RESIDENT & FELLOW SECTION

Section Editor
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Teaching NeuroImages: Hypertrophic olivary degeneration in a young man with *POLG* gene mutation

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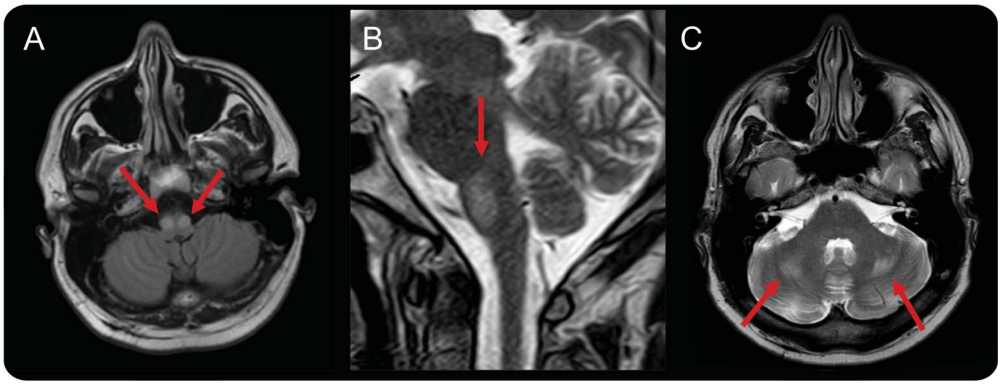
Figure Abnormal findings on MRI



Symmetric hyperintense signal abnormality and enlarged inferior olives on (A) fluid-attenuated inversion recovery and (B) T2 images, characteristic of bilateral hypertrophic olivary degeneration. Additional findings included hyperintense T2 signal changes in the thalami and deep cerebellar nuclei and mild cerebellar vermis atrophy (C).

A 30-year-old man with sensorineural hearing loss presented with subacute somnolence, slurred speech, and unsteady gait following treatment with peginterferon α-2b and ribavirin for chronic hepatitis C virus. Examination revealed scanning speech, horizontal nystagmus, gait ataxia, and symmetric hyporeflexia with distal sensory loss. There was no palatal myoclonus. Metabolic and serologic workup and blood lactate were unrevealing. Brain MRI demonstrated bilateral hypertrophic olivary degeneration (HOD, figure). Whole exome sequencing identified a homozygous pathogenic p.W748S *POLG* mutation.[1] Differential diagnosis of bilateral HOD includes mutations in the nuclear genes crucial to mitochondrial function, *POLG* and *SURF1*.[2]

AUTHOR CONTRIBUTIONS

D.A. had substantial contributions to conception and design of the work, acquisition and interpretation of data, drafting the work, and final approval of the submitted version. V.M. had substantial contributions to conception and design of the work, acquisition and interpretation of data, drafting the work, and final approval of the submitted version. A.K. had substantial contributions to conception and design of the work, acquisition and interpretation of data, drafting the work, and final approval of the submitted version. A.L. had substantial contributions to conception and design of the work, acquisition and interpretation of data, drafting the work, and final approval of the submitted 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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