Tuberous and Tendon Xanthomas in a child: A sentinel presentation of familial hypercholesterolemia (Type II Hyperlipoproteinemia)

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Abstract

Background: Xanthomas are localized lipid-rich deposits in the skin and tendons, often signaling underlying disorders of lipid metabolism. Pediatric xanthomas are uncommon and should prompt evaluation for familial dyslipidemias.

Case Presentation: We report a 12-year-old boy with multiple, gradually progressive nodular swellings over extensor surfaces, sacral region, and Achilles tendons. Laboratory investigations revealed markedly elevated serum cholesterol and LDL levels with normal triglycerides, consistent with familial hypercholesterolemia (Type II hyperlipoproteinemia). Histopathology confirmed lipid-laden macrophages and multinucleated giant cells. The patient responded well to rosuvastatin and ezetimibe, with regression of lesions and improvement in lipid profile.

Conclusion: Early dermatologic recognition of xanthomas can lead to timely diagnosis of inherited dyslipidemias and prevent serious cardiovascular complications.

Key words: Dermoscopy, familial hyperlipoproteinemia Type II, nodules, statins, xanthomas.

Introduction:

Xanthomas are localized deposits of lipids within the skin and tendons, frequently associated with disorders of lipid metabolism[1-3]. They may contain triglycerides or cholesterol esters depending on the underlying metabolic abnormality. Their pathogenesis resembles atheroma formation, particularly regarding the role of modified LDL and lipid accumulation in macrophages[4].

Clinically, xanthomas may present as yellowish papules, nodules, or plaques. Based on morphology and location, they are classified into eruptive, tuberous, tendinous, plane, or subdermal forms. Tendon xanthomas particularly involves the Achilles tendon which are highly suggestive of familial hypercholesterolemia (FH).

Eruptive xanthomas are typically multiple, small (1-4 mm), erythematous-yellow papules occurring on extensor surfaces. Tuberous xanthomas, in contrast, present as firm, sometimes lobulated nodules over joints or extensor surfaces[5].

Hyperlipoproteinemias may be primary (genetic) or secondary. In the Fredrickson classification:Type I, IV, V will have Hypertriglyceridemia, Type II will have Hypercholesterolemia (elevated LDL) while Type III will have Elevated cholesterol and triglycerides. Herein, we present an early-diagnosed pediatric case of familial hypercholesterolemia with classical tendon and tuberous xanthomas.

Case presentation:

A 12-year-old boy presented with multiple nodular swellings of varying sizes over a period of 10 years. The child presented with multiple nodular swellings that began insidiously at around 2 years of age, initially over the bilateral Achilles tendons. Over the subsequent 10 years, the swellings gradually increased in number and size, involving the extensor surfaces of the elbows, knees, feet, and sacral region. The lesions were non-tender, non-itchy, and progressively enlarging without any history of inflammation, ulceration, or discharge (Fig. 1). Nodules ranged from 4 × 3.5 cm (older lesions) to 3 × 3 mm (recent lesions). They were asymptomatic and varied from soft to firm.

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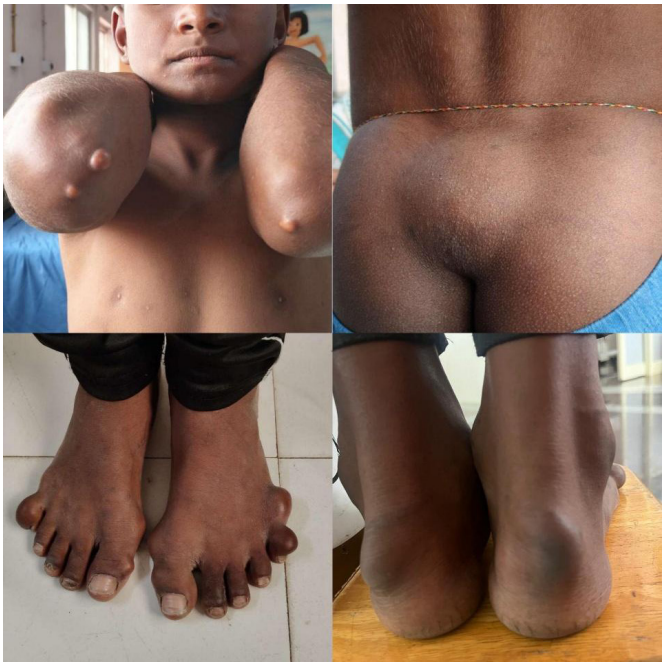

