Images from the Haematologica Atlas of Hematologic Cytology: Gaucher disease

Rosangela Invernizzi
University of Pavia, Pavia, Italy
E-mail: rosangela.invernizzi@unipv.it
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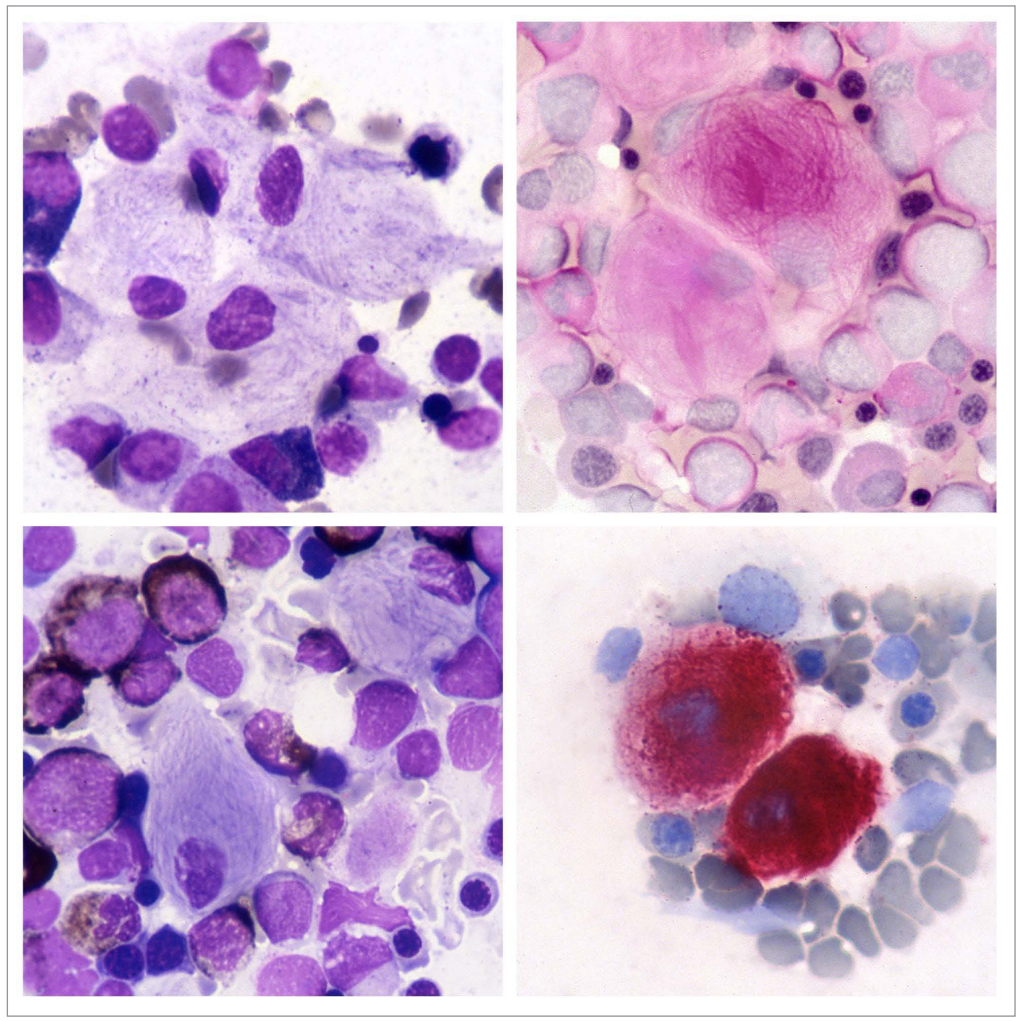

Gaucher disease is an inherited lysosomal storage disease characterized by the accumulation of glucocerebrosides (glucosylceramide), in macrophages of liver, spleen, and bone marrow. This metabolic disorder results from a defect in the lysosomal β-glucocerebrosidase enzyme due to gene mutations. The morphological features of pathological macrophages in the bone marrow are shown in the figure: they are distinctive and important for the diagnosis. Gaucher cells are large cells with a small, round, usually eccentric nucleus and abundant weakly basophilic cytoplasm with a fibrillar or 'onion-skin' appearance (top left). The fibrillar appearance is due to the fact that lysosomes are elongated for lipid accumulation. Gaucher cells have a tendency to occur in groups. They are best seen in the thicker portions of bone marrow smears, i.e. at the ends and margins. Periodic acid Schiff (PAS) staining for polysaccharides shows strong positivity in Gaucher cells and highlights the fibrillar pattern of the cytoplasm (top right); normal PAS positivity is observed in neutrophils. Myeloid cells are normally positive for Sudan black stain, whereas Gaucher cells are negative (bottom left). They exhibit strong reactivity for acid phosphatase (bottom right) that is tartrate-resistant. The diagnosis of Gaucher disease, suggested by morphological findings, should, however, be confirmed by assay of peripheral blood leukocytes for the β-glucocerebrosidase enzyme that is absent or very reduced. Measurement of enzyme levels may then be supplemented by mutational analysis.[1]

Disclosures
No conflicts of interest to disclose.

References

1. Invernizzi R. Storage diseases. Haematologica. 2020; 105(Suppl 1):255-260.