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Chinese patient with cerebrotendinous xanthomatosis confirmed by genetic testing:

A case report and review of literature

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Abstract

BACKGROUND

Cerebrotendinous xanthomatosis (CTX) is a treatable autosomal recessive inherited metabolic disorder. It results from a deficiency of sterol 27-hydroxylase (CYP27A1), which is a mitochondrial cytochrome P450 enzyme that catalyzes the hydroxylation of cholesterol and modulates cholesterol homeostasis. Patients with CYP27A1 deficiency

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