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