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# TUBEROUS XANTHOMA – A RARE CASE REPORT

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**ABSTRACT**
Xanthomas are focal infiltrates of lipid-containing histiocytic foam cells that are usually found within the dermis or tendons. It is a clinical manifestation of lipoprotein metabolic disorders. We report a case of tuberous xanthoma in an 11 year old girl who presented with multiple firm papules and nodules involving multiple sites of the body with elevated serum cholesterol, triglycerides, LDL and VLDL.

## INTRODUCTION

Xanthoma is a symptom rather than a disease, which arises due to a primary error in lipid metabolism or may even arise without any underlying disorder, clinically manifesting as soft to firm papules, plaques or nodules at various sites. There are several types of Xanthomas- Eruptive, Tuberous, Tendinous, Planar (Palmar, Intertriginous, Diffuse, Xanthelasma) and disseminated. Xanthomas can arise either due to extravasated lipids that are taken up by macrophages, resulting in foam cells or foam cells developing as a result of in situ lipid synthesis by macrophages [1].

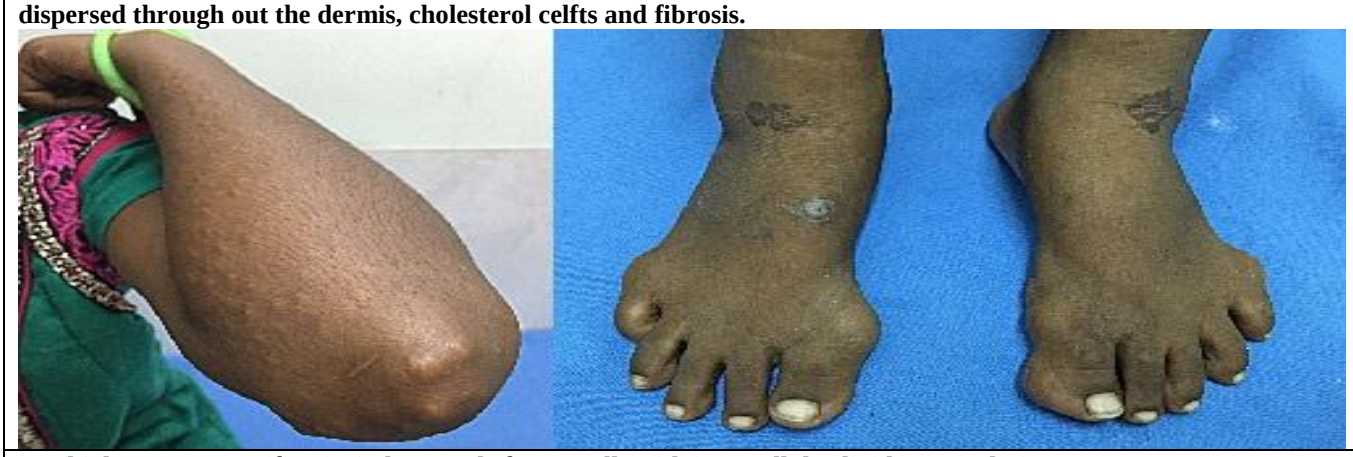
## CASE REPORT

An 11 year old girl was brought to our skin OPD with multiple yellowish to skin colored raised lesions, of varying size. She was apparently asymptomatic until 5 years of age, following which she developed multiple yellowish to skin colored raised lesions, initially over her both knees which increased in number and size in the span of 4 months and later, progressed to involve the gluteal region, elbows, forearms, and feet. Those lesions were painless. She gives history of spontaneous reduction and recurrence of the lesion's size. Born out of non-consanguineous marriage, and normal delivery, her pre-natal, natal and post natal periods were uneventful. She was incidentally diagnosed to have hypothyroidism, for which she has been on Thyroxin tablets regularly since last two years. No history of similar complaints among her siblings or any family members. Developmental history revealed milestones attained at appropriate ages. On examination, the child was moderately nourished, alert and cooperative.

