IJCRT.ORG ISSN : 2320-2882

INTERNATIONAL JOURNAL OF CREATIVE RESEARCH THOUGHTS (IJCRT)

An International Open Access, Peer-reviewed, Refereed Journal

A Rare Presentation Of Tuberous Xanthoma In An Adult Patient: A Case Report And Literature Review

First Author: Dr Vivek kumar Sharma, Dr RPGMC Kangra at Tanda (H.P)

Second Author: Dr Shruti Sharma, Dr RPGMC Kangra at Tanda (H.P)

Corresponding Author: Dr Komal Sharma, Dr RPGMC Kangra at Tanda (H.P)

Abstract:

This case report describes a rare presentation of a nodular lesion on the index finger of an adult patient, histologically confirmed as tuberous xanthoma. The report highlights clinical features, histopathological findings, including Giemsa and Hematoxylin & Eosin (H&E) staining, and discusses the diagnostic process, management strategies, and the patient's clinical course.

Introduction:

Tuberous xanthomas are noncancerous skin lesions marked by accumulation of lipid-rich macrophages within the dermis, often emerging as a consequence of hyperlipidemic states. While they typically manifest at pressure points like the knees and elbows, the case of a 30-year-old male, is atypical due to the lesion's rare occurrence on the index finger. This anomaly presents a unique diagnostic challenge, as such lesions are usually linked with systemic lipid disorders, yet in this instance, the patient exhibited normal lipid levels. The case underscores the necessity of considering a wide differential diagnosis when encountering unusual cutaneous nodules, even in the absence of systemic symptoms.

Case Presentation:

A 30-year-old male, presented to the Dermatology Clinic at Zonal Hospital Dharamshala with a chief complaint of a slowly enlarging lesion on his right index finger (Figure 1). The lesion, located on the distal phalanx, was first noticed by the patient approximately 12 months prior to presentation. The nodule was painless and had a yellowish hue, prompting concerns due to its progressive, albeit slow, increase in size. Physical examination revealed a solitary, well demarcated, firm, nodular lesion measuring approximately 1 cm in diameter. The overlying skin appeared slightly taut, with a yellowish discoloration, characteristic of lipid-rich nodules. There was no tenderness on palpation, and the lesion was non-fluctuant, without evidence of warmth or erythema suggesting an inflammatory or infectious process. No similar lesions were noted elsewhere on the body. The patient denied any pruritus or discharge from the lesion. Patient's medical history was unremarkable. He denied any history of systemic diseases such as diabetes mellitus or cardiovascular disease. There was no reported history of similar lesions in his family which could suggest a genetic predisposition to lipid metabolism disorders. The patient's social history was non-contributory, and he denied the use of any medications, supplements, or tobacco products. He consumed alcohol socially but did not report any excessive use.

Diagnostic Assessment:

The patient presented with a lesion that posed a diagnostic challenge due to its non-specific clinical presentation. The differential diagnoses were broad, primarily including gouty tophus, rheumatoid nodule, infectious aetiologies such as mycobacterial or deep fungal infections, and other dermatological conditions that could present as nodular lesions. The FNA was non-diagnostic. It is not uncommon for FNA to yield insufficient tissue for definitive diagnosis, especially for lesions that are fibrous or have a paucity of cells, such as xanthomas. Consequently, an excisional biopsy was deemed necessary to obtain adequate tissue for histopathological diagnosis. The biopsy showed numerous foamy histiocytes, which are macrophages filled with lipid, and Touton giant cells, which are a type of multinucleated giant cell with a characteristic ring of nuclei (Figure 2 and figure 3). These findings are typical for xanthomas. Additionally, the presence of extracellular lipid deposition supported the diagnosis. This histological picture, in conjunction with the clinical presentation and the absence of systemic disease, confirmed the diagnosis of tuberous xanthoma.

Management and Outcome:

The patient underwent surgical excision of the lesion. Post-operative recovery was uneventful, and the lesion did not recur. Despite the normal lipid profile, the patient was advised on dietary modifications and regular follow-up to monitor for systemic involvement.

Discussion: The case illustrates an uncommon presentation of tuberous xanthoma located on the index finger, a rarity in both the medical literature and clinical practice. Tuberous xanthomas are traditionally associated with both primary and secondary hyperlipidaemias and manifest as firm, painless, yellowish nodules predominantly

over extensor surfaces such as the elbows, knees, and buttocks [1]. Their pathogenesis is linked to the accumulation of cholesterol within macrophages, leading to the characteristic foam cells observed histologically [2].

Interestingly, our patient did not exhibit any signs of systemic lipid abnormalities, which is an unusual finding. Cases of normolipemic xanthomas have been reported, although they are exceedingly rare [2]. In these instances, localized metabolic disturbances in lipid processing within the skin may be implicated, independent of systemic cholesterol levels [2].

