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CASE REPORT

CEREBROTENDINOUS XANTHOMATOSIS

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Hypercholesterolemia is a well-known risk factor for coronary heart disease, however we report a very rare disorder of lipid metabolism which is associated with coronary heart disease despite the fact that all the basic risk markers of dislipdemia like serum cholesterol, HDL, LDL and triglycerides are normal.

CASE REPORT

A 47 years old married male whose parents are first cousins, presented with a history of chronic diarrhea and ataxia since childhood. Because of poor memory and low I.Q., he left school in 7th class.

On examination he was found to have grossly ataxic gait, dysarthria, bilateral cataracts, bilateral Achilles tendon Xanthomas and blood pressure of 180/120 mmHg. He looked wasted and weighed 90 lbs. (40.5 Kg).

Clinical impression of CTX was confirmed by- raised serum Cholesterol level of 1.66 mg/dl (Reference range 0.2-0.6 mg/dl).

His lipid profile (HDL, LDL and total Triglycerides), blood glucose, urea, electrolytes, REG, and echocardiography were all normal. ECG showed ischemic changes. CT scan of brain showed moderate atrophy of cerebrum and cerebellum with small nodular enhancing foci at vermis. Abdominal ultrasound showed a 1.2 cm stone in his gall bladder.

He was put on chenodeoxycholic acid (750 mg/day) along with sodium valproate (200 mg/day) for ataxia and enalapril (20 mg/day) for hypertension. His diarrhoea has subsided and he has gained 40 Ibs in weight during the six months' therapy with chenodeoxycholic acid. Patient did not agree for coronary angiography; the ST-T changes in ECG were taken as sole evidence of coronary heart disease.

His blood pressure is controlled and the frequency of falls has come down from 2-4/wks to 0-1/month. Repeat ultrasound showed no gall stone. During the three years follow up his memory for recent events has also improved. However, the density of cataracts and the size of Achilles tendon Xanthomas have increased. The patient's own marriage is non-consanguineous and his four offspring are so far clinically normal. However, seven of his older siblings died in infancy due to some undetermined cause. It is not possible to state whether some or all of his siblings had succumbed to CTX.

DISCUSSION

Cerebrotendinous Xanthomatosis (CTX) is an auto-somal recessive disorder of lipid storage caused by deficiency of hepatic microsomal 26-alplia hydroxy-lase [16,17,18] Biosynthesis of bile acids is consequently decreased while that of cholestanol is enhanced. Cholestanol is a 5-alpha dihydro derivative of cholesterol. It is an atherogenic compound and accumulates in many tissues. Xanthomas, bile, tendons and the nervous system receive particularly heavy deposits.

Xanthomas in Achilles tendon, progressive ataxia, dementia, cataracts and widespread atherosclerosis are the most frequent presenting features of CTA.

Parkinsonism [7,15], bradykinesia, epilepsy[2,12] and polyneuropathy of demyelinating type or axonal type [14] occur less frequently. Atypical cases with only neurological signs [6] and those without tendinous xanthomas [19] have also been reported. Coronary' artery disease is also met in CTX [4,19].

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CTX is one of the very few hereditary disorders which, though potentially fatal, is amenable to treatment.
The progression of the disease can be halted and some of the existing damage can be reversed with Chenodeoxycholic acid [3,5]. Low density lipoprotein (LDL) adheres is has recently been used for treating CTX[8,13]. Ursodeoxycholic acid has proved ineffective.

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