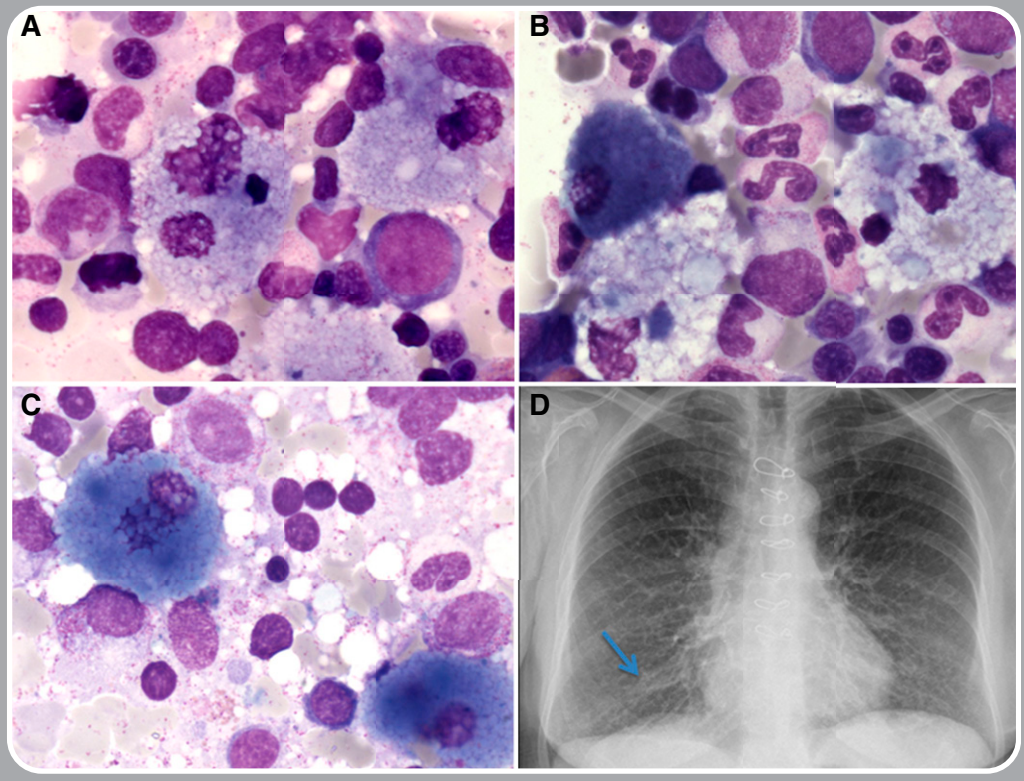
bloodwork
IMAGES IN HEMATOLOGY

Bone marrow smear examination in the diagnosis of Niemann-Pick B disease



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A 73-year-old woman with a medical history of interstitial lung disease, aortic valve surgery, and coronary artery bypass surgery was referred for possible myeloma or lymphoma. Evaluation showed moderate thrombocytopenia (150×10^9/L), monoclonal gammopathy (immunoglobulin G kappa, 2.18 g/L), and hepatosplenomegaly on scans. Bone marrow aspirate (May-Grünwald-Giemsa stain) was infiltrated by numerous foam cells (panels A-B; magnification ×1000) and sea blue histiocytes (panels B-C; magnification ×1000). The foam cells showed cytoplasm filled with numerous small vacuoles. These findings are typical of type B Niemann-Pick disease (NPD). The diagnosis was confirmed by the markedly decreased acid sphingomyelinase activity, whereas the acid β-glucosidase activity was normal, excluding type 1 Gaucher disease. Gene sequencing revealed homozygosity for the mutation encoding p.RΔ610 in the acid sphingomyelinase. Moreover, the chest radiograph showed a typical granular density (panel D).

This is an unusual case of an elderly woman with type B NPD with striking bone marrow findings, suggesting the diagnosis. Although pseudo-Gaucher cells and sea blue histiocytes can be found in bone marrow that is rapidly turning over, the combination of both numerous foam cells and sea blue histiocytes suggests the diagnosis of type B NPD, which should be confirmed by demonstrating the enzyme deficiency and/or gene mutations.

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