CTX, A RARE CASE OF CHRONIC DIARRHEA IN AN ADULT

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Introduction

• Cerebrotendinous Xanthomatosis (CTX) – rare, autosomal recessive disease

• The principle enzyme deficiency is sterol 27-hydroxylase\(^1\) results in defect in bile acid synthesis

Achilles tendon xanthoma\(^1\)
Case

• A 54 year old white female presented decades history of diarrhea and ataxia
• Family history included a degenerative familial ataxia in immediate relatives
• Worsening ataxia requiring a wheelchair due to falls and spasticity
• Poor oral intake, and difficulty with activities of daily living
• On exam wheelchair bound, spasticity, and word finding difficulty
Case

- Infectious workup and endoscopy unremarkable
- MRI with prominent atrophy of cerebellar hemispheres with scattered gliosis, T2/FLAIR along corticospinal tracts
- Brother recently diagnosed with CTX disease, she underwent genetics evaluation and measurement of cholestenol
- She was treated with chenodeoxycholic acid
Discussion

• CTX occurs in less than 5 per 100,000 people\textsuperscript{1}
• Chronic diarrhea in CTX presents early in older childhood

• The underlying pathophysiology in CTX is not completely understood but likely related to excess bile alcohols in multiple organ systems

• Chenodeoxycholic acid is believed to replace the bile alcohols produced in CTX and help with diarrhea
Mechanism of enzyme deficiency in cerebrotendinous xanthomatosis

Cholesterol

\( 7a \)-hydroxy-4-cholesten-3-one

\( \text{cholesterol} \ 7a \)-hydroxylase

\( 4 \)-cholesten-3-one

\( \text{sterol-27-hydroxylase} \)

Chenodeoxycholic acid

7a-hydroxy-4-cholesten-3-one

Mechanism of enzyme deficiency in cerebrotendinous xanthomatosis

Cholcestone

\( \text{cholesterol} \ 7a \)-hydroxylase

Cholestanol

\( \text{cholesterol} \ 7a \)-hydroxylase

CTX
Learning Points

• CTX is a rare disease and diagnosis
• Requires early intervention to prevent neurologic complications
• If treatment is started late irreversible complications develop
