



# An Unusual Case of Xanthoma of Bilateral Achilles Tendon with Gouty Infiltration: A Rare Case Report

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## Abstract

Achilles tendon xanthoma is a benign and rare disease with a high incidence in patients having familial hypercholesterolemia. Patients present with or without pain and thickened tendons. Gout is seen in patients with hyperuricemia. Coexistence of xanthoma and gout is extremely rare. We searched the PubMed literature with 'Xanthoma' and 'Gout' as keywords and could find only one case report. Imaging modalities such as radiography and ultrasonography, play a vital role in diagnosing this condition early, hence helping the patient to commence the use of potentially lifesaving lipid-lowering therapeutic agents. Magnetic resonance imaging is helpful in delineating the morphological changes, exact measurements, and eventually in treatment planning. It helps to rule out early involvement of other tendons by the same pathology. We present a case of a 25-year-old male patient who presented to our institute with bilateral large symmetrical swelling in the posterior aspects of ankles for 4 years, with normal serum and blood parameters. Bilaterality, enormous size and normal blood and serum parameters make this case unique.

## Keywords

- ▶ Achilles tendon
- ▶ xanthoma
- ▶ gout
- ▶ ultrasonography
- ▶ radiography
- ▶ MRI

## Introduction

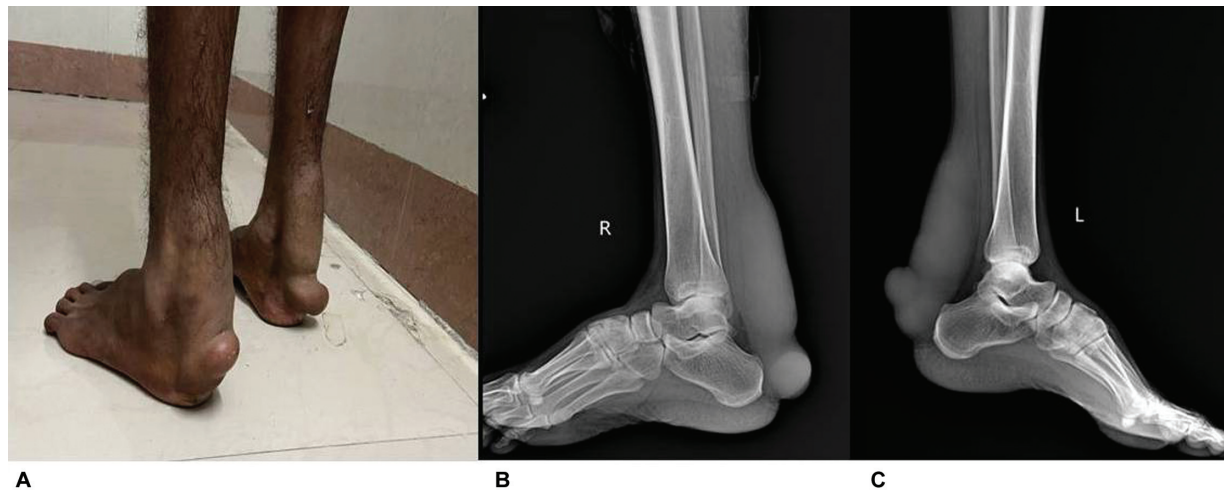
Xanthomas are rare, nonneoplastic lesions seen in the form of accumulation of low-density lipoproteins (LDL) in the tendon or synovium.<sup>1,2</sup> The high levels of LDL are oxidized and engulfed by the macrophages resulting in the formation of foam cells.<sup>2</sup> The xanthomatous deposition occurs most frequently in the tendons close to the skin and predominantly in the extensor tendons such as the Achilles tendon, patellar tendon, and the extensor tendons of the hand.<sup>3</sup> Deposition of LDL in the Achilles tendon triggers an inflammatory response, which leads to the thickening of the tendon, presenting with symptoms similar to tendinopathy.<sup>4</sup> There have been a few

