Teaching NeuroImage: Subcutaneous Swellings and Cognitive Impairment in a Young Male Patient

Author(s):
Rasmi Ranjan Sahoo, D.M1; Saravana Sukriya, DM1; Gadde Sudhish, MD1; Adya Kinkar Panda, MD1; Debahuti Mohapatra, MD1; Pradeepta Sekhar Patro, DM1

Corresponding Author:
Pradeepta Sekhar Patro, drpradeepa07@gmail.com

Affiliation Information for All Authors: 1. Institute of Medical Sciences & SUM Hospital, Bhubaneswar-751030

Equal Author Contribution:

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Saravana Sukriya: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Analysis or interpretation of data
Gadde Sudhish: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Analysis or interpretation of data
Adya Kinkar Panda: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Analysis or interpretation of data
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A 20-year male patient presented with progressive painless subcutaneous swellings over both knees, heels and dorsum of right great toe (Figure 1A), and diminution of vision for last 8 years. Examination revealed high-arched feet and global cognitive impairment. Slit lamp examination showed bilateral lenticular opacities (Figure 1, B and C). Histopathological examination of swelling revealed foamy histiocytes, cholesterol clefts and touton giant cells, suggestive of xanthoma (Figure 1D). Axial T2-weighted magnetic resonance imaging of brain showed bilateral symmetrical cerebellar dentate nuclei hyperintensities and susceptibility-weighted imaging revealed hypointensities due to mineralisation, alongwith moderate atrophy of the cerebellar vermis (Figure 2, A and B). Whole exome sequencing revealed heterozygous pathogenic mutations on exon 3 (c.526delG) and exon 7 (c.1213C>T) in the CYP27A1 gene, suggestive of cerebrotendinous xanthomatosis (CTX). The characteristic features of CTX include progressive neurologic impairment, tendon xanthomas, cataract and premature atherosclerosis.¹ Chenodeoxycholic acid alleviates neurological symptoms and improves prognosis.
References


Figure legends

**Figure. 1. Clinical manifestations and imaging characteristics of cerebrotendinous xanthomatosis.** Tendon xanthomas (A), bilateral lenticular opacities (B & C), and histopathology of subcutaneous swelling showing cholesterol clefts (arrow) and touton giant cell (arrowhead) (D) (H&E, x400).
Figure 2. Clinical manifestations and imaging characteristics of cerebrotendinous xanthomatosis. Axial T2-weighted (A) and susceptibility-weighted (B) magnetic resonance imaging of brain revealed bilateral cerebellar dentate nuclei hyperintensities (horizontal arrows) and punctate foci of hypointensities (oblique arrows), respectively.
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