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CORRESPONDENCE

Neonatal Intestinal Perforations Due to IL10RB Deficiency

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To the Editor: A 2460 g male infant, one of twins born to consanguineous parents at 36 wk of gestation, presented at 12 h of age with sepsis-like features, including poor feeding, hypoactivity, and elevated inflammatory markers. Empirical antibiotics were initiated. Despite initial clinical improvement, recurrent fever and rising acute-phase reactants prompted cerebrospinal fluid analysis on day 7, which was consistent with meningitis; however, all cultures remained sterile. A 14-d course of broad-spectrum antimicrobial therapy was completed. On day 22, refractory fever and diarrhea developed. Although the initial influenza B test was positive, the severity of the clinical course and subsequent negative results argued against an isolated viral infection. The clinical course was further complicated by thrombocytopenia, leukopenia, hypoalbuminemia, and persistent systemic inflammation despite sterile cultures. Screening for hemophagocytic lymphohistiocytosis was unremarkable. By day 27, the patient required intubation and developed abdominal distension with bilious gastric aspirates; ultrasonography demonstrated bowel wall edema, ascites, and reduced peristalsis. Exploratory laparotomy on day 39 revealed multiple ileal perforations, intra-abdominal abscesses, and extensive adhesions. A 20-cm ileal segment was resected with ileostomy formation. Histopathology demonstrated severe chronic active transmural inflammation with necrosis. Systemic methylprednisolone resulted in significant but transient clinical improvement. A second laparotomy was required two weeks later due to duodenal perforation and ileojejunal necrosis. The infant died despite aggressive intensive care. Postmortem whole-exome sequencing identified a homozygous nonsense mutation in interleukin-10 receptor subunit beta (IL10RB) (NM_000628.5:c.477G>A; p.Trp159*), classified as pathogenic (PVS1, PM2, PP5), confirming autosomal recessive very early-onset inflammatory bowel disease.

This case highlights an extreme neonatal presentation of interleukin-10 receptor deficiency and expands its recognized clinical spectrum. Routine immunological evaluation may be unremarkable in such cases [1–3]. Early genetic testing—particularly in consanguineous families—is critical for timely diagnosis and consideration of hematopoietic stem cell transplantation as the only curative option [4].

Declarations

Conflict of Interest None.

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