdiv, Izmir, Turkiye

E-mail Addresses :
esrabadak36@gmail.com

Addresses :

1. Ege Univ, Sch Med, Dept Pediat, Pediat Genet Subdiv, Izmir, Turkiye
2. Ege Univ, Fac Med, Dept Pediat, Div Pediat Genet, Izmir, Turkiye
3. Ege Univ, Fac Med, Dept Pediat, Div Pediat Hematol, Izmir, Turkiye
4. Ondokuz Mayis Univ, Fac Med, Dept Pediat, Div Pediat Hematol & Oncol, Samsun, Turkiye
5. Istanbul Univ, Fac Med, Dept Pediat, Div Pediat Hematol & Oncol, Istanbul, Turkiye

...more addresses

E-mail Addresses :
esrabadak36@gmail.com

Categories/ Classification

Research Areas: Hematology
Citation Topics: 1 Clinical & Life Sciences › 1.225 Hematologic Diseases › 1.225.786 Spectrin
Sustainable Development Goals: 03 Good Health and Well-being

Web of Science Categories Hematology

Funding

View funding text

Funding agency	Grant number	Show All Details
Ege University	17-TIP-006	Show details
Turkish Society of Hematology - Ege University Scientific Research Projects Coordination		

+ See more data fields

Journal information

EUROPEAN JOURNAL OF HAEMATOLOGY

ISSN	0902-4441
eISSN	1600-0609
Current Publisher	WILEY, 111 RIVER ST, HOBOKEN 07030-5774, NJ
Journal Impact Factor	Journal Citation Reports ™
Research Areas	Hematology
Web of Science Categories	Hematology

2.3
Journal Impact Factor ™ (2023)

0.56
Journal Citation Indicator ™ (2023)

Citation Network

In Web of Science Core Collection

22

Use in Web of Science

43