

Teaching NeuroImages: Cerebrotendinous xanthomatosis

A rare treatable adult-onset lipid storage disease

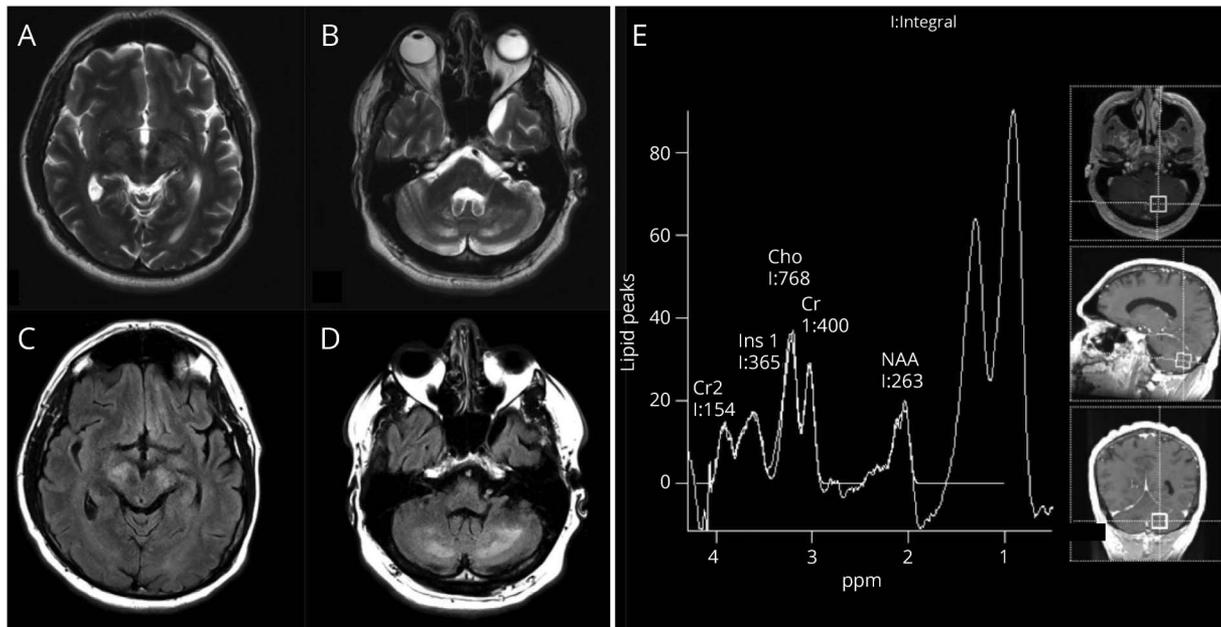
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Figure Brain MRI and spectroscopy demonstrate characteristic findings of cerebrotendinous xanthomatosis



(A, B) Axial T2-weighted and (C, D) fluid-attenuated inversion recovery images demonstrate T2 hyperintensities involving the thalami, midbrain, dentate nuclei, and deep cerebellar white matter. (E) Magnetic resonance spectroscopy reveals typical lipid peaks at 0.9 and 1.3 ppm. Cho = choline; Cr = creatine; NAA = *N*-acetylaspartate.

A 39-year-old previously healthy man presented with insidiously progressive paresthesia in his lower extremities and worsening of gait and balance. MRI demonstrated T2-hyperintense signal abnormalities involving the thalami, midbrain, dentate nuclei, and adjacent deep cerebellar white matter, which are characteristic of cerebrotendinous xanthomatosis (CTX) (figure). Magnetic resonance spectroscopy revealed typical lipid peaks at 0.9 and 1.3 ppm,¹ and serology analysis indicated sterol 27-hydroxylase deficiency,² confirming the diagnosis. CTX is a rare lipid storage disease with unique clinical and imaging findings, and effective treatment with positive outcomes if initiated early, raising the importance of awareness and early recognition of this disease.²

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Author contributions

Mina S. Makary: study concept, analysis of MRI data, drafting and revising the manuscript, final approval. Yaz Y. Kisanuki, Nessim N. Amin: study concept, revising the manuscript, final approval. Hasel W. Slone: study concept, study supervision, analysis of MRI data, revising the manuscript, final approval.

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