cases with the formation of xanthomas in normolipemic states. In such cases the xanthomatous deposits are made of phytosterols (sitosterolemia, cerebrotendinous xanthomatosis) and there may be altered lipoprotein content or structure or an underlying lymphoproliferative disease with xanthomatization of cells infiltrating the dermis. It is also seen in people exposed to endemic injuries, mainly over the gluteal region, extensor surface of the limb, and flexor surface of the joints.<sup>5,6</sup> In the majority (90%) of the cases, involvement is bilateral.<sup>7</sup> In some cases, the tendon becomes edematous, while in other instances there may be interference in the functions of the tendon, leading to achillodynia, cosmetic disfiguration, and very rarely spontaneous tendon rupture.<sup>8</sup>

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**Fig. 1** (A) Clinical image shows soft tissue swellings in bilateral lower extremities extending from the lower calf to the posterior regions of the ankle, with bilobed appearance on both sides. Lateral plain radiographs of right (B) and left (C) lower half of legs and ankles show diffusely thickened Achilles tendon with soft tissue mass extending cranially from the lower third of the calf to the insertion site of the Achilles tendon along the expected course of the tendon.

## Case Report

We report a case of a 25-year-old male who presented with lobulated swelling (►Fig. 1A) in the posterior aspects of the lower-thirds of bilateral legs, extending up to the ankle joints for 4 years. The patient gave no history of trauma or previous surgery. There was no other significant history. On physical examination during the time of USG, the masses were firm, nontender, nonreducible, and nonmobile. Signs of inflammation in the form of discoloration of the overlying skin were not seen. There was no restriction of movements around the ankle joint and no probe tenderness. The lipid profile was normal, serum uric acid levels were lower than the normal reference value, while serum homocysteine levels were slightly elevated as shown in ►Table 1.

Radiographs showed bilateral soft tissue masses in the posterior lower calf and ankle regions with a bilobed appearance. The masses extend from the lower calf up to the ankle along the course of the Achilles tendon (►Fig. 1B, 1C). It measured  $\sim 90 \times 45$  mm (cranio-caudal (CC)  $\times$  antero-posterior (AP)) on the right side and  $102 \times 50$  mm (CC  $\times$  AP) on the left side. Calcifications or underlying bone changes were not seen.

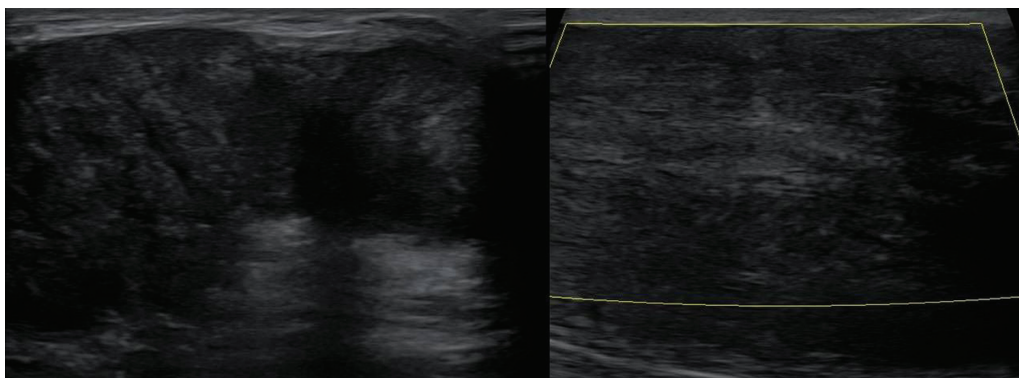
On USG, lanceolate-shaped thickening of Achilles tendons extending from the lower calf to the heel was noticed, which showed loss of normal fibrillar pattern and appeared heterogeneously hypoechoic (►Fig. 2). The involved segment of tendons measured  $100 \times 40 \times 40$  mm (CC  $\times$  AP  $\times$  transverse) on the right side and  $110 \times 40 \times 45$  mm (CC  $\times$  AP  $\times$  transverse) on the left side. Anechoic/cystic spaces were not

