Teaching *NeuroImages*: Early-onset dementia and demyelinating neuropathy disclosing cerebrotendinous xanthomatosis

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Figure Examination and neuroimaging findings in cerebrotendinous xanthomatosis

(A) Left wrist with tendon xanthoma (arrow). Axial brain MRI study shows global cortical atrophy with mild bilateral parieto-occipital gliosis in T2-weighted (B) and fluid-attenuated inversion recovery sequences (C). (D) Tc-99m brain SPECT scan shows hypoperfusion involving the frontal and temporal lobes and basal ganglia.

A 40-year-old woman presented with a 5-year history of numbness in hands and feet, apathy, compulsive behavior, and aggression towards others. Medical history disclosed juvenile-onset cataracts. Examination disclosed tendinous xanthoma. Electroneuromyography disclosed sensorimotor demyelinating polyneuropathy. Neuroimaging studies showed global cortical atrophy. Tc-99m brain SPECT scan showed marked hypoperfusion involving the frontal and temporal lobes (figure). Plasma cholestanol levels and bile alcohol glycoconjugates were elevated, enabling a diagnosis of cerebrotendinous xanthomatosis (CTX).

CTX is an autosomal recessive disorder of bile acid biosynthesis resulting from sterol 27-hydroxylase deficiency.¹ Neurological signs include cerebellar ataxia, spasticity, epilepsy, demyelinating neuropathy,¹ and early-onset dementia, mimicking features of frontotemporal dementia.²

AUTHOR CONTRIBUTIONS

Dr. Souza: case report project conception, organization, and execution, writing of the first draft. Dr. Bortholin: case report project conception, organization, and execution, writing of the first draft. Dr. Naylor: case report project conception and organization, writing of the first draft. Dr. Pinto: case report project conception, writing of the first draft, manuscript review and critique. Dr. Oliveira: manuscript review and critique.

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