



Case Report

Xanthoma- A case report

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ABSTRACT

Xanthomas are lesions that are mostly located within the skin or subcutaneous tissue and consist of cholesterol and cholesterol esters. Xanthomas can occur at any site. They are mostly associated with Familial hypercholesterolemia. Although the clinical picture is variable, a high level of LDL cholesterol is the common manifestation. Here we report a case report on multiple Xanthomas in a 19 years old male.

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1. Introduction

Xanthoma is a localized collection of tissue histiocytes containing lipid in the connective tissue of the skin, subcutaneous tissues, tendons, fascia and infrequently in periosteum.¹ It is not a true tumour but a reactive histiocytic proliferation that occurs in response to alterations in serum lipids. We are reporting the case of a 19 years old male who had presented with swelling in bilateral gluteal region and thighs for one and a half years.

2. Case Report

A 19 years old boy presented with multiple painful swellings in the bilateral gluteal regions and thighs since one and a half years. On general examination, patient was conscious. On local examination, the swellings were firm to hard measuring 5.2 x 4.2 cm in size in the right gluteal region, 10 x 8 cm in the left gluteal region and 6.0 x 5.0 cm in the left thigh. An excision biopsy was done and sent to the department of Pathology at St. Stephen's Hospital, Delhi.

Grossly, two skin covered polypoidal soft tissue pieces were received which measured 8.0 x 5.0 x 4.5 cm and 11.0

x 8.5 x 7.5 cm. Cut surface was greyish yellow with brown spots. (Figure 1)



Fig. 1: Polypoidal soft tissue pieces

On microscopic examination, sections showed lesions covered by atrophic epidermis. The dermis and subcutis showed diffuse infiltration by foamy macrophages along with minimal chronic inflammation, extensive fibrosis and hyalinization. Focally, cholesterol clefts with multinucleate

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giant cells were also seen. (Figure 2)

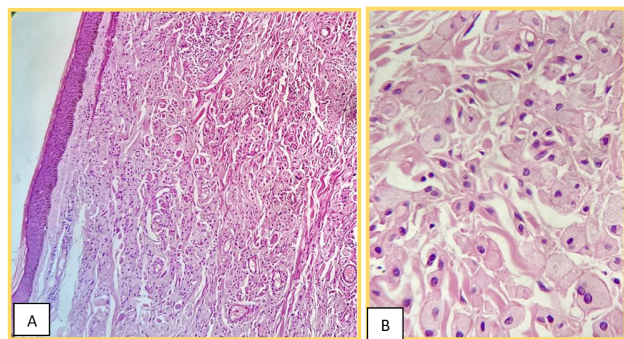


Fig. 2: A,B: Dermis and subcutis showing diffuse infiltration by foamy histiocytes (H&E, 10X 10 and 10X 40)

3. Results and Observation

The superficial location, gross appearance and associated clinical findings led to the final diagnosis of Xanthoma. The clinician was asked to follow up the patient, who was lost to follow up. Xanthomas can occur at any soft tissue site. The clinical picture varies from soft to semisolid skin macules and papules to large nodules, usually yellow coloured due to the presence of carotene contained in lipids.

4. Discussion

Xanthomas are masses that are mainly located within the skin or subcutaneous tissue and consist of cholesterol, cholesterol esters, triglycerides and numerous lipid-laden foamy macrophages.² The most frequent site for xanthomas is the Achilles tendon.^{3,4} Other sites include the extensor tendons of the hands and feet, as they are subject to mechanical stress. The mechanical stress encountered in tendons is similar to recurrent trauma, which is thought to predispose to the development of xanthomas.² Minor trauma/injury like mechanical stress has also been seen a deciding factor since it causes a release of histamine which increases the capillary permeability and therefore accelerates xanthoma formation by accumulation of LDLC.⁵ Xanthomas have been known to commonly occur in patients with Familial hypercholesterolemia.⁶

A high level of LDLC is the most common clinical manifestation and the main cause of xanthoma in patients with FH. In these patients, markedly elevated LDLC levels are secondary to LDL receptor defects and result in lipid leakage from the vasculature into the surrounding tissue. This results in uptake of lipids by macrophages which leads to accumulation of undegraded lipids and formation of foam cells.⁷

Histologically, xanthomas consist of foamy macrophages with fibrosis and cholesterol clefts. Extracellular cholesterol crystallizes into clefts, inducing an inflammatory reaction

with giant cells and fibrosis.²

CD68 is particularly useful as a marker for various giant cells, Kupffer cells and osteoclasts. In addition to these, the foam cells found in xanthomas are strongly immunopositive for CD68.

Various imaging modalities have been found to be useful in examination of xanthomas. Sonography is simple, economical and widely available which is superior to gross clinical examination. A MRI is a way to distinguish xanthomas from tumours as the former exhibits morphological and signal intensity abnormalities.⁵

Studies have shown statins have the ability to soften xanthomas. Massive xanthomas may require surgical intervention. Surgery is only suggested for xanthomas that are extremely large and painful and cause mobility problems. Certain studies, however, have reported postoperative recurrence rate of xanthomas. Postoperative cholesterol-lowering therapy may reduce the likelihood of recurrence; therefore, for multiple, massive tendinous and tuberous xanthomas, local surgical excision combined with postoperative cholesterol-lowering therapy appears to be the most effective treatment option.⁸

5. Conclusion

Xanthomas are benign masses that occur in the skin or subcutaneous tissue anywhere on the body. They have a varied clinical presentation ranging from being asymptomatic to symptoms arising from compression of adjacent structures. They are commonly seen in patients of Familial hypercholesterolemia. They have no malignant potential and need medical treatment with drugs such as statins unless they are extremely large in size causing discomfort to the patient, in which case surgical excision is the treatment.

6. Source of Funding

Nil.

7. Conflicts of Interest

All authors declare no conflicts of interest regarding the publication of this paper.


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