**Table 1** Biochemical laboratory parameters with the normal reference range

Biochemical laboratory parameter analysis		
Parameters	Obtained value	Reference value
Cholesterol (mg/dL)	138	<200
HDL (mg/dL)	48	40–60
LDL (mg/dL)	64.6	<100
VLDL (mg/dL)	25.4	7–40
Non-HDL cholesterol (mg/dL)	90	<130
Triglycerides (mg/dL)	127	<150
Cholesterol total/HDL ratio	2.88	0–4.0
LDL/HDL	1.35	0–3.5
Uric acid (md/dL)	2.6	3.5–7.2
Homocysteine (Umol/L)	<b>21.38</b>	< 16.20
Lipoprotein A (mg/dL)	23.9	Up to 30

Abbreviations: HDL, high-density lipoprotein; LDL, low-density lipoprotein; VLDL, very low-density lipoprotein.

Note: Bolded values suggest S.uric acid being normal range, inspite of patient having Gouty, uric acid crystals embedded in the tendon. Also to note that S.homocysteine is higher than normal.



**Fig. 2** Sagittal sonogram of the Achilles tendon showing a heterogeneously hypoechoic lesion with diffusely thickened Achilles tendon with loss of the normal fibrillar pattern. No color uptake is seen on color Doppler imaging.

seen within the lesions, and color flow was absent on color Doppler. After eliciting relevant history and ruling out other pathologies, we gave the possibility of xanthoma of the Achilles tendon as the first differential.

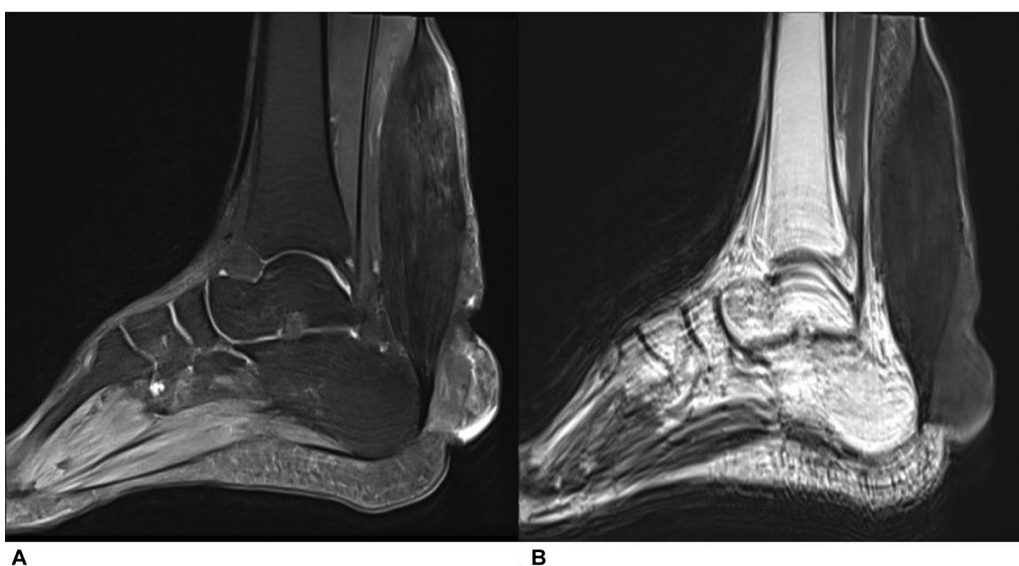
MRI was done for confirmation and to know the extent of the lesion, which showed thickened Achilles tendons with a maximum anteroposterior thickness of 3.5 cm and an approximate length of 13.5 cm craniocaudally on the left side. The thickened tendons showed heterogeneous signal intensity on proton density-weighted (PDW) images with a few high signal areas in between. On T1-weighted images, the tendons were hypointense, while heterogeneous hyperintense lesions were seen in the subcutaneous plane posterior to the insertion of Achilles on PDW sequence, appearing uniformly hypointense on T1-weighted sequence. Left-sided peroneus longus tendon was also involved, showing similar signal intensity changes as seen in Achilles tendon (► **Fig. 3**).