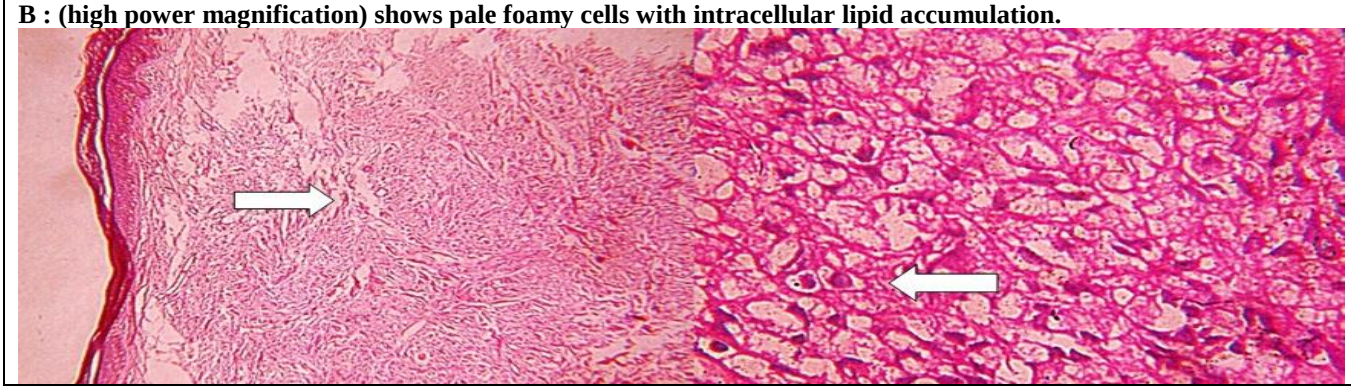
Dermatological examination revealed multiple yellowish to skin colored, non tender papules over bilateral forearms, knees and gluteal region and firm, non tender nodules over bilateral elbows, gluteal region and feet, of varying size with largest nodule over the inferio-medial aspect of right gluteus measuring 3.5 x 5.0 cm in diameter, pedunculated, firm and non-tender. Nails, oral mucosa and hair were normal. Blood investigations revealed elevated levels of total cholesterol, triglycerides, LDL and VLDL with normal routine baseline investigations. Skin biopsy taken from a nodule in the right elbow revealed, atrophic epidermis with numerous foam cells (xanthoma cells) in dermis, confirming the clinical diagnosis of tuberous xanthoma associated with dyslipidemia. The patient was advised to start on oral statins and also surgical removal of the largest lesion.

**Figure 1 & 2. shows clinical picture of skin colored non-tender papules and firm nodules of varying size, over gluteal region, dorsal aspect of hands, elbow and feet**.

**Figure 3. Tuberous xanthoma. A: (low power magnification) shows epidermal atrophy with numerous foam cells dispersed through out the dermis, cholesterol celfts and fibrosis.**



**B : (high power magnification) shows pale foamy cells with intracellular lipid accumulation.**



## DISCUSSION

Xanthomas are common presentation of a focal or generalized disorder of lipid metabolism. They are often linked with high risk of arteriosclerotic vascular diseases, pancreatitis, etc. During childhood, Type I and Type IIa are seen and are hereditary [2]. They are characterized by accumulation of lipid laden macrophages.

Pathogenesis involves a complex process by means of dysregulation of macrophage sterol flux. Xanthomas are predisposed by increased levels of cholesterol rich LDL and VLDL remnants. Under normal circumstances, around 80% of LDL clolesterol are taken up by LDL receptor mediated endocytosis. The residual LDL is cleared by scavenger receptor pathways of macrophages. In familial hypercholesterolemia, the accumulation LDL and VLDL remnants are primarily scavenged by macrophages without feedback regulation, resulting in continuous cellular lipid accumulation and foamy cells.

Clinically tuberous xanthomas are characterized by yellow to skin colored nodules that are firm, painless, indurated with an erythematous halo and are generally localized to extensor surfaces of buttocks, knees, elbows and knuckles. The lesions can be seen in inguinal and axillary folds and in the sites of trauma and they may be

fissured, pedunculated and suppurative. Frequently associated with primary hypercholesterolemia which includes familial hypercholesterolemia (Friederickson type II) and dyslipoproteinemia (Friederickson type III, an autosomal dominant genetic disorder of lipid metabolism) and in secondary hypercholesterolemia with biliary disease, monoclonal gammopathy and hypothyroidism.

In familial hypercholesterolemia, raised LDL levels are due to increased production and decreased resorption of LDL secondary to dysfunctional LDL receptors. As a result of altered endothelial function, elevated serum total cholesterol and LDL with normal triglycerides are found and manifests as atherosclerosis and coronary artery disease [3].

Histopathological examination reveals large and small aggregates of foamy cells, which are macrophages engulfing lipid droplets. Xanthoma cells are mononuclear and may also show Touton giant cells with a wreath of nuclei. Early lesions show a mixture of non foamy cells, neutrophils, lymphocytes and macrophages and well developed lesions show infiltrates consisting mainly of foamy cells which are later replaced by collagen bundles. Cholesterol clefts are also seen [4].

Treatment options include lifestyle modification with dietary changes and pharmacotherapy including statins, eczema, niacin and bile acid sequestrants like cholestyramine. This condition responds well to a combined therapy involving statins, cholesterol absorption inhibitors and a bile acid sequestrant, if needed [5]. Invasive procedures namely lifelong lipid apheresis and liver transplantation can also be considered [6]. Tuberous xanthoma shows slower rate of regression after appropriate therapy.

## CONCLUSION

Xanthomas are indicators of underlying lipid abnormality. Hence, it should be diagnosed and treated as early as possible to reduce the complications. All family members should be screened even in the absence of cutaneous lesions. As tuberous xanthomas are seldom larger, this case is reported because of its atypically larger clinical presentation.

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## CONFLICT OF INTEREST:

The authors declare that they have no conflict of interest.

## STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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