HYPERLIPIDEMIA IN A CHILD (A Case Report)

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Introduction

Hyperlipidemias are a heterogeneous group of disorders with both genetic and non-genetic etiology. The genetic type of hyperlipidemias occur in a frequency much greater than was formerly suspected. The exact basic molecular defects in these disorders have been identified. This brief report is of a case of primary hyperlipidemia in a child.

CASE REPORT

A 5-year-old boy, born of nonconsanguineous parents, presented with whitish nodules over both gluteal regions and knees, which were gradually increasing in size and number since one year. There was no history of abdominal pain, chest pain, vomiting, photosensitivity reactions or bone pains. Examination revealed a child of medium build, with weight 15 kg and height 102 cm. He had arcus juvenilis bilaterally in the cornea and multiple xanthomata over both gluteal regions and extensor aspects of the knee joints (Fig). There was no hepatosplenomegaly. There were no clinical features of hypothyroidism or nephrotic syndrome. Cardiovascular, respiratory and nervous system examination were within

Fig. : Xanthoma over gluteal region.

normal limits. Fundus examination was normal. His blood pressure was within normal limits (90/60 mm Hg right arm supine). A disorder of lipid metabolism was suspected and the patient was investigated accordingly.

Hemogram, liver function tests and urinalysis were within normal limits. ECG and chest radiogram were normal. Serum cholesterol was 840 mg/dL. Lipid profile showed a marked increase in LDL, β -lipoproteins, triglycerides and chylomicron levels and decreased HDL levels (Table). The serum cholesterol levels in the parents and one sibling, aged 8 years, were normal. (Mother - 202 mg/dL, Father - 216 mg/dL, sibling - 166 mg/dL). A diagnosis of familial hypercholesterolemia was made. The child was treated with a low cholesterol (less than 200 mg per day) low fat diet (less than 7% of total calories) and tablet nicotinic acid 5 mg TDS. Serum cholesterol levels decreased to 620 mg/dL after three months. However the xanthomata did not subside.

TABLE

Serum lipid profile of the patient

Lipid component	Levels (mg/dL)	Normal (mg/dL)
Cholesterol	840	150 - 250
Triglycerides	205	46 - 130
HDL	28	30 - 70
LDL	781	100 - 160
VLDL	41	24 - 1
Chylomicrons	32.4	< 28
Pre- β -lipoproteins	368	< 240
β -lipoproteins	792	< 550

Discussion

Hyperlipidemias can be primary or secondary to systemic diseases like hypothyroidism, nephrotic syndrome, obstructive liver disease or porphyria. In this case, there were no symptoms or clinical features suggestive

of these disorders. The primary hyperlipidemias which present with a marked increase in serum cholesterol and triglycerides are familial hypercholesterolemia, familial combined hyperlipidemia and polygenic hypercholesterolemia. Familial combined hyperlipidemia rarely occurs before 20 years and is not associated with xanthomas [1,2]. Polygenic hypercholesterolemia was considered as a differential diagnosis. However, the classical features of familial hypercholesterolemia, like corneal arcus and xanthomas, do not occur in this condition. Moreover the cholesterol levels will not be as high as observed in this case [3].

Familial hypercholesterolemia occurs as autosomal dominant and recessive forms. The autosomal recessive form is characterized by onset of symptoms beyond the second decade of life and the serum cholesterol levels range from 270-550 mg/dL. Autosomal dominant form is more devastating with serum cholesterol levels ranging from 600-1200 mg/dL. Xanthomas and arcus juvenilis occur by 4 years of age. Serum triglycerides can also be raised [3]. Parents of autosomal dominant type can have normal cholesterol levels. They can be identified as heterozygotes by genetic studies. The other possibility could be a fresh mutation [3]. This case fits in with autosomal dominant form of familial hypercholesterolemia. However, the diagnosis can be confirmed only by genetic studies. Autosomal dominant familial hypercholesterolemia has an incidence of one in a million. The primary biochemical abnormality in this is a defect in the LDL receptor which is responsible for LDL degradation through receptor mediation. The LDL receptor gene is present in the short arm of chromosome 19. Almost twenty different defects in the chromosome, causing familial hypercholesterolemia, have been described [3].

Therapy for familial hypercholesterolemia is unsatisfactory. Drugs like nicotinic acid

and cholestyramine reduce cholesterol levels by 20-30%. HMG-CO-A reductase inhibitors are very effective in achieving reduction in LDL cholesterol levels by 25-45% in adults. They have not been approved for use in children. Portocaval shunt, plasma exchange, selective LDL apheresis, alone or in combination are effective therapeutic interventions in this disorder. Extra-corporeal LDL elimination and liver transplant have been introduced as alternatives. LDL receptors are expressed in amniotic cells in early pregnancy and can be used in prenatal diagnosis [3].

In spite of these therapeutic interventions the prognosis of this disorder is poor. Coronary atherosclerosis has its onset before 10 years of age, and results in death from myocardial infarction before 30 years of age.

Hyperlipidemias have been reported earlier in India. These were cases of Type I (hyperlipoproteinemia, hyperchylomicronemia) [4], Type II (autosomal recessive hypercholesterolemia) [5] and Type IV (familial hyper b-lipoproteinemia heterozygous variant). This case is being reported for its rarity, the need for awareness of this condition and its early management to prevent complications.

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