Fine needle aspiration cytology was performed, which showed mildly cellular smears consisting of numerous singly

scattered round to oval cells with round to oval nucleus and moderate amount of vacuolated cytoplasm. A few multinucleate giant cells were seen. Background appeared lipoidal. Numerous needle-shaped crystals suggestive of uric acid and plate-shaped crystals suggestive of cholesterol were also seen. A diagnosis of Achilles tendon xanthoma along with uric acid crystals (gout) was rendered on FNAC (► **Fig. 4**). On histopathology, the lesion was seen arranged in sheets amidst fibromuscular tissue composed of round to polygonal cells with round to oval nucleus and moderate to abundant amount of vacuolated cytoplasm and numerous multinucleated giant cells. Numerous needle-shaped crystals were also seen, thus confirming the cytological diagnosis (► **Fig. 5**).

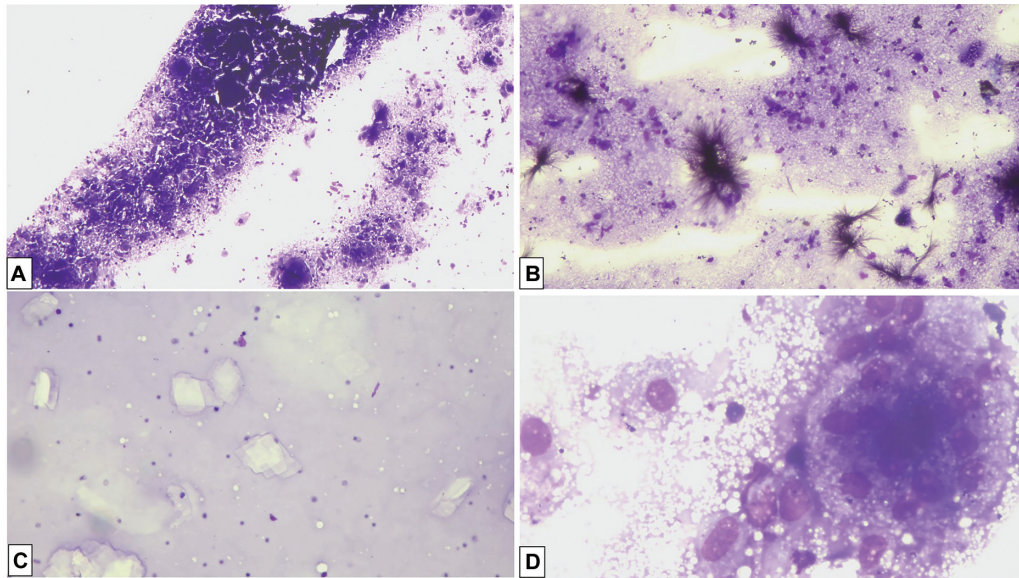
## Discussion

Xanthomas are benign, rare, nonneoplastic lesions, which are formed by accumulation of collagen, lipid-laden macrophages, giant cells, and inflammatory cells in response to the

