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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