

Case Report

Imaging of bilateral tendoachilles xanthomatosis in a young woman

Suba Arul Devi¹, K. M. Machaiah¹, Amanda C. Pinto², Santosh P. V. Rai¹

Departments of ¹Radiodiagnosis, and ²Pathology, Kasturba Medical College, Mangalore, Manipal Academy of Higher Education, Manipal, India.

*Corresponding author:

Suba Arul Devi,
Junior Resident, Department
of Radiodiagnosis, Kasturba
Medical College, Mangalore,
Manipal Academy of Higher
Education, Manipal, India.

subamuthamizhan@gmail.com

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ABSTRACT

Bilateral tendoachilles xanthomatosis is a rare entity and is frequently found in association with the primary hyperlipidemia. It is characterized by the deposition of cholesterol in tendons. An early diagnosis with different imaging modalities changes the course of the disease. This case report illustrates the bilateral tendoachilles xanthomas on MRI imaging in a patient who presented with painless progressive swelling of bilateral ankle joints for several years. Intralesional subtotal resection was successfully done and histological evaluation further confirmed the diagnosis.

Keywords: Xanthomatosis, Familial hypercholesterolemia, Tendon sparing procedures, Tendon xanthomas, Achilles tendon xanthoma

INTRODUCTION

Xanthomas are rare non-neoplastic lesions that are frequently seen within the tendons and synovium.^[1] It is essentially pathognomonic of heterozygous familial hypercholesterolemia (FH). FH is an autosomal dominant disorder with a prevalence of approximately one in 500 individuals.^[2]

Histologically tendinous xanthoma is characterized by the accumulation of multinucleated giant cells, cholesterol crystals, and other inflammatory cells. They appear clinically as a hard lump over the tendon region beneath the skin. The Achilles tendon, patellar tendon, and extensor tendon over the digits and elbow are the most common sites of involvement. These are most commonly seen in the 3rd decade of life with female predominance as compared with males (4:3).^[3]

It is commonly associated with raised lipid levels. It occurs due to a defect in the LDL receptor and it is associated with FH.^[4] Achilles tendon xanthoma manifests clinically depending on the site of involvement and the size of the lesion. Some lesions are purely asymptomatic. Some lesions are clinically present with a large mass lesion that impair joint mobility, severe pain, and cause cosmetic disfigurement.

In the presence of a xanthoma, the tendons appear as a diffusely enlarged globular structure with a loss of normal architecture. On x-rays, it appears as an area of abnormal soft-tissue thickening without calcification and bony erosion. MRI is the imaging modality of choice as it allows the accurate delineation of soft-tissue characteristics and provides accurate diagnosis preoperatively.

The typical features of xanthomas can be identified in imaging modalities, but the definitive investigation to confirm the diagnosis of tendoachilles xanthomas is histopathology. It shows the

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clusters of multinucleated giant cells, hemosiderin deposits, and histiocytes (foamy macrophages).^[5]

Earlier diagnosis of Achilles tendon xanthoma through appropriate imaging modality and correctly timed intervention is essential to reduce morbidity and, thereby, eliminates the need for complete resection. Here, we are presenting a case of bilateral tendoachilles xanthomatosis diagnosed in the earlier stage by MRI in a young woman with primary hypercholesterolemia and subtotal resection with tendon-sparing surgery was successfully done and it significantly improved the patient outcome.

CASE REPORT

The study was conducted only after obtaining permission from the Institutional Ethics Committee. A 21-year-old female presented with complaints of painless progressive swelling over the bilateral ankle and difficulty in walking. Physical examination revealed diffuse swelling noted in the region of tendoachilles insertion in bilateral ankle joints [Figure 1]. Laboratory investigations revealed raised LDL - 568 mg/dL and triglyceride levels - 152 mg/dL. Her parents were simultaneously investigated to rule out FH, where both were found to have normal cholesterol levels in their lipid profile. Neurological evaluation has been done for our patient to look for unsteadiness of gait, frequent falls, chronic diarrhea, early-onset cataracts, and developmental delay that may be a hallmark of cerebral xanthoma and were also ruled out.