Figure 1: Showing lesions over Elbow joint, Sacrum, foot and Achilles tendon.

The child was born to second-degree consanguineous parents, and there was a significant family history was noted wherein the child's paternal uncle developed premature coronary artery disease and underwent coronary angioplasty at the age of 36 years, further supporting the hereditary nature of the condition (Figure 2).

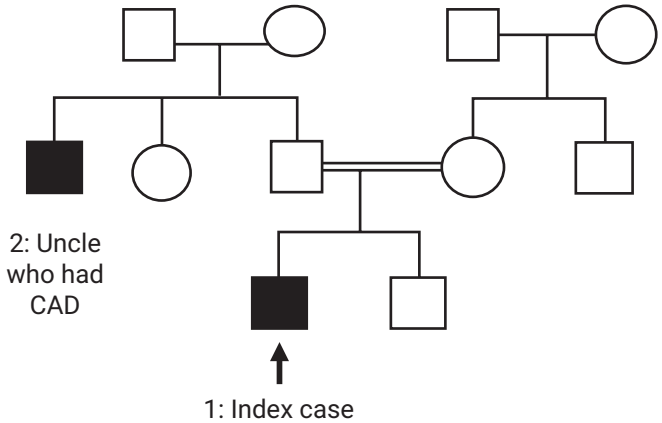

Figure 2: Pedigree chart

Anthropometric assessment revealed a weight of 34.8 kg and height of 144 cm. The calculated BMI was 16.8kg/m², corresponding to the 10TH to 25THpercentile for age and sex (as per WHO growth charts), indicating normal nutritional status.

At presentation, vital parameters were stable with heart rate 92 beats per minute, respiratory rate 20 cycles per minute, blood pressure 98/64 mmHg (within normal limits for age), and oxygen saturation of 98% on room air.

Detailed ophthalmological evaluation revealed no evidence of corneal arcus, xanthelasma, or retinal abnormalities.

Laboratory evaluation revealed: Raised total cholesterol (598.3 mg/dl) and LDL (426.9 mg/dl) levels with normal HDL, VLDL and Triglycerides (115.8 mg/dl). These findings indicated severe hypercholesterolemia with normal triglycerides with family history of coronary artery disease suggestive of familial hyperlipoproteinemia Type II.

Lipid profile screening of both parents revealed elevated total cholesterol and LDL cholesterol levels in the father (Total cholesterol: 295 mg/dL, LDL: 225 mg/dL), while the mother's lipid profile was within normal limits, supporting an autosomal dominant inheritance pattern.

Ultrasonography of the neck showed no evidence of carotid intimal thickening or focal plaques. Color Doppler evaluation demonstrated normal carotid artery flow patterns with no evidence of luminal narrowing or atherosclerotic changes.

Ultrasonography of lesions showed encapsulated hyperechoic solid lesions consistent with benign fat-containing masses with normal abdominal ultrasound and ECG. Dermoscopy of lesions showed homogeneous yellow (lipid material) background (Fig. 3).

