

Cerebrotendinous Xanthomatosis (CTX): A Rare Etiology of “ Hot-Cross Bun” Sign

Aims & Materials

Aims:

- To present a rare case of Cerebrotendinous Xanthomatosis (CTX) manifesting with the Hot-Cross Bun sign.
- To highlight clinical features, radiological findings, and diagnostic challenges.
- To emphasize importance of early recognition and treatment.

Materials:

- Case study of a 26-year-old female patient.
- Clinical history: chronic diarrhea since age 3, cataracts at 22, progressive gait ataxia & cognitive decline since 24.
- Physical findings: tendon xanthomas, distal limb wasting, pes cavus, cerebellar signs.

Methods & Results

Methods:

- Bone Mineral Density: Osteoporosis
- Nerve Conduction Study: Sensorimotor demyelination with axonal changes
- MRI Brain:
 - T2 hyperintensity & blooming of dentate nuclei
 - Pontine 'Hot-Cross Bun' sign
 - Enlarged Virchow Robin spaces

Results:

- Clinical + Radiological correlation confirmed diagnosis of:
- Cerebrotendinous Xanthomatosis (CTX) with Hot-Cross Bun sign.

Discussion & Conclusion

Discussion:

- Radiological hallmarks of CTX: dentate nucleus hyperintensity, cerebellar/cerebral atrophy, white matter changes.
- Differentials: Marinesco–Sjogren syndrome (no xanthomas), Myotonic dystrophy type 1 (muscle weakness predominant).

Conclusion:

- CTX is a rare but treatable metabolic disorder.
- Hot-Cross Bun sign can occur outside MSA-C.
- Key clinical clues: diarrhea, ataxia, cognitive decline, tendon xanthomas.
- Early recognition enables timely treatment and improved outcomes.