

ly in listeria endocarditis(8). Early suspicion and prompt therapeutic intervention is important for decreasing the high mortality due to listeriosis in various groups of compromised hosts.

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Pseudo-Homozygous Type IIa Hypercholesterolemia

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Hyperlipoproteinemias are disturbances of lipid transport which result from accelerated synthesis or retarded degradation of lipoproteins that transport cholesterol and triglycerides through plasma(1). Although hyperlipoproteinemias appear in childhood, they are unfortunately diagnosed at an older age when they present with life threatening complications, such as

atherosclerosis. If they can be diagnosed during early childhood the associated complications can be prevented. We are describing a case of pseudo-homozygous hypercholesterolemia type IIa(2), a recently described rare variety of hyperlipoproteinemias, which was diagnosed early in life.

Case Report

A 10-year-old male child was admitted to our institution with a history of multiple swellings in the region of buttocks, the extensor surface of both elbows, and on the fingers since the last 5 years. Initially these swellings were of the size of a pea which gradually increased to have variable dimensions. At first they appeared on the dorsal aspect of fingers of upper limb and then on and around elbow, knee and intergluteal region. These swellings were not associated with pain or limitation of joint movements. Trauma to the intergluteal swellings during sitting sometimes caused minor bleeding. The child

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was a strict vegetarian and a product of non-consanguineous marriage with a rural background. He had no history of a similar illness or a history suggestive of premature atherosclerosis in the family.

Clinical examination of this child revealed presence of multiple nodular xanthomas on proximal interphalangeal (*Fig. 1*), elbow and knee joints which were of the size of 1 to 5 cm. In the intergluteal region multiple parallel elevations resembling a streak like pattern (*Fig. 2*) were seen which have been described as streak like xanthomas. These ranged in size from 0.5 to 3.0 cm. An ophthalmological examination revealed presence of arcus-cornae and xanthelasma. On examination of the precordium, a grade III, rough and rasping ejection systolic murmur was heard which was also associated with a thrill in the carotid arteries. No signs of congestive heart failure were present. A valvular aortic stenosis was later on confirmed by echocardiography.

Based on the clinical presentation, a provisional diagnosis of hyperlipidemia

was suspected. The serum lipid profile revealed: cholesterol-798.2 mg/dl, triglycerides-111.5 mg/dl, high density lipoproteins-43.8 mg/dl, very low density lipopro-

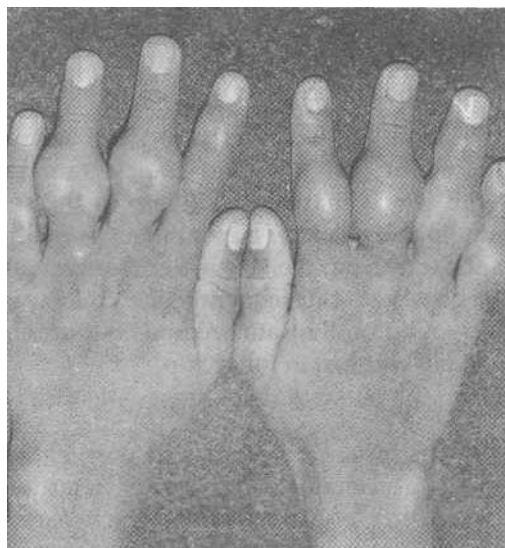


Fig. 1. Showing nodular xanthomas around the interphalangeal region of hands.

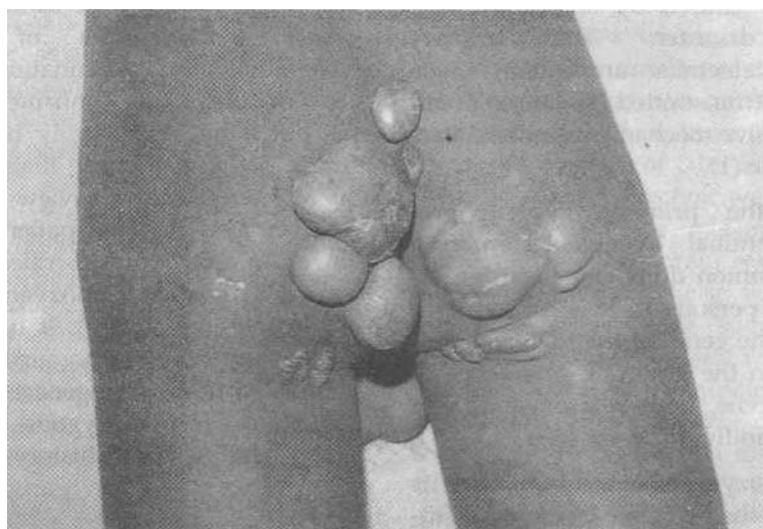


Fig. 2. Intergluteal xanthomas of various size

teins-22.3 mg/dl, and low density lipoproteins (LDL) 732.1 mg/dl. Histopathological examination of a nodule excised from the elbow showed a classical picture suggestive of xanthoma tuberosum multiplex. Routine hematological profile, X-ray of skull, spine, chest and wrist were normal. Rheumatoid factor was negative and ECG showed no abnormality.

