Teaching NeuroImages: Neuroferritinopathy

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Figure Brain imaging

(A) Cavitation of the basal ganglia on axial CT scan of the brain. (B) Axial susceptibility-weighted MRI shows iron deposition in basal ganglia.

A 51-year-old music teacher presented with difficulty playing his flute and mild dysarthria related to impaired tongue movements. Over the subsequent 11 years, he developed fluctuating chorea and bradykinesia, stereotypic foot tapping, and a grossly unsteady gait, resulting in him becoming chairbound 9 years later. He further developed profound dysarthria and dysphagia. Cognitive impairment was not observed. There is a strong family history, with the patient's father and 3 siblings also affected by this movement disorder.

CT scanning showed cavitation of the basal ganglia while susceptibility-weighted MRI demonstrated iron accumulation in basal ganglia, cerebellum, and

cerebral cortex (figure). DNA analysis revealed the common 460InsA mutation for neuroferritinopathy, an autosomal dominantly transmitted condition caused by mutation of the ferritin light chain gene.¹

AUTHOR CONTRIBUTIONS

Dr. Lehn: drafting/revising the manuscript, analysis or interpretation of data. Dr. Mellick: drafting/revising the manuscript, study concept or design, analysis or interpretation of data, acquisition of data, study supervision. Dr. Boyle: drafting/revising the manuscript.

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