

## Case Report

# Bilateral prepatellar tuberos xanthomas unmasking a concealed lipid metabolic disorder: A case report

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## ABSTRACT

Tuberous xanthomas, though uncommon, may be the initial presentation of a concealed systemic lipid metabolic disorder. The typical imaging features, when aligned with the clinical and biochemical profile, can lead to a correct diagnosis for initiation of prompt and early treatment. We present a case of a 47-year-old female with bilateral prepatellar tuberos xanthomas as the initial clinical and imaging pointer for the diagnosis of familial dysbetalipoproteinemia. This case report highlights the importance of early recognition of these imaging findings of tuberous xanthomas for the diagnosis of underlying dyslipidemia and to prevent atherosclerotic cardiovascular complications.

**Keywords:** Hyperlipidemia, Lipid metabolic disorder, Metabolism, Prepatellar xanthoma, Tuberous xanthoma

## INTRODUCTION

Xanthomas are localized deposits of lipids in soft-tissue structures, including skin and subcutaneous fat (tuberous xanthomas) and tendons (tendinous xanthomas).<sup>[1]</sup> Tuberous xanthomas present as firm, yellow nodules over pressure areas such as the extensor aspect of the knees, elbows, and buttocks, while tendinous xanthomas are seen as fusiform enlargement or thickening of the affected tendons, most commonly the Achilles tendon.<sup>[2]</sup> Histopathologically, all forms of xanthomas are composed of aggregates of lipid-laden macrophages, known as foam cells, containing cholesterol and cholesterol esters.<sup>[3]</sup> Although these lesions are focal or multifocal in distribution, they may serve as early indicators of an underlying concealed systemic lipid metabolism disorder. They may also represent the initial clinical manifestation of such conditions.<sup>[1]</sup> From a vital clinical perspective, this deranged metabolic profile may remain concealed until the unfortunate occurrence of a fatal acute cardiovascular adverse event. This case report aims to highlight the distinct imaging characteristics of rare prepatellar tuberous xanthomas, which are less frequently seen compared to tendinous xanthomas, and underscore their diagnostic value as radiological markers of an underlying underdiagnosed dyslipidemic disorder such as familial dysbetalipoproteinemia (FD). It also reinforces the importance of preventive musculoskeletal radiology for mitigating potential morbidity and mortality.

## CASE REPORT

A 47-year-old woman presented with a 5-year-long history of focal nodular swelling along the anterior aspect of the right knee shortly followed by the left knee, with insidious onset and

gradual progression [Figure 1a]. The patient failed to recall any history of trauma to the knees. On local examination, the nodular swellings were symmetric on both sides with no erythema or cutaneous discharge on inspection and were firm and non-tender on palpation. The swellings were not fixed to the underlying patella or quadriceps and patellar tendons. A thorough physical examination revealed no evidence of any other similar swelling in the body.

Routine blood investigations revealed mild anemia (hemoglobin = 10.3 g/dL) and an abnormal lipid profile with nearly equally elevated total cholesterol (342 mg/dL) ( $n$ : <200 mg/dL) and triglycerides (339 mg/dL) ( $n$ : <150 mg/dL) and reduced low-density lipoprotein cholesterol (LDL-C) (85 mg/dL) ( $n$ : ~100 mg/dL) [Table 1]. The

| Biochemical parameter             | Result              | Biological reference value |
|-----------------------------------|---------------------|----------------------------|
| Hemoglobin                        | 10.3 g/dL           | >12 g/dL                   |
| Total leucocyte count             | 6,300/ $\mu$ L      | 4,000–11,000/ $\mu$ L      |
| Total cholesterol                 | 342 mg/dL           | <200 mg/dL                 |
| LDL cholesterol                   | 85 mg/dL            | ~ 100 mg/dL                |
| HDL cholesterol                   | 34 g/dL             | >40 mg/dL                  |
| VLDL cholesterol                  | 68 mg/dL            | 10–30 mg/dL                |
| apoB                              | 70 mg/dL            | <130 mg/dL                 |
| Fasting blood glucose             | 75 mg/dL            | <100 mg/dL                 |
| 2-h post-prandial blood glucose   | 186 mg/dL           | >200 mg/dL                 |
| Serum agarose gel electrophoresis | Broad $\beta$ -band | -                          |

LDL: Low-density lipoprotein, HDL: high-density lipoprotein, VLDL: Very-low-density lipoprotein