This child was then prescribed a low cholesterol diet and oral cholestyramine. There was a marginal decrease in the serum lipid levels after a month, but a significant decrease in the size of xanthomas was not observed. Subsequently the child was discharged on the same regime, but unfortunately was lost in follow up.

Discussion

A disturbance in lipid transport, either due to accelerated synthesis or retarded degradation of lipoproteins which transport cholesterol and triglycerides through plasma, results in hyperlipoproteinemias. This may be due to a primary defect in the synthesis or degradation of lipoprotein particles or secondary to a constellation of abnormalities caused by and underlying metabolic disorder. The primary hyperlipoproteinemias are either single gene defects (transmitted by simple dominant or recessive mechanisms) or multifactorial disorders(1).

Among the primary hyperlipoproteinemias, familial hypercholesterolemia (FH) is a common dominant disorder, affecting 1:500 persons(3). It is caused by a mutation in the gene for the LDL receptor. Depending on the mode of inheritance of the mutant gene, both heterozygous and homozygous individuals are seen.

The homozygous individual inherits two copies of the FH gene and consequently has six to eight folds elevation in plasma LDL cholesterol levels. These patients often

present with planar cutaneous xanthomas at birth, but always develop the lesions by 6 years of age. The xanthomas are present at points of cutaneous trauma such as knees, elbows, and buttocks. Tendon xanthomas, arcus corneae and xanthelasma are also characteristic. Coronary artery atherosclerosis frequently has its clinical onset before 10 years of age and cholesterol deposition in the aortic valve may produce symptomatic aortic stenosis(4). Homozygous individuals usually succumb to the complications of myocardial infarction before 20 years of age.

A cholesterol level of greater than 600 mg/dl with normal serum triglycerides and characteristic clinical picture is highly suggestive of the diagnosis of homozygous familial hypercholesterolemia. Elevation in plasma LDL occurs in type 2a.

The patient reported by us had a characteristic clinical picture which prompted us to have a serum lipid profile. The elevated plasma LDL cholesterol, along with the presence of cutaneous and tendon xanthomas(5,6), valvular aortic stenosis(4), and arcus corneae with xanthelasma suggested a diagnosis of FH. The histopathological examination of an excised nodule further confirmed the diagnosis, but a negative family history was a strong point against the diagnosis. Subsequently an extensive review of the literature revealed that the patient in fact belonged to a recently described, rare entity known as pseudo-homozygous type 2 a hypercholesterolemia(2). It has been named as pseudo-homozygous because it resembles in both pathogenesis and clinical picture to FH homozygous type Ha, but lacks a positive family history.

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Interhemispheric Arachnoid Cyst with Agenesis of Corpus Callosum

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Arachnoid cysts are benign developmental cysts that occur throughout the cerebrospinal axis in relation to the arachnoid membrane and the subarachnoid space(1). Intracranial arachnoid cysts usually occur in close proximity to arachnoid cisterns, most often in the Sylvian fissure(2) and they become symptomatic in early childhood(3). We report an interhemispheric arachnoid cyst associated with agenesis of corpus callosum in a neonate.

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Case Report

A term male baby born vaginally to a grand multigravida mother with Apgar scores of 5 and 7 at 1 and 5 minutes, respectively and weighing 3500 grams had focal clonic seizures of left upper limb with associated orofacial movements on the second day of life. Clinical examination of the neonate was essentially normal with a head circumference of 37 cm and length of 54 cm.

Investigations which included arterial blood gases, serum electrolytes, blood sugar, serum calcium and cerebrospinal fluid examination and culture were normal. Ultrasonography of the skull revealed 70 x 81 x 70 mm anechoic well defined cystic structure in the midline. Computed tomography of the brain revealed a large midline interhemispheric cyst displacing the brain parenchyma suggestive of an arachnoid cyst. The lateral ventricles, third ventricle and fourth ventricle were well visualized. Agenesis of corpus callosum was also seen. No other brain malformation or atrophy was noted (Figs. 1 & 2). Neonatal seizures were controlled with phenobarbitone and phenytoin therapy. The neonate was discharged on day fifteen