

## Cerebrotendinous Xanthomatosis with Cerebellar Vacuolation

### Serebellar Vakuolizasyonlu Serebrotendinöz Ksantomatozis

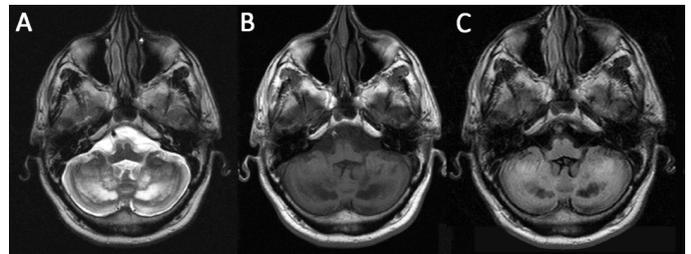
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Cerebrotendinous xanthomatosis is an autosomal recessive lipid storage disease characterized with xantoma of the tendons, as well as ocular, cardiac and neurological involvement. The disorder is caused by a mutation in the CYP27A1 gene coding the sterole-27-hydroxylase enzyme converting cholesterol to cholic acid and chenodeoxycholic acid. The mutation causes the serum cholesterol level to rise, which is diagnostic as well as clinical features of the disease. As a result of cholestanol accumulation in tissues, patients develop progressive pyramidocerebellar symptoms, cognitive failure and peripheral neuropathy. The progressive course of the disease can only be prevented if treated early with chenodeoxycholic acid.

Thirty-four year old male patient presented with complaints of progressive difficulty in walking, loss of balance and forgetfulness; his medical history showed chronic diarrhea, cataracts and osteoporosis. Bilateral swellings were noticed on his achilles tendons initially when he was 17, and he had complaints of forgetfulness and slowly progressing ataxia since 20 years of age. His parents were first degree relatives. His neurological examination showed his speech to be dysarthric and that he had horizontal nystagmus, bilateral pyramidocerebellar symptoms and mild cognitive failure. In addition, he had bilateral xanthomas on the achille tendons (Figure 1). His cranial MRI showed hyperintense lesions in the T2 weighted sections and hypointense lesions consistent with vacuoli in the T1 weighted and FLAIR sections in the cerebellar white matter (Figure 2). His laboratory examination showed serum cholestanol level to be 3.76 mg/dl (normal: 0-0.6 mg/dl). Achille tendon biopsy was consistent with xanthoma. Chenodeoxycholic acid treatment was initiated.



**Figure 1:** Xanthoma presenting as a painless swelling on the Achilles tendon.



**Figure 2:** Patient's coronal T2 (A), T1 weighted (B) and FLAIR (C) cerebral MRI sections. Vacuoli can be seen within the cerebellar parenchyma.

### References

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