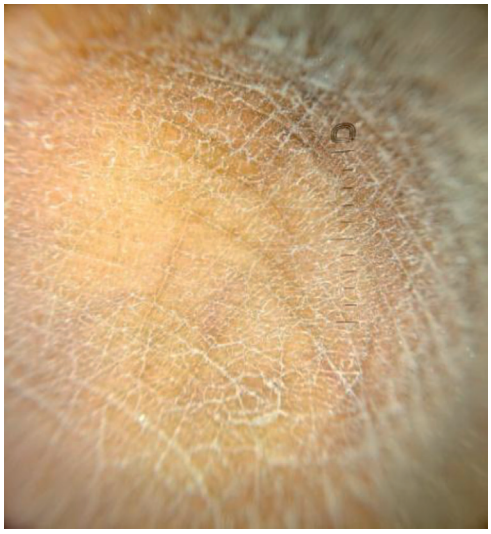

Figure 3: Dermoscopy of lesion

Biopsies from the nodules over the knee (5 × 4 cm, firm) and lateral foot (3 × 2 cm, soft) were taken and histopathology of these lesions showed - numerous lipid-laden macrophages, spindle-shaped nuclei with vacuolated cytoplasm, fibroblasts and multinucleated giant cells with background fat globules and fibrous strands (Fig. 4). These findings were consistent with xanthoma.

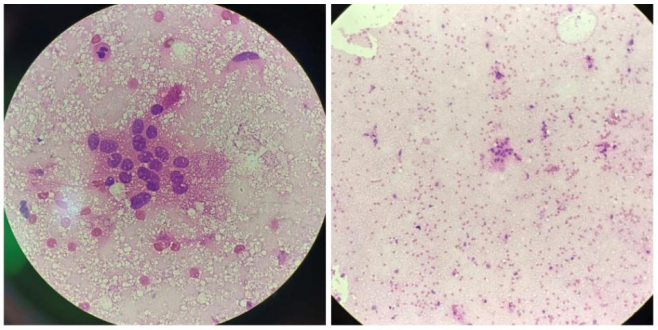

Figure 4: Histopathological findings of nodules.

The patient was started with Statins (Rosuvastatin), Ezetimibe (a selective cholesterol absorption inhibitor) and lifestyle modifications. Follow-up showed no appearance of new lesions, gradual regression in the size of existing nodules and improvement in serum lipid levels

Genetic counselling was provided to the family regarding the inherited nature of the disorder, cardiovascular risks, and the need for cascade screening. Although genetic testing for LDL receptor or related gene mutations could not be performed due to resource limitations, the clinical phenotype and biochemical profile were strongly suggestive of familial hypercholesterolemia.

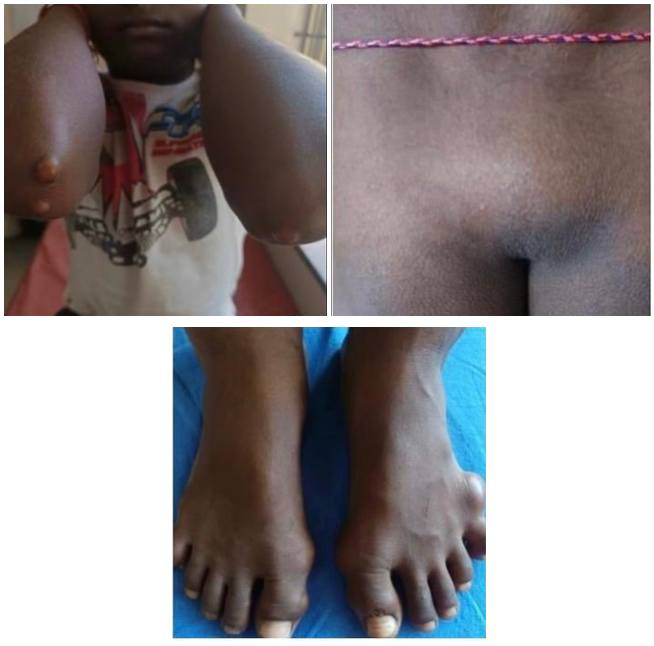

Figure 5: Post treatment picture : Mild regression of lesions

Discussion:

Xanthomas in children are uncommon and commonly point toward primary disorders of lipid metabolism such as familial hypercholesterolemia. Secondary causes include diabetes, hypothyroidism, nephrotic syndrome, medications (retinoids, oestrogens), or rarely sarcoidosis[6].

