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Teaching NeuroImages: Progressive asymmetric parkinsonism and tendon xanthomas

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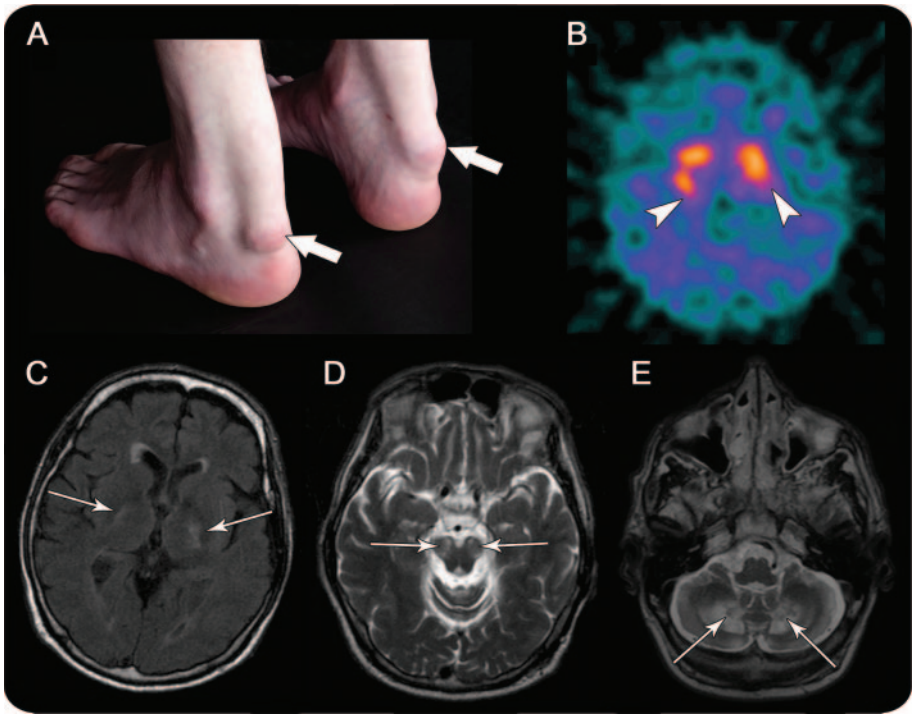
Figure Clinical and radiologic signs



(A) Bilateral tendon xanthomas (arrows). (B) I-123-Ioflupane (I-123 FP-CIT) SPECT: reduced bilateral, asymmetric putaminal uptake (arrowheads). MRI (arrows): diffuse volume loss with signal abnormality, (C) in the globus pallidus, internal capsules on axial fluid-attenuated inversion recovery, (D) cerebral peduncles, substantia nigra, and (E) extensive white matter involvement of the cerebellar hemispheres including dentate nuclei on axial T2-weighted imaging.

A 45-year-old man presented with rapidly progressive gait and balance deterioration, bradykinesia, and speech and swallowing difficulties. He had longstanding cognitive impairment and bilateral cataract surgery in childhood. On examination he had asymmetric parkinsonism with pyramidal signs and bilateral Achilles tendon xanthomas (figure). Imaging at age 43 (figure) and increased urinary bile alcohols confirmed cerebrotendinous xanthomatosis (CTX).[1,2] Treatment was started with chenodeoxycholic acid but clinical deterioration continued. Cocareldopa was added at age 45 with transient benefit.

CTX is an autosomal recessive disorder caused by reduced mitochondrial sterol-27-hydroxylase activity (*CYP27A1* gene), leading to accumulation of cholestanols. Features include infantile diarrhea, childhood cataracts, xanthomas, and psychiatric and neurologic abnormalities. While clinical improvement has been described with chenodeoxycholic acid, statins, and

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levodopa for patients with parkinsonian features, other patients continue to progress.[1]

AUTHOR CONTRIBUTIONS

Olga Kirmi: primary author, organization, image collection. Elaine Murphy: author, clinical input, clinical photograph. Miryam Carecchio: author, clinical input. Tom Sulkin: figure author and interpretation of DAT scan and MRI. Julia Rankin: interpretation of clinical and biochemical findings, diagnosis. Fergus Robertson: author, neuroradiology input, MR interpretation, supervisor.

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