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. 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. Additionally, hemosiderin deposits with calcification were present in the cerebellar hemispheres (figure, G, H). Biochemical 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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