Iranian Journal of Neurology

Clinical Quiz

Ir J neurol 2012; 11(2): 80-81

# An adult man with progressive dysarthria, dysphagia and disequilibrium

Received: 29 Oct 2011
Accepted: 12 Jan 2012

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**Keywords**
Cerebrotendinous xanthomatosis

## Case

The patient was a 40 year old man, who referred to our clinic because of progressive dysarthria, dysphagia and disequilibrium for the previous 15 years. According to his medical history, he had bilateral cataract surgery when he was only 2 years old and mild mental retardation since early childhood.

On neurologic examination, he had severe dysarthria (anarthria) and jaw opening dystonia with drooling. Furthermore, he had bilateral hand dystonia and mild upper limb dysmetria. Deep tendon reflexes were brisk and there was bilateral Babinski and leg spasticity. Due to his severe dystonia, spasticity and ataxia, he had become bedridden and was unable to walk.

The brain MRIs are presented below (Figure 1):

1- What abnormalities are seen on brain MRI?

2- What is the most probable diagnosis?

3- Is there any treatment for this patient (other than symptomatic treatments)?

## Answers

1- The brain MRI shows bilateral symmetrical hyperintensity in internal capsules, cerebral peduncles, optic radiations and dentate nuclei.

2- Early cataract, mental decline with pyramidal, extrapyramidal and cerebellar signs with the above MRI findings, in addition to bilateral enlargement of Achilles tendons (Figure 2), which was not mentioned in the history and exam, make cerebrotendinous xanthomatosis (CTX) the most probable diagnosis.

3- Early treatment with chenodeoxycholic acid could stop the progression of the disease.

## Discussion

Cerebrotendinous Xanthomatosis (CTX) is a rare autosomal recessive lipid storage disease. This disease is caused by mutation in the gene encoding the enzyme 27-sterol hydroxylase, which is a mitochondrial enzyme and converts cholesterol to cholic acid and chenodeoxycholic acid.[1,2]

Deficiency in this enzyme causes accumulation of cholestanol (cholesterol precursor) and its deposition in different tissues such as lens, tendons and central nervous system.[1,2]



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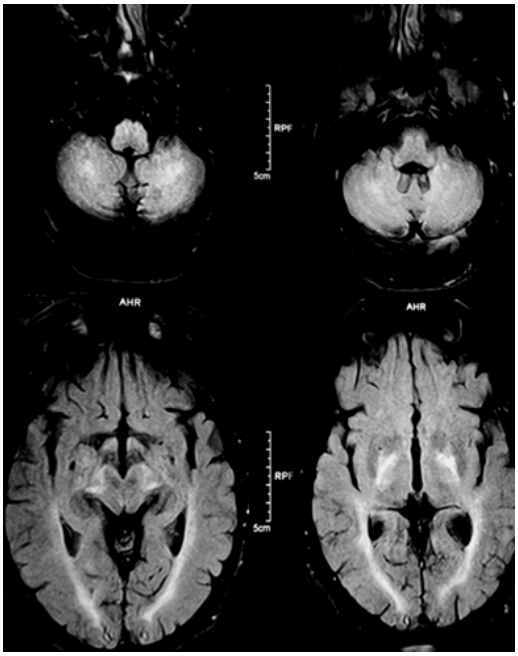


**Figure 1.** Axial brain MRI of the patient

The major symptoms of CTX comprise early cataract, tendon xanthomata and progressive neurologic deficits.[1,2]

The major neurologic manifestations including pyramidal signs, cerebellar dysfunction, mental decline, extrapyramidal signs, seizure and peripheral neuropathy start in adolescence or adulthood.[3,4]

Brain MRI abnormalities are seen in most of the cases and include cerebral and cerebellar atrophy, hyperintensities in dentate nucleus, and internal capsule on T2 and FLAIR sequences.[3,4]

Treatment with chenodeoxycholic acid (CDCA) decreases the elevated cholestanol levels and arrests the progression of the disease.[5,6]

Statins are being used in some cases in conjunction with CDCA, but their usefulness is controversial.[5,6]

Early diagnosis is essential, because early treatment will reverse the neurologic symptoms.

Finally, keep in mind to look at Achilles tendons of every patient with progressive neurologic deficit and early cataract.

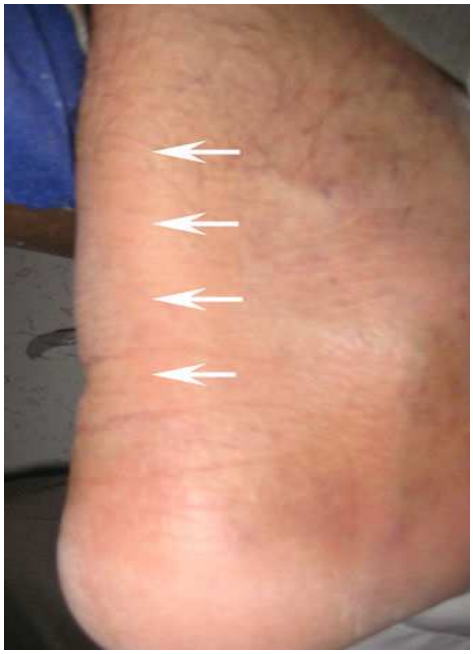

**Figure 2.** Bilateral enlargement of Achilles tendons (white arrows)

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