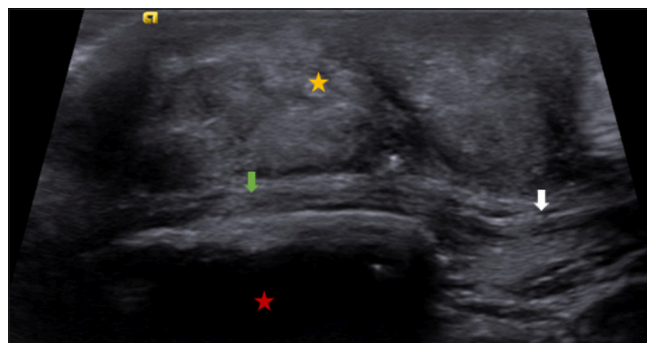


**Figure 1:** (a) Clinical photograph of the right knee showing a large nodular swelling along the anterior aspect. (b) Lateral plain radiograph of the right knee showing soft-tissue density swelling in the prepatellar region with normal underlying patella and distinct fat planes along the quadriceps and patellar tendons.

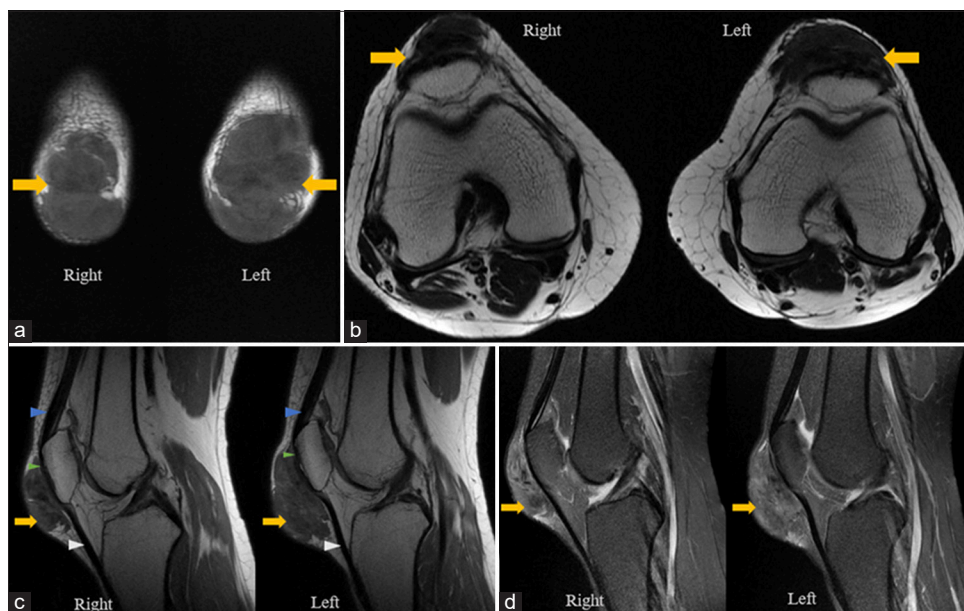
high-density lipoprotein (HDL) cholesterol was decreased (34 mg/dL;  $n$  > 40 mg/dL), and very-LDL (VLDL) was elevated (68 mg/dL;  $n$  = 10–30 mg/dL). Apolipoprotein B level was reduced (70 mg/dL;  $n$  < 130 mg/dL) with relatively decreased apolipoprotein A-I, yielding an increased cholesterol: HDL ratio (>10). Other routine parameters, including fasting glucose, liver and renal function tests, and thyroid profile, were within normal limits, ruling out secondary causes of dyslipidemia.

The patient underwent plain radiography of both knees in orthogonal planes, which revealed ill-defined, homogeneous soft-tissue density opacities in bilateral prepatellar regions, with no evidence of erosion, scalloping, or sclerosis of the underlying patellae [Figure 1b]. The fat planes adjacent to the quadriceps and patellar tendons were distinct. High-resolution ultrasonography of bilateral anterior knees showed lobulated soft-tissue mass lesions relatively hyperechoic to the adjacent muscles, separate from the adjacent tendons, with no significant internal vascularity [Figure 2]. The patient also underwent magnetic resonance imaging (MRI) of bilateral knees using a 3-Tesla MRI scanner, which revealed irregular and lobulated prepatellar soft-tissue mass lesions located in the subcutaneous plane with intermediate signal intensity (to muscle) on both T1- and T2-weighted sequences and higher signal than the tendons [Figure 3]. No evidence of fluid signal was noted within the lesions or any blooming foci on gradient-echo sequences. The lesions were not seen to be arising from the quadriceps or patellar tendon or even from the prepatellar quadriceps continuation. A subtle fat plane was also noted separating the patella from the prepatellar lesion. The internal architecture of the bilateral knee joints was normal.

