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Clinical Image
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# An Aggressive Form of Cerebrotendinous Xanthomatosis

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## Clinical Image

This 34-year-old man presented to our clinic with progressive gait difficulties and swelling of his tendons. On examination, he had bilateral ataxia, upper motor neuron signs, and tumors in his tendons (Figure 1A-1C). Brain MRI showed diffuse Encephalopathy (Figure 1D-1F). Cerebrotendinous Xanthomatosis was diagnosed after genetic testing confirmed the presence of pathogenic variants on the CYP27A1 gene. Cerebrotendinous Xanthomatosis is a rare, potentially treatable autosomal recessive disorder characterized by impairment of the hepatic conversion of cholesterol to chenodeoxycholic acid, leading to massive deposition of cholesterol in organs. It presents with xanthomas, juvenile cataracts, and progressive neurological dysfunction [1].

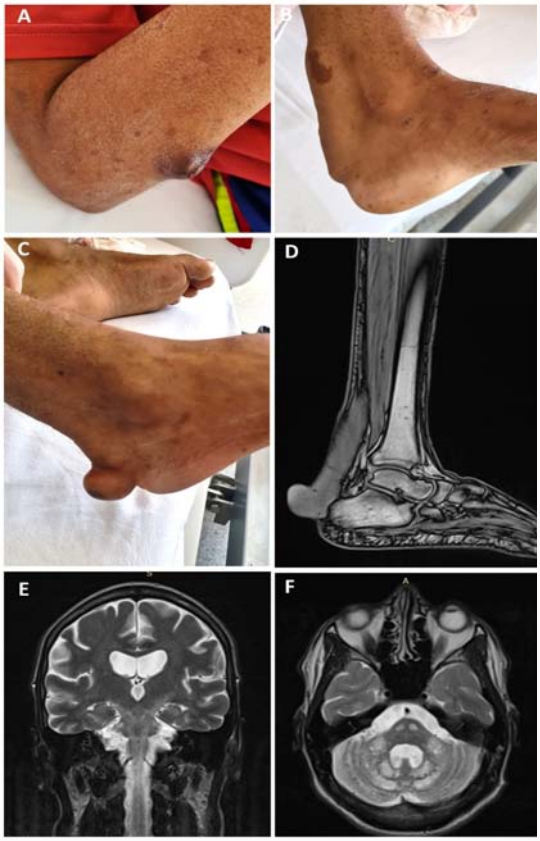


**Figure 1:** Physical examination and MRI images. The patient's tendons on physical examination (A-C), MRI T1 Vibe imaging of the right foot enlarged tendon (D), and brain MRI showing bilateral hyperintensities on the corticospinal tracts, middle cerebellar peduncles, and dentate nucleus (T2 images – E, F).

## References

1. Moghadasian MH, Salen G, Frohlich JJ, Scudamore CH. Cerebrotendinous xanthomatosis – A rare disease with diverse manifestations. Arch Neurol. 2002;59(4):527-9.

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