

## **Cerebrotendinous xanthomatosis revealed in drug-resistant epilepsy diagnostic workup.**

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### **Abstract**

Cerebrotendinous xanthomatosis (CTX) is a treatable disorder of bile acid production caused by mutations in the mitochondrial enzyme sterol 27-hydroxylase. This inborn error of bile acid metabolism results in lipid pathologic accumulation in multiple tissues. Progressive neuropsychiatric disturbances are a frequent manifestation of this disease. Although seizures have been frequently noticed as part of CTX manifestations, there have not been reports of CTX being diagnosed in drug-resistant epilepsy diagnostic workup nor of seizure response to chenodeoxycholic acid treatment. Here, the authors present a case of a drug-resistant epilepsy patient with a complex phenotype where a diagnosis of CTX was done and showed a significant reduction in seizure frequency after chenodeoxycholic acid supplementation. This report illustrates the importance of considering treatable neurometabolic disorders in epileptic patients showing complex phenotypes.