Figure. Cerebrotendinous xanthomatosis (CTX) is a rare autosomal recessive disorder caused by mutations of the sterol 27-hydroxylase gene leading to deposition of cholestanol and other sterols. Cranial MRI typically shows gliosis of cerebellar dentate nuclei (gray arrow) surrounded by liquefied defects (white arrows). Chromatography detected increased serum cholestanol and cholest-8-en-3β-ol, as well as bile alcohols in urine. CTX is one of the very few treatable causes of progressive ataxia. Patients benefit from early therapy, and progression of the disease can be prevented.

Cerebrotendinous xanthomatosis: A treatable ataxia

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A 44-year-old man presented with progressive cerebellar ataxia and spastic paraparesis (see the video on the *Neurology* Web site). Gait difficulties had started at age 16 years, and at age 42 years bilateral cataracts were diagnosed. His cognitive function

progressively declined, and seizures repeatedly occurred. Although diarrhea and tendon xanthomas were not overt, clinical and laboratory investigations excluded the differential diagnosis of the autosomal recessive Marinesco-Sjögren-syndrome (early-onset ataxia with cataracts and cognitive impairment). Cerebrotendinous xanthomatosis was diagnosed on the basis of cranial MRI and increased serum cholestanol levels (figure), slow theta activity in EEG, and peripheral neuropathy. Daily treatment with 1,000 mg of chenodeoxycholic acid and 10 mg of simvastatin was initiated.^{1,2} Follow-up examinations showed normal sterol metabolism after 25 weeks (figure) and no further neurologic deterioration at 10 months.

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