A plain radiograph of both ankles was done in view of progressive swelling which showed bilateral soft-tissue opacification in the region of tendoachilles insertion with no evidence of calcification and bony abnormalities. USG was done as a next-line investigation which revealed a diffusely enlarged heterogeneous



Figure 1: Physical examination revealed diffuse swelling was noted in the region of Tendoachilles insertion in bilateral ankle joints.

bilateral Achilles tendon with multiple hypoechoic foci within it resulting in the loss of the normal fibrillary pattern of the Achilles tendon. MRI was done as a further investigation to look for lesion characteristics which showed bilateral symmetrical fusiform enlargement involving the Achilles tendons which are heterogeneously hypointense on T1 sequence and appear hypointense on the T2 sequence. T1 coronal [Figure 2a] showed diffuse hypointensities with interspersed striations of hyperintensities and T2 axial images [Figure 2b] showed diffuse hypointensities in bilateral Achilles tendons measuring for a length of 12 cm with a maximum AP diameter of 3.6 cm. Subcutaneous tissue overlying the ankle joint shows mild thickening.

Under spinal anesthesia, surgery was performed with the patient in the prone position and both ankles were dorsiflexed throughout the surgical procedure. A midline incision was made over the soft-tissue swelling region. A diffusely enlarged swollen Achilles tendon was exposed [Figure 3] and another midline vertical incision was made over the tendon showing a xanthoma lesion. Resection of localized xanthomatous tissue was made with a tendon-sparing procedure performed by preserving the surrounding normal tendon tissues.

Histopathology confirmed the diagnosis of tendoachilles xanthomatosis, which revealed the florid cholesterol deposition surrounded by foreign body giant cells with sheets of foamy histiocytes and no malignant cells [Figure 4].

DISCUSSION

Xanthomas are pseudotumors of connective tissue characterized by an accumulation of foam macrophages, multinucleated giant cells, and other inflammatory cells. These tumors often present macroscopically as a yellowish and well-encapsulated lobulated mass lesion due to the significant deposition of cholesterol crystals.

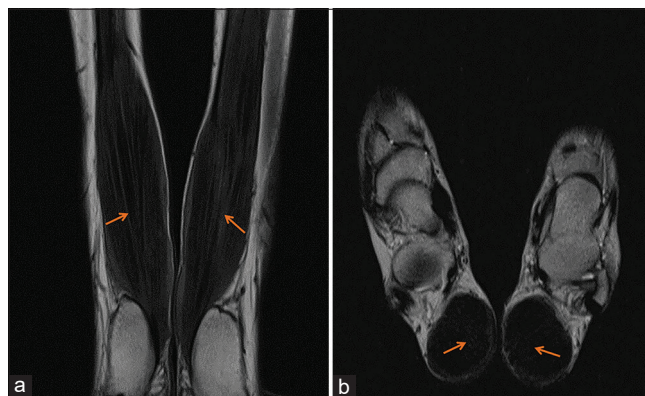


Figure 2: (a) T1 coronal (orange arrow mark) showed diffuse hypointensities with interspersed striations of hyperintensities within bilateral Achilles tendon. (b) T2 Axial MRI imaging (orange arrow mark) showing bilateral diffusely enlarged hypointense Achilles tendon.

Tendon xanthomas are the most classical finding of cerebrotendinous xanthomatosis and most commonly occur in the second or third decade. Lipid deposition is also responsible for the appearance, of palpebral xanthelasma.

Homozygous or heterozygous FH is the most common state associated with tendinous xanthomatosis. Achilles tendon xanthomas have also been found associated with some types of hyperlipidemia and chromosome mutations or with non-FH.^[6]

Careful clinical assessment of the Achilles tendon often lacks sensitivity. Different imaging modalities have been used to evaluate these xanthomas such as radiographs of particular joints or regions, CT, MRI, and ultrasound. The plain radiograph and

MRI depict the typical findings of xanthomas and the diagnosis was often confirmed by histological evaluation.

A plain x-ray is usually the initial modality of choice which was showing the bilateral and symmetrical thickening of soft tissues in the region of the bilateral Achilles tendon and no evidence of calcification.

Sonography is a low cost, readily available, and simple modality used for the identification of xanthomatosis and it is superior to gross clinical assessment. Achilles tendon xanthomas have been described either as hypoechoic nodules of lipid deposits within an affected tendon or as having a diffusely heterogeneous echo pattern. In our case, it showed a diffusely enlarged heterogeneous bilateral Achilles tendon with multiple hypoechoic foci of lipid deposits within it resulting in the loss of the normal fibrillary pattern of the Achilles tendon.

