Received: 26 September 2023 | Accepted: 18 December 2023

DOI: 10.1002/jha2.844

eJHaem

HAEMATOLOGY IMAGES

Pediatric bone marrow with wrinkled histiocytes

Luis F. Carrillo[1] | Daisy Alapat[2]

[1]Department of Pathology, Duke University, Durham, North Carolina, USA

[2]Department of Pathology, University of Arkansas for Medical Sciences (UAMS), Little Rock, Arkansas, USA

Correspondence
Luis F. Carrillo, Department of Pathology, Duke University Medical Center, 40 Duke Medicine Circle, Davison Building, Room 246AM, Durham, NC 27710, USA.
Email: luis.carrillo@duke.edu

An 8-year-old female child presented with leg pain and pancytopenia. Her medical history was unremarkable. Upon physical examination, the patient demonstrated hepatosplenomegaly without neurological deficits. Radiology studies were concerning for a marrow infiltrative process. Bone marrow biopsy was normocellular and revealed prominent histiocytic hyperplasia in the background of trilineage hematopoiesis (Figure 1, panel A; original magnification 200×; hematoxylin and eosin [H&E] stain). Histiocytes contain abundant eosinophilic cytoplasm that appears fibrillar, resembling wrinkled tissue paper (Figure 1, panel B; original magnification 1000×). Bone marrow aspirate also showed histiocytes with abundant blue-gray cytoplasm and wrinkled paper-like fibrillar pattern. The nucleus is small, with coarse chromatin, irregular nuclear contour, and indistinct nucleolus (Figure 1, panel B, inset, original magnification 1000×; Wright–Giemsa stain). CD68 immunohistochemical stain highlights histiocytes (Figure 1, panel C; original magnification 500×). PASD (Periodic acid-Schiff with diastase) stain demonstrates a striated cytoplasm that is reddish-magenta within the histiocytes (Figure 1, panel D; original magnification 500×). These findings were consistent with storage disorders, such as Gaucher disease. Low beta-glucosidase activity of 0.176 nmol/mL blood/h (reference range, 1.69–22.6) and genetic testing by single-gene analysis revealed heterozygous mutations in exon 11 c.1448T>C (p.Leu483Pro) and exon 9 c.1214G>A (p.Ser405Asn) of the *GBA1* gene, confirming the diagnosis. Subsequently, the patient started treatment with Imiglucerase.

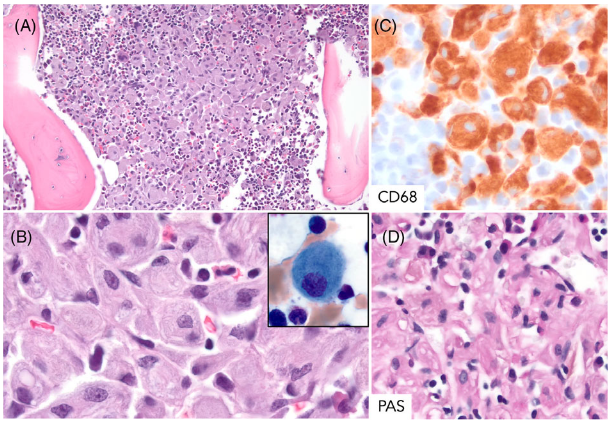

FIGURE 1 (A) Core biopsy (hematoxylin and eosin [H&E] 200×): normocellular for age (90% cellularity) with prominent histiocytic hyperplasia in the background of trilineage hematopoiesis. (B) Core biopsy (H&E 1000×): histiocytes contain abundant eosinophilic cytoplasm that may appear fibrillar. Inset: bone marrow aspirate (Wright–Giemsa stain 1000×) histiocytes with abundant blue-gray cytoplasm and wrinkled paper-like fibrillar pattern. The nucleus is small with coarse chromatin, irregular nuclear contour, and indistinct nucleolus. (C) CD68 immunohistochemical stain highlights histiocytes. (D) PASD (Periodic acid-Schiff with diastase) stain demonstrates a reddish-magenta color within the histiocytes' cytoplasm.

CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

FUNDING INFORMATION

The author received no financial support for the research, authorship, or publication of this article.

Luis F. Carrillo and Daisy Alapat contributed equally to this study.

CLINICAL TRIAL REGISTRATION

The authors have confirmed clinical trial registration is not needed for this submission.

DATA AVAILABLITY STATEMENT

Data sharing is not applicable to this article as no new data were created or analyzed in this study.

ETHICS STATEMENT

The information presented in this manuscript is de-identified, and there is minimal risk to the patient's privacy or confidentiality. No material from other sources is included in this manuscript.

ORCID

Luis F. Carrillo https://orcid.org/0000-0003-1045-7971

How to cite this article: Carrillo LF, Alapat D. Pediatric bone marrow with wrinkled histiocytes. eJHaem. 2024;1–2. https://doi.org/10.1002/jha2.844