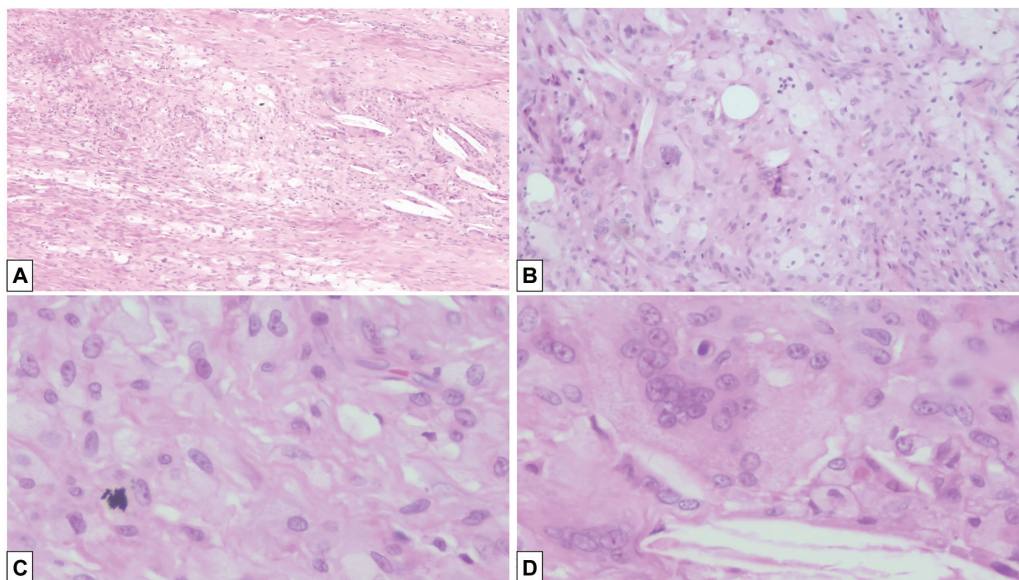


**Fig. 3** Images in the sagittal plane, proton density-weighted (A) showing thickened Achilles tendon with linear high signal stripes and heterogeneous signal intensity lesion with focal areas of high signal intensity in the subcutaneous plane, posterior to the site of insertion of the Achilles tendon. T1-weighted images (B) showing the thickened and hypointense tendon with hypointense lesion in the subcutaneous plane, posterior to the site of insertion of the Achilles tendon.





**Fig. 4** Cytology. (A) Mildly cellular smears consisting of singly scattered xanthoma cells (x50 magnification, MGG stain), (B) singly scattered xanthomatous cells in a lipoidal background along with needle-shaped crystals of uric acid (x100 magnification, MGG stain), (C) plate-shaped cholesterol crystals in a fluid background (x100 magnification, MGG stain), (D) high-power view showing xanthomatous cells with foamy vacuolated cytoplasm along with multinucleated giant cells (x400 magnification, MGG stain) MGG: May–Grunwald Giemsa.



**Fig. 5** Histopathology (A) Low-power view showing a lesion composed of round to polygonal cells with abundant foamy vacuolated cytoplasm and a few needle shaped crystals (x50 magnification, H and E stain). (B) Proliferation of xanthoma cells and multinucleate giant cells amidst fibromuscular tissue (x100 magnification, H and E stain). (C) High-power view showing xanthoma cells with minimal nuclear atypia (x400 magnification, H and E stain). (D) High-power view showing multinucleate giant cells and cholesterol crystals (x400 magnification, H and E stain) H and E: hematoxylin and eosin.

deposition of LDL in the tissue. It can occur in the skin, especially eyelids and are less commonly seen in the tendons and synovium.<sup>9</sup> Tendinous xanthomas most commonly involve the extensor tendons of the hand and elbow, the Achilles tendon and the patellar tendon. Xanthomatosis of the Achilles tendon is rare, but an early and accurate diagnosis is warranted due to clinical implication of strong association between xanthomatosis and primary hyperlipidemia

(predominantly of types IIa and III). It usually occurs in patients with familial hypercholesterolemia since childhood and develop by 20 years of age in patients with heterozygous hypercholesterolemia.<sup>10</sup> It may be a precursor to coronary artery disease.<sup>11</sup> There have been case reports of their development in patients on antiretroviral therapy due to drug-induced hyperlipidemia.<sup>12</sup> Several studies have observed that not all hyperuricemia patients suffer from gout

and not all gout patients have hyperuricemia.<sup>13,14</sup> In our case, the patient had normal blood lipid levels and low serum uric acid levels unlike other case report, where the patient had Achilles tendon xanthoma and gout, but with hypercholesterolemia and hyperuricemia.<sup>15</sup>

