

Clinical Image

Open Access, Volume 5

Bone marrow atypical hemophagocytosis with pancytopenia: Hemophagocytic lymphohistiocytosis or metabolic disorder?

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Received: Oct 08, 2024

Accepted: Oct 30, 2024

Published: Nov 06, 2024

Archived: www.jcimcr.org

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DOI: www.doi.org/10.52768/2766-7820/3328

Keywords: Pancytopenia; Inborn errors; Hemophagocytosis; Lysinuric protein intolerance; Bone marrow.

Description

A 10-month-old girl, was admitted to the pediatric department for early-onset pancytopenia. Digestive disorders were noted in her history. Clinically, she was non-dysmorphic and eutrophic, with findings of hepatomegaly and a spleen tip; the rest of the examination was unremarkable.

Laboratory tests revealed pancytopenia with normochromic normocytic anemia (hemoglobin = 10 g/dL), leukoneutropenia (White Blood Cells = $3.1 \times 10^9/L$; Neutrophils = $0.8 \times 10^9/L$), and thrombocytopenia (Platelets = $111 \times 10^9/L$).

Additionally, hepatic cytolysis (Aspartate Aminotransferase = 96 U/L), hemolysis markers (Lactate Dehydrogenase = 1500 U/L, Haptoglobin < 0.1 g/L), elevated ferritinemia (2076 µg/L), and hypernatremia (190 mmol/L) were observed. Immunoglobulin assays, immunological tests, and viral tests were negative. Abdominal ultrasound showed isolated homogeneous hepatosplenomegaly.

Bone marrow examination revealed a normocellular marrow. There was an increased presence of apoptotic neutrophils (Figure 1A, all images x100, May Grünwald Giemsa stain). Additionally, selective phagocytosis of naked nuclei by immature granular cells was observed (Figure 1B and 1C), an abnormal process where these cells engulfed the nuclei. Numerous hemophagocytic images were present (Figure 1B and 1D), where histiocytes displayed cytoplasm filled with engulfed erythroblasts.

These findings pointed towards a rare metabolic disorder, the lysinuric protein intolerance. This rare condition is characterized by a defective transporter of dibasic amino acids (arginine, ornithine, lysine), leading to their deficient plasma levels and impaired urea cycle function [1]. Diagnosis is confirmed by decreased plasma levels of these amino acids, their increased urinary excretion, and identification of mutations in the SLC7A7 gene [2].

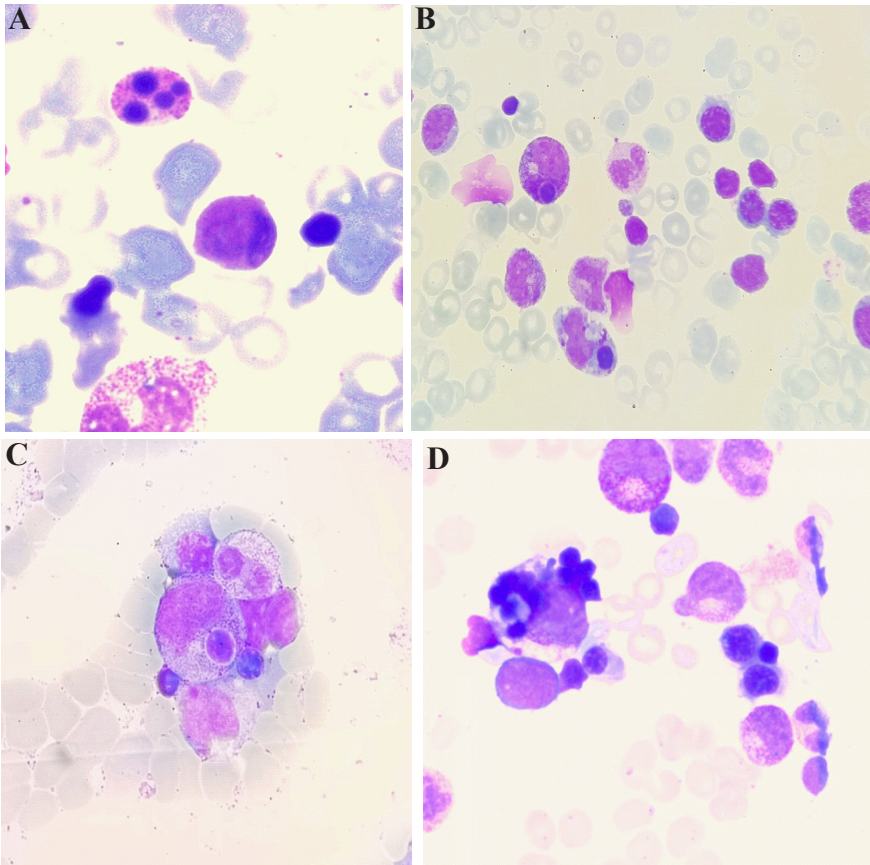


Figure 1: Clinical image.

Statements and declarations

The authors declare that our data collection does not require ethics committee approval.

Verbal consent was obtained from the parents before data collection.

Conflict of interest: The authors declare that there is no conflict of interest to disclose.

Funding: The authors declare that the study received no funding.

The data that support the findings of this study are available from the corresponding author upon reasonable request.

References

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