Teaching NeuroImage:

Cerebrotendinous xanthomatosis

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A 57-year-old man with cerebrotendinous xanthomatosis (CTX) was admitted to the hospital after a fall. He had been diagnosed with CTX in his 30s and had had numerous complex-partial seizures, which occur in 50% of adult patients.¹ Seizures were characterized by staring spells and speech deficits, occasionally generalizing to tonic-clonic leg movements. He had marked enlargement of the tongue and Achilles tendons (figure, A–D). Neurologic examination was notable for dementia, spasticity, and ataxia. Brain MRI revealed lesions in the temporal lobes, globus pallidus, and dentate nucleus of the cerebellum (figure, E, F), thought to be from lipid accumulation and reactive astrocytosis.² Additionally, hemosiderin deposits with calcification were present in the cerebellar hemispheres (figure, G, H). Bio-

chemical testing revealed a high plasma cholestanol level (3.04 mg/dL, >10 times normal). The patient had been treated with chenodeoxycholic acid, but did not receive it for over a year because of short supply worldwide. Lack of recent therapy was associated with an increased frequency of seizures, prominent tongue protrusion, and further enlargement of the Achilles tendons.

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Figure Patient with cerebrotendinous xanthomatosis

Physical examination demonstrated enlargement of the tongue (A) and Achilles tendons (B). Axial (C) and sagittal (D) T1-weighted MRI scans confirmed hypertrophy of the Achilles tendons. Brain MRI FLAIR sequences showed cortical and subcortical hyperintensities in the temporal lobes (arrows, E) and globus pallidus (arrowheads, E). T2-weighted MR images revealed cerebellar hyperintensities within the dentate nucleus (F). Hypointensities were seen on T1-weighted and susceptibility MRI scans within the cerebellum at the level of the midbrain (G, H).

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