Article published online: 2025-01-30

Cytoplasmic Vacuoles in Lymphocytes of a Child with Sialic Acid Storage Disorder

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Ibnosina J Med Biomed Sci

Abstract

This is an image-based clinical vignette demonstrating unusual cytoplasmic vacuoles in the lymphocytes in a stained peripheral blood smear from a 6-month-old infant who presented with seizures, hypotonia, and hepatosplenomegaly and was found to have the extremely rare lysosomal disorder, namely, sialic acid storage disease. Although not specific, it is important to recognize this morphologic finding for reflex biochemical and genetic testing.

Keywords

- hypomyelination
- infantile sialic acid storage disorder
- lysosomal membrane transporter
- N-acetylneuraminic acid
- sialic acid

Image and Case Description

This is a composite of photomicrographs showing lymphocytes with cytoplasmic vacuoles noted upon review of a stained peripheral blood smear from a 6-month-old infant who presented with a complaint of seizures and was found to have hypotonia and hepatosplenomegaly (►**Fig. 1**). At the time of review, the complete blood count (CBC) data included the following: white blood cell (WBC) = 6.5×10^9/L; hemoglobin = 14.2 mg/L; and platelets = 86×10^9/L. A storage disease was suspected and workup to establish the diagnosis included total sialic acid (18,966 nmol/mg) and free sialic acid (18,575 nmol/mg), both of which were significantly elevated.

Comments

Sialic acid storage disease is a rare lysosomal disorder, inherited as an autosomal recessive condition, rendering cells unable to clear sialic acid.[1] An underlying genetic defect in 1 of more than 40 possible genes that control this process can result in intracellular accumulation/storage of the sugar N-acetylneuraminic acid (sialic acid) in multiple tissues, including the central nervous system, heart, muscles, and lymphocytes.[2] While the presence of rare cytoplasmic vacuoles in lymphocytes is not specific, their increased number and the context of suggestive clinical history may help narrow the differential diagnosis. Similar vacuoles in the cytoplasm of lymphocytes may also be seen in ceroid lipofuscinoses (Batten's disease), other lipid storage diseases (e.g., Wolman's disease), glycogen storage diseases (e.g., Pompe's disease), and mucopolysaccharidoses (e.g., Hurler's syndrome).[3] A specific diagnosis relies on biochemical and/or genetic testing.[4]

Conclusions

This is an image-based clinical vignette that demonstrates unusual cytoplasmic vacuoles in the lymphocytes in a stained peripheral blood smear from a 6-month-old infant who presented with seizures, hypotonia, and hepatosplenomegaly and was found to have the extremely rare

DOI https://doi.org/10.1055/s-0044-1801759.
ISSN 1947-489X.

Thieme Medical and Scientific Publishers Pvt. Ltd., A-12, 2nd Floor, Sector 2, Noida-201301 UP, India

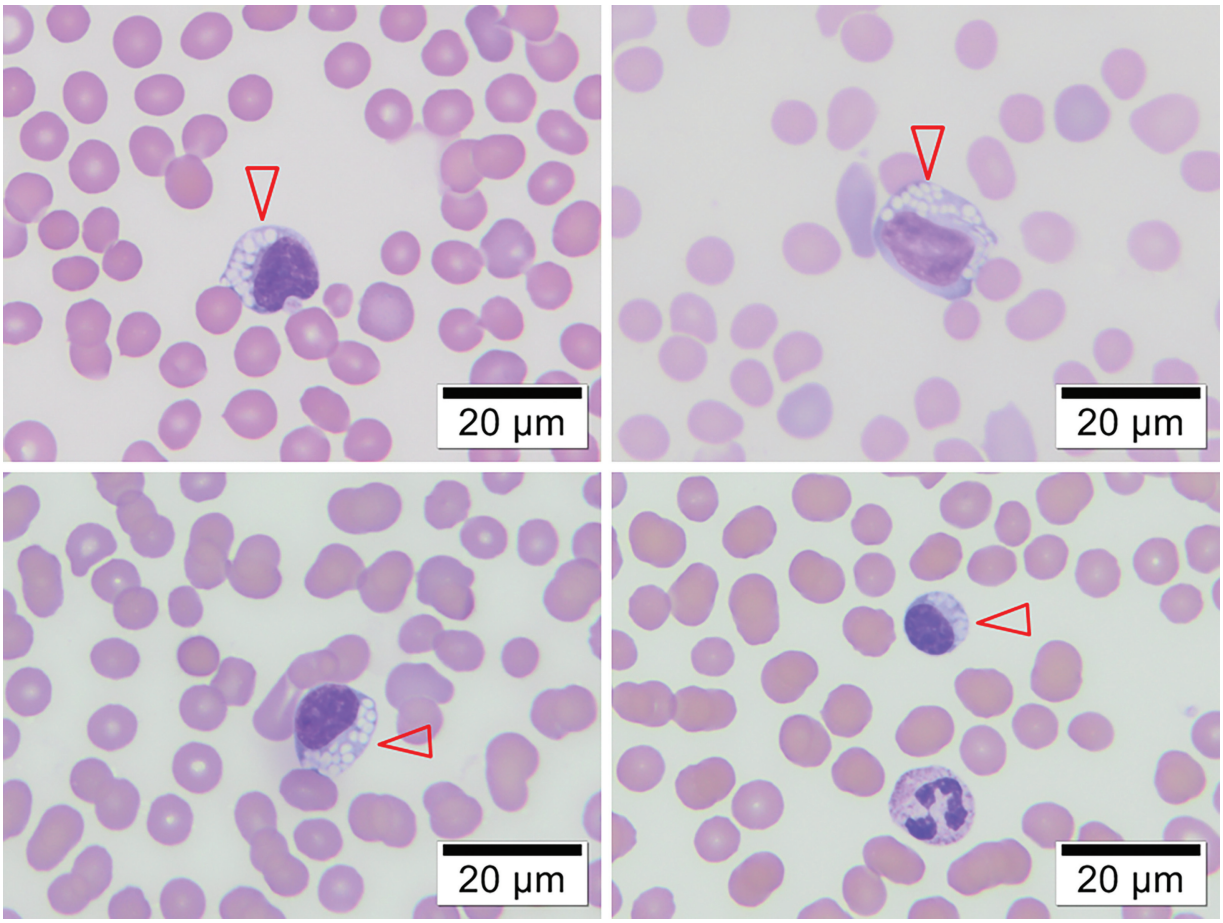

Fig. 1 Peripheral smear showing lymphocytes with multiple vacuoles in the cytoplasm. Figure magnification: ×100 oil.

lysosomal disorder, sialic acid storage disease. Although not specific, it is important to recognize this morphologic finding for reflex biochemical and genetic testing.

Authors' Contributions

S.B.K. made the diagnosis, wrote the first draft, and prepared the composite photograph. S.S. performed the literature search and edited the manuscript.

Compliance with Ethical Principles

No ethical concerns.

Funding and Sponsorship

None.

Conflict of Interest

None declared.

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