38 year old male with childhood onset chronic diarrhea, progressive learning disabilities in school, bilateral cataracts noted in his twenties along with significant tendon xanthomas.

At the age of 38 years he presented with ataxia, progressive memory loss, attention deficit disorder, and sleep disturbances. Physical examination showed striking tendinous xanthomas and cataracts.

His fasting lipid profile in mg/dL: TC 220, TG 254, calculated LDL-C 130, and HDL-C 39, with a plasma cholestanol level that was significantly elevated at 8.5 mg/L (normal < 5.0 mg/L). He was found to be a compound heterozygote for defects at the CYP27A1 gene locus (R127W and splice site 845-1G>A).

On CDCA therapy at a dose of 250 mg orally three times daily, his chronic diarrhea disappeared, with improvements in his neurologic function over the past 3 years.
Cerebrotendinous Xanthomatosis
