International Journal of Medical and Pharmaceutical Research

Online ISSN-2958-3683 | Print ISSN-2958-3675
Frequency: Bi-Monthly
Available online on: https://ijmpr.in/

Original Article

A Case of Familial Hypercholesterolemia, Type IIA (With Tuberous Xanthomas and Arcus Senilis)

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Received: 07-01-2026
Accepted: 25-01-2026
Published: 07-02-2026

ABSTRACT

Familial hypercholesterolemia is an autosomal dominant disorder due to mutations in the low-density lipoprotein receptor gene, characterized by skin and tendon xanthomas, xanthelasma and premature arcus senilis. A female patient presented with multiple large swellings over extensor aspects of both the elbows, inner aspects of both the arms and buttocks which on biopsy were found to be tuberous xanthomas. Her lipid profile revealed severe hypercholesterolemia and markedly raised LDL levels. A diagnosis of Familial Hypercholesterolemia was made. She was advised life style modification; surgical excision was done and treated with Rosuvastatin 40mg OD. As Homozygous FH is an important risk factor for atherosclerosis and premature coronary artery disease in children and young adults, early diagnosis and treatment with screening of first-degree relatives is essential to minimize the progression of cardiovascular disease.

Keywords: Familial Hypercholesterolemia, Tuberous Xanthomas, Elevated LDL Cholesterol, Premature Coronary Artery Disease, Rosuvastatin Therapy.

INTRODUCTION:

Tuberous xanthomas present as plaques or papules or nodules over the skin present in lipoprotein metabolism disorders. These are benign and formed by accumulation of lipid laden macrophages that develop in the dermis subcutaneous tissue.[1] Tuberous xanthomas are firm painless yellowish nodules most commonly seen over extensor aspects of limbs and buttocks. [2,4] They are indicative of a derangement in lipoprotein metabolism, in particular familial hypercholesterolemia.

CASE REPORT:

A 17yr old Female patient came with the complaints of asymptomatic raised lesions over elbows, arms, buttocks since 10 yrs which started as small asymptomatic peanut sized lesions over both the buttocks since 10 yrs followed by the appearance of similar lesions over both the elbows since 3 yrs which later gradually progressed to tumour like growths. Patient noticed similar lesions over both the arms 2yrs back. She is a product of consanguineous marriage. Father died of MI at the age of 38 yrs. Her mother also had hypercholesterolemia. On examination she had bilateral corneal arcus.

Figure:1. ARCUS SENILIS

Her Cutaneous examination showed 3 to 7 cm sized firm oval nodules of skin colour over both the buttocks. Skin coloured firm plaques of size varying from1 cm to over 8 cm were present over both the elbows. Multiple yellow coloured papules were noticed over both the arms. Palms and soles are normal ; Hair, scalp and nails are normal.

