CTX, A RARE CASE OF CHRONIC DIARRHEA IN AN ADULT

Brian Sowka DO,
Thu Nguyen DO,
James Groskreutz MD,
Internal Medicine, Gundersen Health System, La Crosse, WI

GUNDERSEN HEALTH SYSTEM®

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

GUNDERSEN HEALTH SYSTEM®

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

T2 Flair MRI

GUNDERSEN HEALTH SYSTEM®

Discussion

- CTX occurs in less than 5 per 100,000 people[1]

GUNDERSEN HEALTH SYSTEM®

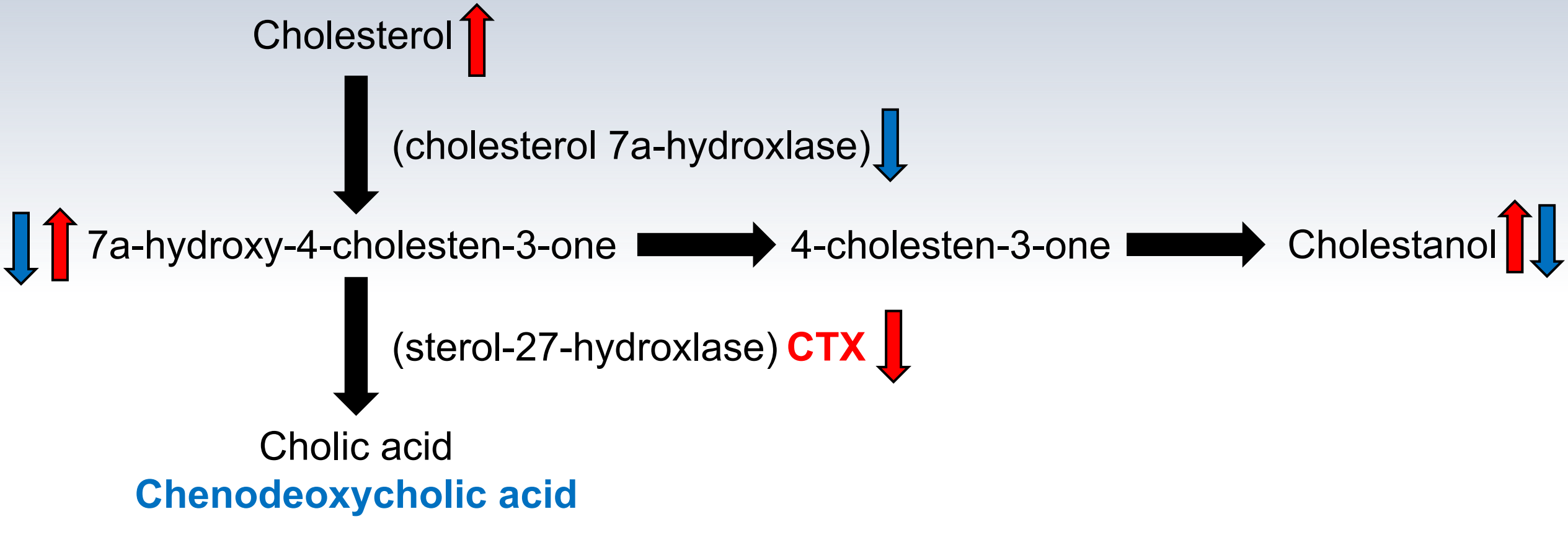
Discussion[2]

- 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

Cholesterol

Chenodeoxycholic acid

GUNDERSEN HEALTH SYSTEM®



Mechanism of enzyme deficiency in cerebrotendinous xanthomatosis[2]

Learning Points

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

References

1. Walman AT. Cerebrotendinous xanthomatosis. *UpToDate*. May 2020. Accessed March 11, 2021 from https://www.uptodate.com/contents/cerebrotendinousxanthomatosis
2. Shuke N, et al. Cerebrotendinous xanthomatosis: a comprehensive review of pathogenesis, clinical manifestations, diagnosis, and management. *Orphanet Journal of Rare Diseases*. 2014; 9: 179. doi: 10.1186/s13023-014-0179-4.
3. Leonard J. What to know about complex partial seizures. Medical News Today. 2018. https://www.medicalnewstoday.com/articles/320588

GUNDERSEN HEALTH SYSTEM®