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HOMOZYGOUS FAMILIAL HYPERCHOLESTEROLAEMIA

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A boy with multiple xanthomatosis with deranged lipid profile consistent with homozygous familial hypercholesterolaemia is reported for its rarity and exotic presentation and biochemical abnormalities of lipids in parents.

Key Words : Xanthoma, Hypercholesterolaemia

Introduction

Familial hypercholesterolaemia is inherited as autosomal dominant disorder.[1] It is characterised by an increase in the levels of LDL and is the commonest cause of tendon xathomas. Homozygotes have 2 mutant alleles and therefore are almost completely unable to clear LDL from the plasma and present in childhood with cutaneous xathomas. Heterozygotes have one normal allele and can remove about half the plasma LDL. Homozygotes of this desease have been reported at the rate of about 1 in 1 million from Western and Oriental races.[2,3] Cases have been reported from the Indian population also.[4-6]

Case Report

A 6-year-old, thin built, male child presented with yellowish nodules over interphalangeal joints of hands and feet, elbows, buttocks, knees and heels of 4 years duration. The lesions were progessively increasing in size and extent. The child was active and there was no impairment of mental intelligence. There was no signficant history of loss of weight or appetite. The parents had non-consanguineous marriage. His 3-year-old sister had also developed similar but fewer lesions on heels, elbows and natal cleft for the past 1 year.

There was no lymphadenopathy and organomegaly. CVS and CNS examinations were normal. Corneal arcus was present but on fundus examination there was no evidence of lipaemia retinalis.

Cutaneous examinition (Figs. 1 and 2) revealed yellowish, soft to firm papulonodular lesions as well as plaques at various sites i. e., xanthelasma palpebrum over both upper eyelids, tuberous

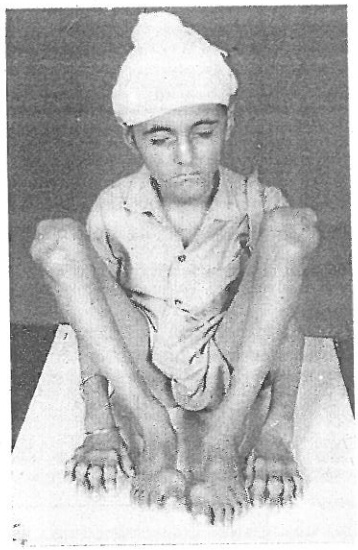

Fig. 1. Showing xanthomas over hands, feet, knees and xanthelasma palpebrum over upper eyelids

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xanthomas over the knees, elbows and buttocks, tendinous xanthomas over achilles tendon and extensor tendons of hands and feet, plane xanthomas over cubital fossa, popliteal fossa, finger creases, flexor aspects of both forearms, trunk and iliac creast. X-ray hands showed soft tissue swellings in the region of interphalangeal and metacarpophalangeal joints with no bony abnormality. Skin biopsy was consistent with xanthomatosis.

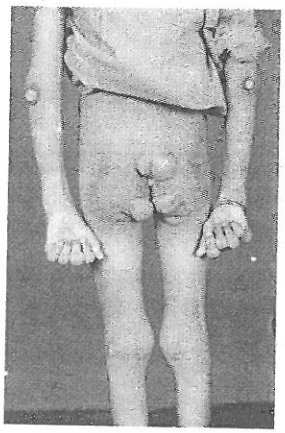

Fig. 2. Showing xanthomas over forearms, elbows, buttocks and politeal fossa

Table I. Lipid profile of the patient (mg/dl)

	Before treatment	After 3 months treatment	Normal Range
Serum total lipids	1155	823	400-900
Serum triglycerides	200	123	50-200
Serum total cholestrol	495	361	150-200
Serum HDL cholestrol	45	28	30-70
Serum LDL cholestrol	415	308	80-160
Serum VLDL cholestrol	35	25	20-40

Lipid profile of the patient is shown in table I. Lipogram of both parents also showed considerably elevated serum cholesterol. The patient was advised cholesterol restricted diet and was put on gemfibrozil 300 mg daily to be taken one hour before meals. They tolerated the drug well except for the feeling of general weakness. Skin lesions softened to some extent after 3 months of therapy.

Comments

In familial hypercholesterolaema (Type IIa of the Fredrickson classification) plasma cholesterol is elevated with normal triglyceride level. The presence of tendinous xanthomas and family members with elevated plasma cholesterol helps in diagnosis.[1] This was the case with our patient. All secondary causes of hyperlipaedemia were ruled out. For management of this condition besides diet control, a number of therapies have been advocated and institution of such therapy may increase the long term survival rates. Gemfibrozil is effective in patients with type II (both a and b) and IV hyperlipaedemia.

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