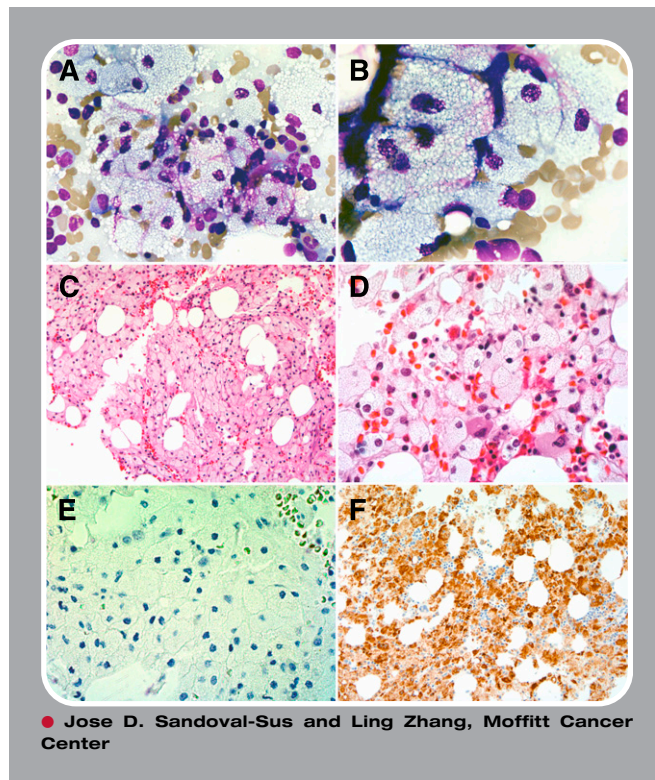


## Familial hypertriglyceridemia manifests with pancytopenia and bone marrow pseudo-Niemann-Pick cells



**A**n asymptomatic 51-year-old man with familial hypertriglyceridemia presented with pancytopenia (white blood cells,  $2.8 \times 10^9/L$ ; hemoglobin, 11.9 g/L; and platelets,  $77 \times 10^9/L$ ). His only medications were 3 antihyperlipidemic drugs. Lymphadenopathies, hepatosplenomegaly, and/or bleeding were absent. Aside from elevated triglycerides (1739 mg/dL; normal range, 150-199 mg/dL), an extensive diagnostic workup was unremarkable. Bone marrow aspirate revealed panhypoplasia and increased histiocytes, which were remarkably large with abundant cytoplasmic accumulation of foamy or lipid components resembling Niemann-Pick cells (panels A-B, Wright-Giemsa; magnification  $\times 600$  and  $\times 1000$ , respectively). About 70% to 80% of marrow space was replaced with the abnormal histiocytes (panels C-D, hematoxylin and eosin; magnification  $\times 200$  and  $\times 600$ , respectively). There was no evidence of hemophagocytosis, dysplasia, or blastosis. On immunohistochemistry, the histiocytes were CD1a<sup>-</sup> (panel E; magnification  $\times 600$ ), CD68<sup>+</sup> (panel F; magnification  $\times 600$ ), and S-100<sup>-</sup>. Flow cytometry, cytogenetics, and stains for acid-fast bacilli and Gomori methenamine silver were normal. Finally, glucocerebrosidase and sphingomyelinase activity, and mutation analysis, excluded Gaucher and Niemann-Pick diseases.

Pseudo-Niemann-Pick cells (PNPCs) have characteristic vacuolated cytoplasm instead of the “striated” one of pseudo-Gaucher cells (PGCs). Also, PNPCs are seen in lipid diseases whereas PGCs are found in high cell turnover disorders (eg, acute leukemias, multiple myeloma, Langerhans cell histiocytosis, and mycobacterial infection). Familial hypertriglyceridemia with pancytopenia due to PNPC marrow infiltration has not been previously reported.



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