Clinical manifestations of the Achilles tendon xanthomas depend predominantly on the size of the lesions and the smaller lesions are unnoticeable, whereas the larger clinically apparent lesions are mostly noticed with cosmetic disfigurement. Patients may also have localized pain, irritation, and restriction of movements.<sup>16</sup>

Plain radiography is often the first imaging modality in these cases, and lateral views are used for the evaluation of anteroposterior diameter of the Achilles tendon.<sup>17</sup> Xanthomatosis appears as an abnormal thickening with noncalcified soft tissue masses.<sup>18,19</sup> A few studies showed that in patients with hyperlipidemia, the Achilles tendon was thickened with the thickness ranging from 7.5 to 21.5 cm in contrast to the normal thickness which is < 0.7 cm in males and < 0.6 cm in females.<sup>18</sup>

Ultrasonography is widely performed for the detection of xanthomatosis in various tendons. The involved tendons show focal hypo-echoic areas or a diffuse heterogeneous echo pattern with loss of normal fibrillar pattern. It is also a practical modality to monitor the treatment response.<sup>20</sup>

Computed tomography is usually not considered due to its low contrast within soft tissue lesions.<sup>21</sup> MR imaging depicts morphologic and signal abnormalities in the form of loss of normal flat or concave ventral margin of the Achilles tendon as seen in our case. Previously reported cases showed uniform low signal intensity on MR images, indicating collagen fibers and intervening high signal intensity areas that could be cholesterol-laden foam cells and/or an inflammatory response. Our case showed slightly hyperintense signal in the lesion along with the fibrillar pattern, which could be due to the coexistence of xanthoma and gout. However, more research is required to evaluate the genesis of various MR appearances. MRI is also highly sensitive in ruling out early disease in other visualized tendons and in underlying bone and marrow. The involved tendons show higher signal intensity on T1- and T2-weighted spin echo images compared with the normal tendon.<sup>22</sup> In our case, sonography could not indicate the presence of gouty crystal deposition, likely due to predominant xanthomatous component appearing inhomogeneous in attenuation. In some unusual cases, patients may have necrosis and hemorrhage.<sup>16</sup>

The differentials for bulky, enlarged, and disfigured Achilles tendon include xanthoma, tendinopathy, chronic degeneration, tendonitis, peritendinitis, bursitis, trauma, nodules of rheumatic arthritis, tophaceous gout, infection, and neoplasm.<sup>16</sup> However, owing to the clinical history, bilateralism and imaging findings, most of them were excluded. The nodules in cases with gout can be seen at any part of the joints, articular cartilage, synovial membranes, joint capsules, tendons, ligaments, interosseous or subcutaneous tissue.

Treatment options could range from conservative treatment to total excision with reconstruction or subtotal/partial resection. Surgery is reserved only for patients having severe disfigurement, pain or restriction of movements with only

partial resection and removal of the lesions to preserve the functions. Our case did not present with restriction of movements.

## Conclusion

In conclusion, xanthoma of the Achilles tendon is a rare disorder, appearing as lanceolate thickening of tendons on imaging, which is the mainstay in its diagnosis, if presented with relevant history. In case of inconclusive laboratory parameters, both imaging and microscopy play a key role in early and accurate diagnosis for effective management and alerting the patient and physician alike for its association with hyperlipidemia, hypercholesterolemia, gout, and cardiac disease. Ultrasonography is the modality of choice, while MRI is used for the extent of involvement, treatment planning, and visualizing early changes in other tendons in vicinity. Treatment ranges from conservative to excision, with the surgical option reserved for patients with severe cosmetic disfigurement, pain, and restriction of movements.

## Funding

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## Conflict of Interest

None declared.

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