Teaching Video NeuroImages:

Vertical supranuclear ophthalmoparesis

A diagnostic pearl for Niemann-Pick C

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Figure Bone marrow biopsy

(A and B) Photomicrograph from bone marrow aspirate shows presence of large cells with abundant amount of cytoplasm filled with multiple vacuoles (red arrows) suggesting the morphology of Niemann-Pick disease (May-Grünwald-Giemsa stain, $\times 1,000$). (C) Similar cells (blue arrows) are also observed in the bone marrow trephine biopsy section (hematoxylin & eosin, $\times 1,000$).

A 30-year-old man presented with 10 years of progressive cognitive decline (poor academic performance followed by forgetfulness and calculation and visuospatial difficulties), 5 years of gait and limb ataxia and dysarthria, and 3 years of dysphagia. His 2 elder siblings had similar symptoms and died in their 20s. Examination revealed splenomegaly, global cognitive impairment, brisk gag reflex, pancerebellar ataxia, bipyramidal signs, and vertical saccade paresis (video on the *Neurology*[®] Web site at Neurology.org). Vertical pursuit was preserved. Bone marrow aspirate and trephine biopsy revealed non-Gaucher-type storage foam cells (figure). Considering the diagnosis of Niemann-Pick C, he was treated symptomatically. Vertical supranuclear ophthalmoparesis is a red flag suggestive of Niemann-Pick C in young-onset dementia.^{1,2}

AUTHOR CONTRIBUTIONS

A.G. collected information, prepared the draft, and reviewed the manuscript. N.K. collected information and prepared the draft. R.S. collected the information and prepared the draft. P.R. collected information and prepared the draft. V.Y.V. prepared the draft and reviewed the manuscript. V.L. edited and reviewed the manuscript. All authors reviewed, edited, and approved the final version.

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DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

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