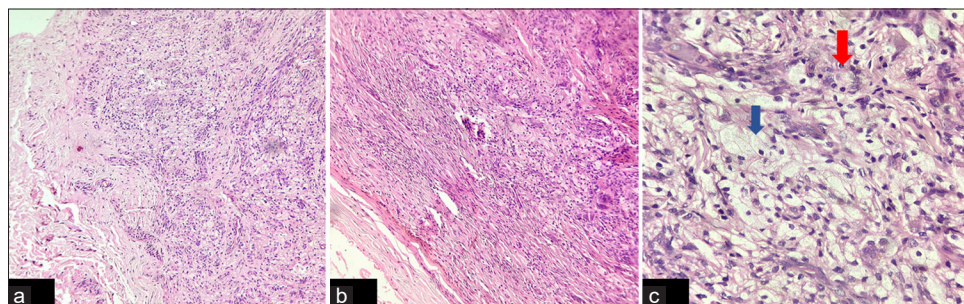
An ultrasound-guided biopsy showed dense infiltration of lipid-laden macrophages (foam cells) with finely vacuolated cytoplasm due to cholesterol accumulation. Scattered multinucleated foreign body giant cells were also seen with a few areas of fibrosis and chronic inflammatory infiltrates [Figure 4]. A histopathological diagnosis of xanthomas was made.



**Figure 2:** High-resolution ultrasound image of the right prepatellar xanthoma in longitudinal section shows an echogenic lobulated soft-tissue mass lesion (yellow star) separate from the underlying patella (red star), patellar tendon (white arrow), and prepatellar quadriceps continuation (green arrow).



**Figure 3:** Bilateral prepatellar tuberous xanthomas on magnetic resonance imaging – (a) Coronal T1W, (b) axial T2W, (c) sagittal proton-density, and (d) sagittal proton-density fat-saturated MRI images of bilateral knees show irregular, lobulated soft-tissue intensity mass lesions (yellow arrow in a-d) of intermediate signal intensity in prepatellar subcutaneous plane separate from the quadriceps (blue arrowhead in c) and patellar (white arrowhead in c) tendons, and prepatellar quadriceps continuation (green arrowhead in c) with normal underlying patellar marrow signal intensity. A subtle cleavage plane can also be seen between the lesions and the underlying patellae in (b).



**Figure 4:** Histomorphology of xanthoma—light microscopic images at (a and b)  $\times 100$  and (c)  $\times 400$  magnifications show lipid-laden macrophages (foam cells) (blue arrow), histiocytes, lymphocytes, fibrocollagenous tissue, and foreign body giant cells (red arrow).

Serum agarose gel electrophoresis revealed a broad  $\beta$ -band, suggesting impaired lipoprotein metabolism. An extended lipid profile also showed reduced apolipoprotein B (apoB) (70 mg/dL) ( $n$ :  $<130$  mg/dL).

Mixed hyperlipidemia with low LDL-C levels, broad  $\beta$ -band on gel electrophoresis, reduced apoB, and bilateral prepatellar tuberous xanthomas led to a working diagnosis of FD. *APOE* genetic testing was, however, not performed.

## DISCUSSION

Xanthomas are localized clusters of abnormal lipid-laden macrophages (foam cells) and present as an uncommon

manifestation of disturbance in systemic lipid metabolism. These may be associated with an elevated risk of atherosclerotic cardiovascular disease and occasionally, pancreatitis. Xanthomas may also be seen in individuals with a normal lipid profile. Tuberous xanthomas are clinical manifestations of abnormalities in lipoprotein metabolism (usually dysbetalipoproteinemia), whereas tendinous xanthomas are more commonly seen with familial hypercholesterolemia.<sup>[4,5]</sup> Certain genetic disorders exhibit varied phenotypic presentations of xanthomas, as highlighted in Table 2.<sup>[4-7]</sup>