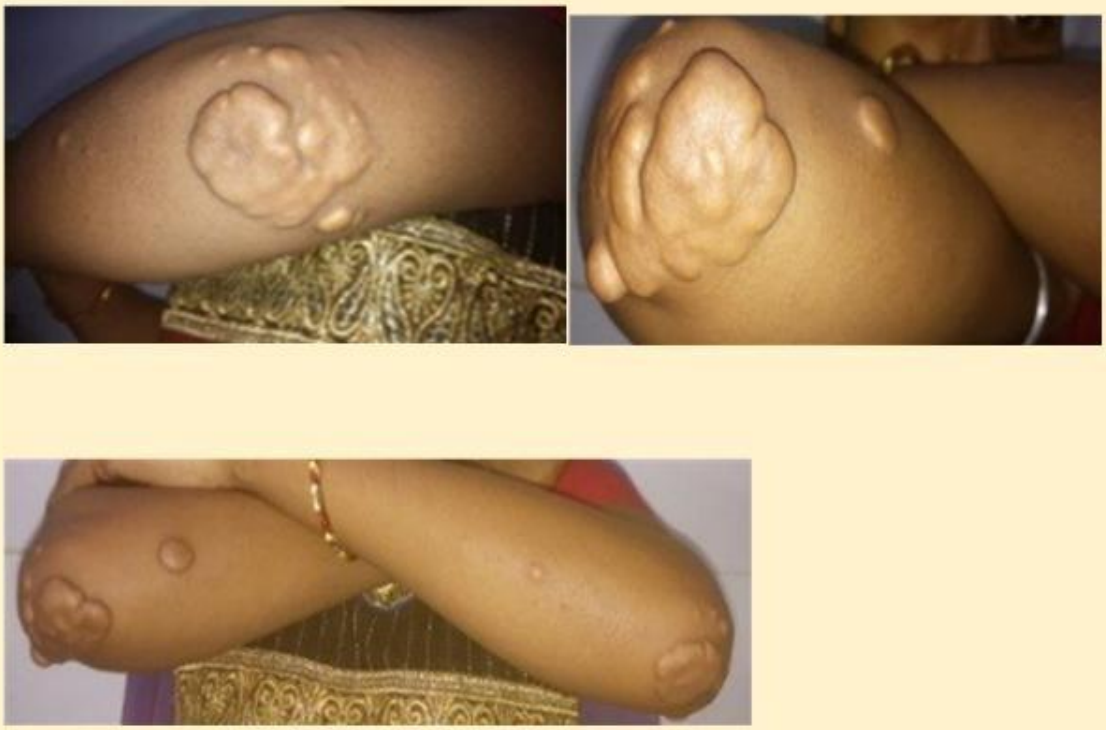

FIGURE: 2,3,4: TUBEROUS XANTHOMAS OVER ELBOWS

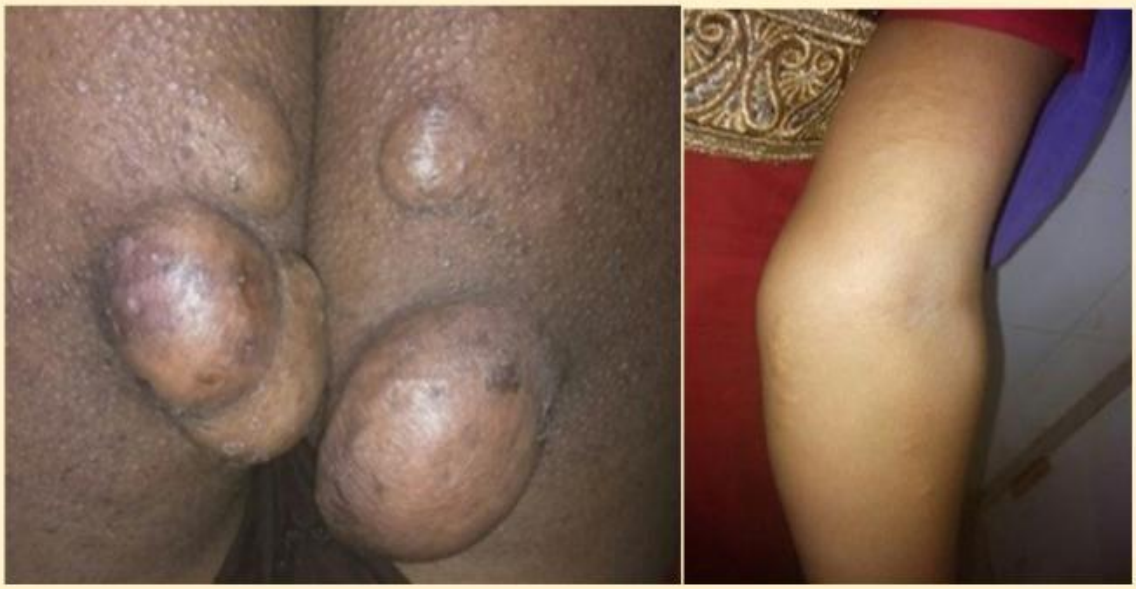

FIGURE: 5,6: TUBEROUS XANTHOMAS OVER BUTTOCKS, ARMS AND FOREARMS

Her Lipid profile showed total cholesterol as 386 mg/dl, low density lipoprotein as 335 mg/dl. Her triglycerides and high density lipoproteins are normal.

Lipid profile

Lipid fraction	value (mg/dl)	normal range(mg/dl)
Total cholesterol	386	<240
LDL	335	<150
VLDL	16	<40
TRIGLYCERIDES	80	<160
HDL	39	<40

LDL: LOW DENSITY LIPOPROTEIN, VLDL: VERY LOW DENSITY LIPOPROTEIN,

HDL: HIGH DENSITY LIPOPROTEIN

TABLE:1: LIPID PROFILE OF PATIENT CONFIRING TYPE LLA HYPERCHOLESTEROLEMIA

Biopsy from the nodule revealed infiltration of the dermis with admixture of foam cell histiocytes and lymphocytes.

