

Images in hematology-oncology

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A 23-year-old man with thrombocytopenia

A 23-year-old man presented with thrombocytopenia. Physical examination was unremarkable. The blood analysis revealed hemoglobin 17.6 g/dl, leukocyte count 10,300/mm³, platelet count 104,000/mm³. Computed Tomography (CT) of the abdomen was normal. The patient underwent a bone marrow biopsy.

In the microscopic examination, bone marrow sections exhibited enlarged macrophages present in clusters and aggregates. Immunohistochemically these cells expressed

CD68 and the cytoplasm was strongly PAS positive. The bone marrow aspirate showed a significant number of histiocytes with a pale blue to gray cytoplasm which showed a fibrillary pattern resembling wrinkled tissue paper.

What is your diagnosis?

Probable Gaucher disease, requires assay of enzyme levels or genotypic confirmation

DISCUSSION

Gaucher disease is an autosomal recessive, lysosomal storage disease, characterized by glycosylcerebroside deposition in reticulo-endothelial cells, due to deficiency of lysosomal glucocerebrosidase. The disease is the most common lysosomal storage disorder. Three clinical subtypes of Gaucher disease have been distinguished, with different degrees of clinical severity. The most common is type I (chronic non-neuronopathic form), type II (acute neuronopathic form) and type III (intermediate between types I and II) (1).

Histopathologically in the bone marrow, there is an accumulation of macrophages engorged with the excess metabolic product. These macrophages have a distinctive appearance, with a pale blue cytoplasm in the standard Wright-Giemsa stain and a delicate “folded tissue paper” appearance of the cytoplasm. The cells are strongly PAS positive (2).

For all types of storage disease, analyses of blood mononuclear cells or fibroblasts for enzyme activity is

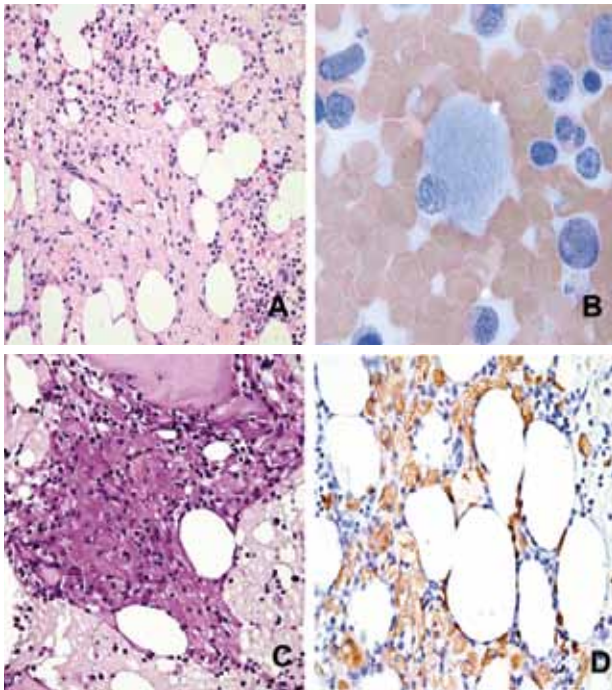


Fig 1 (A&B&C&D). (A): The macrophages forming large aggregates; (B): “Folded tissue paper” appearance of the cytoplasm; (C): PAS positivity and (D): CD68 expression in the histiocytes

required for diagnosis. Morphology of bone marrow histiocytes alone is not sufficient because, in some patients, these cells are not prominent in the bone marrow and

because other bone marrow conditions characterized by high rates of cell turnover may also exhibit storage disease-type histiocytes (3).

References

1. Ramzi S. Cotran, Vinay Kumar, Tucker Collins. Pathologic Basis for Disease. W.B. Saunders Company, 1999.
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