MRI is a highly specific imaging modality used to diagnose tendinous xanthomas and further determine underlying tendon anomalies. It showed the typical imaging features of xanthoma in our case such as bilateral symmetrical fusiform enlargement with altered signal intensities seen involving the Achilles tendons and mild thickening of subcutaneous tissue overlying the ankle joint.

Here, in our case, histology confirmed the diagnosis of tendinous xanthomata and helps to differentiate it from other conditions with similar imaging features such as tophaceous gout, chronic mucoid tendon degeneration, and tendinosis.

Although small areas of hypointensities with interspersed striations of hyperintensities are seen on T1 STIR mainly due to tendon enlargement, it can also be found in diseases of other etiologies which diffusely involve the tendon, thus limiting the diagnostic usefulness of MRI as a high specific modality.[Table 1] illustrates the differential diagnosis of diffuse Achilles tendon enlargement.

An early diagnosis of xanthoma modifies the disease progression and alters the treatment process, improving a clinical status of a patient. The common complication following surgical reconstruction of the Achilles tendon repair is re-rupture which occurs in about 2–8% of cases^[7,8] Various authors suggested the use of various tendons to fill the gap after surgical excision. The usual options are gracilis, the tensor fascia lata, FHL, and semitendinous for reconstruction.^[9] Dinçel *et al.*^[10] proposed tendon reconstruction in case of a large defect with quadriceps



Figure 3: Intraoperative surgical image (black arrow mark) showing a diffusely enlarged swollen Achilles tendon.

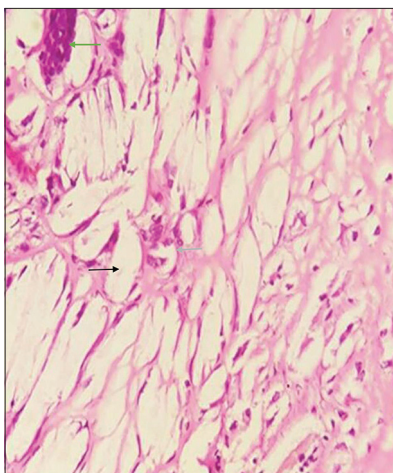


Figure 4: Histopathology (×400) image revealed the florid cholesterol cleft deposition (black arrow mark) surrounded by foreign body giant cells (gray arrow mark) with sheets of foamy histiocytes (green arrow mark).

Table 1: The differential diagnoses of diffuse Achilles tendon enlargement.

Differential diagnoses

- Tophaceous gout
- Chronic tendon degeneration
- Tendinosis
- Partial-thickness tears
- Giant cell tumor of the tendon sheath

tendon graft and bony fragments. Pearce *et al.*^[11] suggested an FHL graft for reconstruction. Gallant *et al.*^[12] proposed the reconstruction with the peroneus brevis tendon.

In our study, early diagnosis allowed the patient treated with subtotal resection and tendon-sparing procedure with at least 50% of the tendon salvaged. Then, our patient started improving after 2 weeks and started independent walking after 4 weeks and significantly improved clinically with no evidence of recurrence thereafter and medically managed for hyperlipidemia. At the time of discharge, our patient lipid profile was in the normal range.

CONCLUSION

Bilateral tendoachilles xanthomatosis in a younger age group is a rare entity and if not treated at the appropriate time, it leads to significant adverse outcomes. Accurate characterization of the lesion is made possible with the use of highly specific imaging modalities such as MRI. Earlier diagnosis through appropriate imaging modality and correctly timed intervention through surgery is essential to improve morbidity and reduce the chance of recurrence.

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TEACHING POINTS

1. Tendoachilles xanthomatosis is a rare non-neoplastic lesion characterized by the accumulation of foamy macrophages, multinucleated giant cells, and other inflammatory cells.
2. The most common sites are the Achilles tendon, extensor tendon of the elbow, and patellar tendon.

MCQs

1. Tendoachilles xanthomatosis is characterised by the deposition of
 - a) Proteins
 - b) Cholesterol crystals
 - c) Calcium
 - d) Uric acids
- Answer key: b
2. Most common site of involvement of tendoachilles xanthomatosis
 - a) Extensor tendons of elbow
 - b) Supraspinatus tendon
 - c) Triceps tendons
 - d) Quadriceps tendon
- Answer key: a

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

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