Lipid metabolism involves proteins of lipoproteins, which are co-factors of enzymes and also such enzymes as lipoprotein lipase, hepatic lipase and lecithin-cholesterol acetyltransferase. The primary genetic defects concerning the apolipoproteins and function disorders of the enzymes may lead to lipid deposition in tissues and xanthoma formation. Injury-induced cutaneous lesions mainly occur in the gluteal region, on the limbs on the extensors side and at joints flexions[7,8].

The etiological possibilities for Type II hyperlipoproteinemia include: Mutations in the LDL receptor (most common), Mutations in Apo B100, Gain-of-function PCSK9 mutations, LDL receptor adaptor protein defects or because of, ABCG5/ABCG8 mutations (Sitosterolaemia).

Clinically, Type II hyperlipoproteinemia may present with tendon/tuberous xanthomas, corneal arcus and Premature CAD. Our patient, being young, did not show corneal arcus or cardiac involvement.

The differential diagnosis were Xanthoma disseminatum, Neurofibromatosis, Lipomas, Langerhans cell histiocytosis, Storage disorders and foreign body reactions. The dermoscopy and histopathology aid in confirming lipid-rich lesions.

Early lipid-lowering therapy is crucial to prevent cardiovascular complications and also helps in decreasing the size cutaneous swellings. The standard therapy includes :High-intensity statins, Ezetimibe and Lifestyle modification. Advanced therapies for resistant cases includes Lomitapide, Mipomersen, PCSK9 monoclonal antibodies, CETP inhibitors and Laser and Cryo-surgery[8,9]. Our patient responded well to conventional therapy with regression of xanthomas, highlighting the importance of early detection.

The diagnosis of familial hypercholesterolemia was based on clinical features and biochemical findings consistent with established diagnostic criteria : Simon broome criteria, including markedly elevated LDL cholesterol (>190 mg/dL), presence of tendon and tuberous xanthomas, and a positive family history of premature coronary artery disease, fulfilling clinical criteria for probable familial hypercholesterolemia.

Conclusion:

This case emphasizes that pediatric xanthomas, especially tendon and tuberous types are important clinical indicators of underlying familial hypercholesterolemia. Early dermatologic identification and timely lipid-lowering therapy can prevent life-threatening cardiovascular events.

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References:

1. *Braun-Falco O, Plewig G, Wolff HH, Burgdorf WHC. Dermatology. Lublin: Polish Medical Publishers; 2004. p. 1159-1170.*
2. *Bito T, Kawakami C, Shimajiri S, Tokura Y. Generalized eruptive xanthoma with prominent deposition of naked chylomicrons: evidence for chylomicrons as the origin of urate-like crystals. J Cutan Pathol. 2010;37:1161-1163.*
3. *Wójcik A, Budzisz E, Rotsztejn H. Skin surface lipids and their measurements. Postepy Dermatol Alergol. 2011;28:498-505.*
4. *Dwivedi S, Jhamb R. Cutaneous markers of coronary artery disease. World J Cardiol. 2010;2:262-269.*
5. *Holsinger JM, Campbell SM, Witman P. Multiple erythematous-yellow, dome-shaped papules. Am Fam Physician. 2010;82:517.*
6. *Maejima H, Katsuoka K. Sarcoidosis coexisting with eruptive xanthoma. Eur J Dermatol. 2011;21:298-299.*
7. *Henning JS, Fazio MG. Yellowish papules on a middle-aged man: eruptive xanthoma. Am Fam Physician. 2011;83:73-74.*
8. *Pietroleonardo L, Ruzicka T. Skin manifestations in familial heterozygous hypercholesterolemia. Acta Dermatovenerol Alp Panonica Adriat. 2009;18:183-187.*
9. *Karsai S, Czarnecka A, Raulin C. Treatment of xanthelasma palpebrarum using a pulsed dye laser: a prospective clinical trial in 38 cases. Dermatol Surg. 2010;36:610-617.*

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