Superficial prepatellar soft-tissue structures include the skin, subcutaneous fat, superficial prepatellar bursa, and fascia,

**Table 2:** Genetic lipid metabolism disorders causing tuberous and tendinous xanthomas.

| Disorder of lipid metabolism                              | Type of xanthoma (s)  |
|---|---|
| Familial hypercholesterolemia                             | <ul style="list-style-type: none"> <li>• Tuberous and tendinous xanthomas</li> <li>• Additional xanthelasma</li> </ul>                            |
| Familial dysbetalipoproteinemia (type III hyperlipidemia) | <ul style="list-style-type: none"> <li>• Tuberous and tuberoeruptive xanthomas</li> <li>• Xanthomas striatum palmaris (characteristic)</li> </ul> |
| Sitosterolemia (Phytosterolemia)                          | Tuberous and tendinous xanthomas  |
| Cerebrotendinous xanthomatosis                            | Tendinous xanthomas   |

along with prepatellar quadriceps continuation. Our case revealed symmetric prepatellar soft-tissue lesions without involvement of any tendon(s) or underlying bone(s). In addition to the clinical background, plain radiography and MRI findings ruled out the common differential diagnoses. Prepatellar bursitis (with possible etiologies including rheumatoid arthritis, gout, bacterial infection, and tuberculosis) was ruled out as the lesions were painless and non-tender with no evidence of intralesional fluid on ultrasound or MRI. A 5-year-long history, bilaterality of the lesions, and absence of aggressive imaging features, including invasion of adjacent structures, made the possibility of soft-tissue sarcomas such as liposarcoma and synovial sarcoma unlikely. Since there was no history of trauma to the knees and the lesions were bilateral with no signs of fluid collections on imaging, the diagnosis of Morel-Lavallée lesion was discounted.

Bilaterality, intermediate signal of the lesions relative to muscle and higher signal than tendons on MRI, involvement of the extensor aspect of the knees in the subcutaneous plane, and absence of bone erosion/scalloping pointed toward a provisional diagnosis of tuberous xanthomas, which was reinforced by an elaborate lipid profile and confirmed on histopathology.<sup>[8]</sup> Other possible sites of tuberous xanthomas include elbows, palms, feet, and buttocks, which were free of any lesions in our case.

FD (type III dysbetalipoproteinemia; Fredrickson classification) is an uncommon disorder of lipid metabolism with a prevalence of 0.1–0.4% in the general population. It is characterized by impaired clearance of triglyceride-rich lipoprotein remnants due to low affinity to LDL receptors, leading to accumulation of chylomicrons and VLDL remnants. These are associated with *APOE* genetic abnormalities.<sup>[9,10]</sup>

Diagnosis is suggested by mixed hyperlipidemia (nearly equally elevated levels of cholesterol and triglycerides),

low LDL-C and apoB levels, and confirmed by detection of *APOE*  $\epsilon 2\epsilon 2$  genotype. Broad  $\beta$ -band on gel electrophoresis represents accumulation of remnant lipoproteins and is a hallmark of FD. Characteristic phenotypic features include tuberous and tuberoeruptive xanthomas over pressure areas such as elbows and knees, and palmar xanthomas in hand and wrist creases. Tendon xanthomas are not usually seen.<sup>[4,9]</sup>

It is of vital importance to recognize this biochemical derangement as it carries a 5–10 times higher risk of premature ischemic heart disease and increased risk of peripheral arterial disease compared to the general population. Calorie and saturated fat restriction with a combination of statins and fibrates forms the cornerstone of management and aids in mitigating the atherosclerotic cardiovascular disease risk.<sup>[9]</sup>

## CONCLUSION

Bilateral prepatellar tuberous xanthomas may serve as the first indicator of an otherwise concealed and uncommon systemic metabolic disorder such as FD. The typical imaging features, when aligned with the characteristic site(s) of involvement, clinical history, and biochemical lipid profile, are essential for diagnosing these xanthomas and can be confirmed on histopathological correlation. It is therefore pertinent for a radiologist to be aware of such entities, as prompt recognition can lead to initiation of early preventive management to mitigate atherosclerotic cardiovascular complications.

**Ethical approval:** The Institutional Review Board at Vardhman Mahavir Medical College and Safdarjung Hospital Institutional Ethics Committee has waived the ethical approval, number 2025-156/CR-89, for this study, dated 8th July 2025.

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