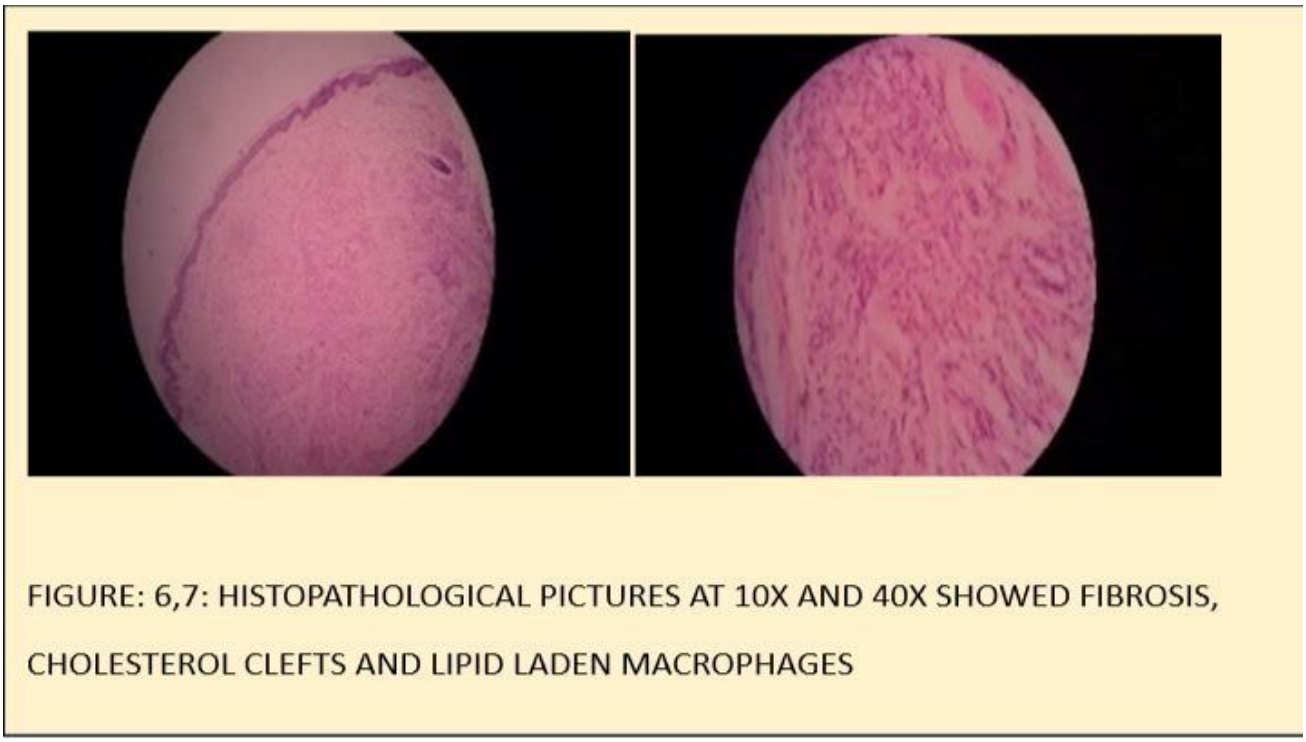

FIGURE: 6,7: HISTOPATHOLOGICAL PICTURES AT 10X AND 40X SHOWED FIBROSIS, CHOLESTEROL CLEFTS AND LIPID LADEN MACROPHAGES

Routine investigation were normal. 2D Echo and Thyroid profile are normal. Surgical excision was done and Rosuvastatin 40 mg OD was started. Life style modification was advised.

DISCUSSION:

Familial hypercholesterolemia is an autosomal codominant disorder with raised LDL levels due to increased production and reduced resorption of LDL secondary to dysfunctional LDL receptors due to mutations in LDL receptor gene. Heterozygotes express half the number of LDL receptors and homozygotes have between 0% and 25%.[5]. As Homozygous familial hypercholesterolemia is an important risk factor for atherosclerosis and premature coronary artery disease in children and young adults[5,8], early diagnosis and treatment of first degree relatives is essential to minimize the progression of cardiovascular diseases. Screening of family members in our case revealed hypercholesterolemia in her mother and elder sister.

Xanthomata are cutaneous markers of underlying lipid metabolism disorders, classified by Frederickson into five classes. Tuberous xanthomas are usually found in type IIa familial hypercholesterolemia.[3,6]. Histologically, apart from foamy macrophages, they have also been found to contain primitive mesenchymal cells, elongated perivascular, and fibroblast like cells, and lysosome-filled macrophages, indicating possible stages in the evolution of dermal mesenchymal cells into mature, cholesterol-rich foam cells.[7]

Pharmacologic treatments includes statins, bile acid sequestrants, ezetimibe, niacin that affect various steps in the pathways of cholesterol absorption and metabolism. The most common medication is a statin in combination with a cholesterol absorption inhibitor, and if needed a third drug such as a bile acid sequestrant.[9] The treatment options must include lifestyle and diet modifications also. Pharmacologic treatments and invasive procedures including lifelong lipid apheresis and liver transplantation.[10].

CONCLUSION:

In conclusion, this case was reported for the atypical presentation of multiple giant tuberous xanthomas which are generally <2cm in size and presence of arcus senilis which is considered rare. A thorough workup is essential for all the family members to identify the underlying condition to start treatment early and to prevent further complications.

Declaration:
Conflicts of interests: The authors declare no conflicts of interest.
Author contribution: All authors have contributed in the manuscript.
Author funding: Nill

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