Research (Drs. Nishino and Nonaka), National Institute of Neuroscience, National Center of Neurology and Psychiatry; and Second Department of Internal Medicine (Dr. Katayama), Nippon Medical School, Tokyo, Japan.

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Address correspondence and reprint requests to Dr. S. Amemiya, Division of Neurology, Second Department of Internal Medicine, Nippon Medical School, 1-1-5, Sendagi, Bunkyo-ku, Tokyo 113-8603, Japan; e-mail: amemy-@wc4.so-net.ne.jp

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Neuro*Images*

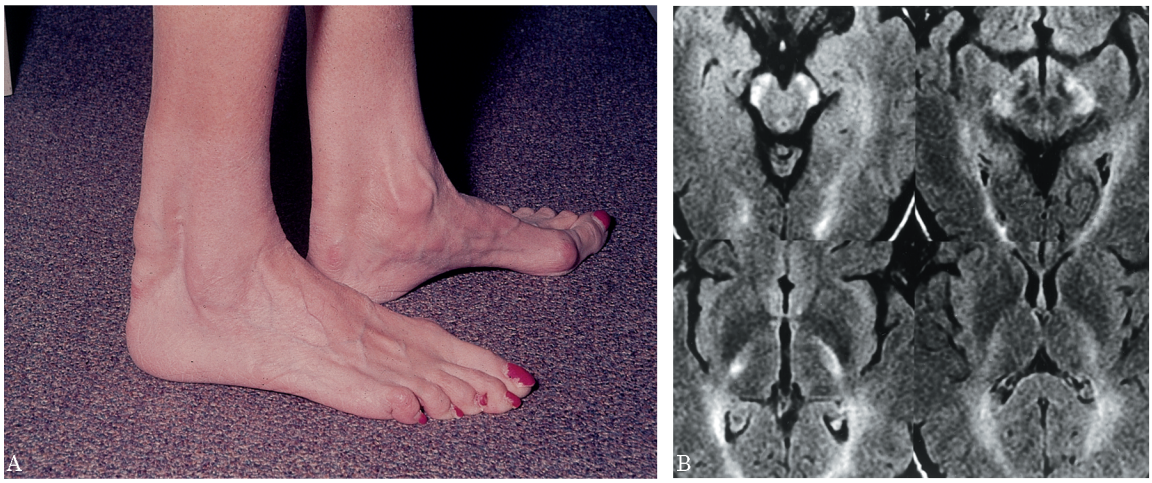

Figure. (A) Bilateral Achilles tendon xanthomas. (B) Axial FLAIR MRI images show abnormally hyperintense signal involving the corticospinal tracts at the brainstem level, the white matter of both internal capsules, and the peritrigonal white matter.

Cerebrotendinous xanthomatosis

James D. Fleck, MD, José Biller, MD, Vincent P. Mathews, MD, Indianapolis, IN

A 58-year-old woman presented with a several-year history of progressive gait difficulties. Her medical history was noteworthy for bilateral cataracts diagnosed in her thirties with subsequent surgical replacement. Physical examination showed nodular thickening of both Achilles tendons and high-arched feet (figure, A). Neurologic examination showed mild spasticity in the lower extremities, normal power, diffuse hyperreflexia, a left Babinski sign, moderate truncal ataxia, mild lower extremity ataxia, stocking distribution sensory loss to light touch and proprioception in the legs, and a markedly wide-based and ataxic gait. Neuropsychological testing showed widespread neurocognitive deficits with poor attention and concentration and seriously impaired memory. Plasma cholestanol was elevated (1.6 mg/dL; reference range, 0.2 ± 0.2 mg/dL). She was treated with chenodeoxycholic acid.

Cerebrotendinous xanthomatosis is a rare autosomal recessive lipid storage disease that results in excessive cholestanol deposition in body tissues, including the central and peripheral nervous systems. Its biochemical basis is a defective activity of the mitochondrial enzyme sterol 27-hydroxylase that results in defective bile acid synthesis. Long-term treatment with oral chenodeoxycholic acid typically corrects the biochemical abnormalities and may improve neurologic function.[1]

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