Management of tuberous xanthomas typically involves addressing the underlying lipid disorder; however, in normolipemic cases, this strategy is less clear. Surgical excision remains the mainstay of treatment for symptomatic relief or cosmetic concerns, as was the successful approach in our case [1]. There is ongoing debate about the need for aggressive lipid-lowering therapy in normolipemic patients with xanthomas [3].

Conclusion:

Tuberous xanthomas, while typically associated with hyperlipidemia, can present in patients with normal lipid profiles. This case underscores the importance of considering tuberous xanthomas in the differential diagnosis of nodular lesions of the fingers, regardless of the lipid status. It also highlights the necessity for a multidisciplinary approach to management, involving dermatologists, pathologists, and primary care physicians.

Keywords:

Tuberous xanthoma, Cutaneous xanthomas, Hyperlipidemia, Lipid-laden macrophages, Histopathology, Touton giant cells, Dermatological manifestations, Normolipemic xanthoma.

References:

1. Shenoy C, Shenoy MM, Rao GK. Dyslipidemia in Dermatological Disorders. N Am J Med Sci. 2015 Oct;7(10):421-428.

2. Fleischmajer R, Tint GS, Bennett HD. Normolipemic tendon and tuberous xanthomas. J Am Acad Dermatol. 1981;5(3):290-296.

3. Yamamoto A, Matsuzawa Y, Yokoyama S, Funahashi T, Yamamura T, Kishino B. Effects of probucol on xanthomata regression in familial hypercholesterolemia. Am J Cardiol. 1986;57(16):29.

Figure 1 Clinical photograph: A nodular lesion noted on index finger

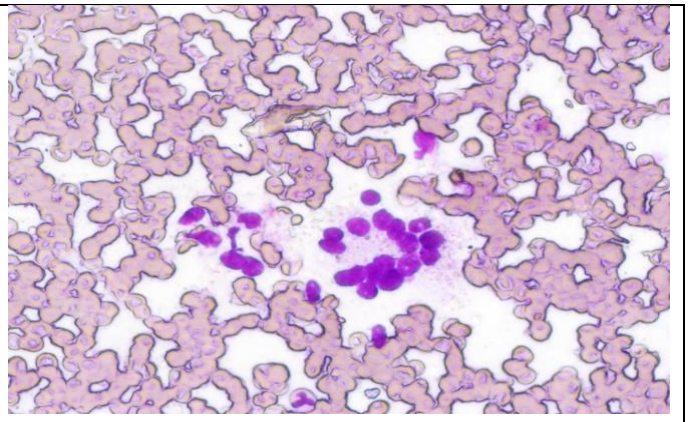

Figure 2Giemsa stain 400X: Touton shaped giant cells are seen in a haemorrhagic background

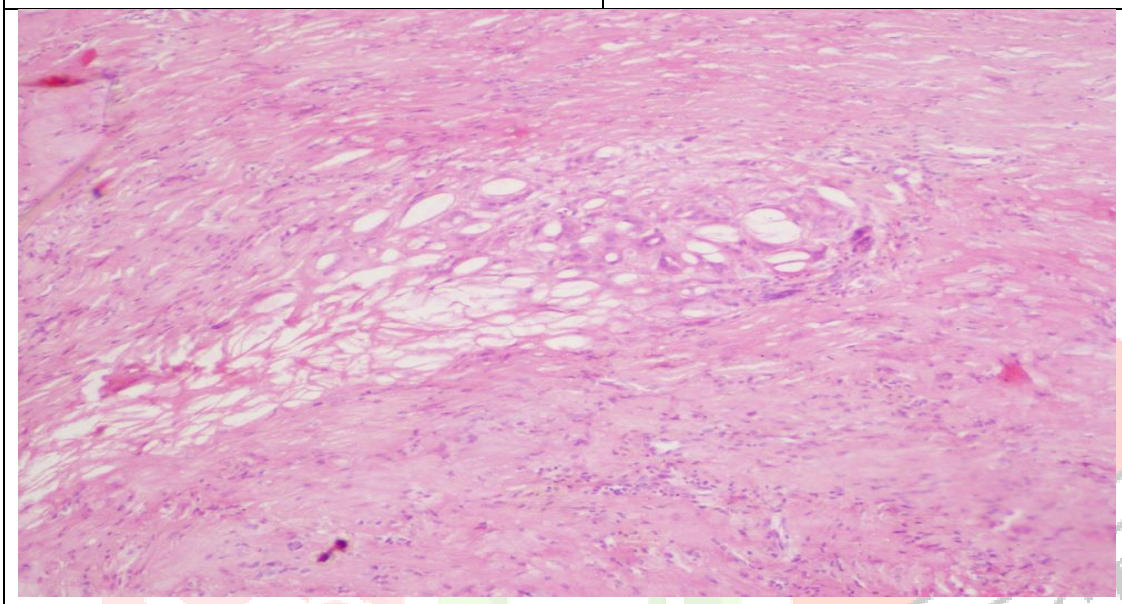

Figure 1 H&E stain 100X: Foamy histiocytes and fibrocytes are seen with extracellular lipid deposition