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Original Research

Atypical Imaging Findings of Achilles Tendon – Xanthomatosis

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ABSTRACT
Achilles tendon xanthoma is a rare condition characterized by the accumulation of lipid-laden macrophages within the tendon, often associated with lipid metabolism disorders, particularly hyperlipidemia and familial hypercholesterolemia. This case study presents a 60-year-old female with long-standing bilateral, multi-lobulated subcutaneous swelling over the Achilles tendons, with no signs of inflammation or joint pain. Imaging studies, including X-ray, ultrasound, and MRI, revealed bilateral Achilles tendon enlargement and characteristic lobulated soft tissue masses without signs of tendon rupture or calcification, confirming the diagnosis of Achilles tendon xanthoma. Laboratory investigations showed elevated cholesterol levels, supporting the link between tendon xanthomas and lipid disorders. The condition is often confused with tendinitis, tendon degeneration, or tophaceous gout, but imaging played a critical role in distinguishing it from these conditions. Early diagnosis and management of hyperlipidemia are essential to prevent potential tendon damage and complications.

Keywords: Achilles tendon xanthoma, hyperlipidemia, ultrasound, MRI, tendon pathology.

Introduction

Achilles tendon xanthomatosis is a rare manifestation of tendon pathology, often associated with systemic lipid metabolism disorders such as familial hyperlipidemia. Xanthomas are non-neoplastic lesions that form as a result of lipid-laden macrophages accumulating in tissues, primarily tendons, synovium, and skin. The Achilles tendon, being a common site for such lesions, frequently presents as a bilateral condition, often in the context of elevated lipid levels, particularly hyperlipidemia [1, 2]. These lesions are typically painless, though they can lead to tendon degeneration, rupture, or functional impairment over time, making their identification crucial for early intervention.

Despite its rarity, Achilles tendon xanthomatosis can be associated with a variety of systemic conditions, including familial hypercholesterolemia, which is often accompanied by other lipid metabolism abnormalities [3, 4]. The imaging findings of these lesions are important for distinguishing them from other tendinopathies or conditions like gout, which may also involve the Achilles tendon [5, 6]. Radiographic and ultrasonographic imaging have been widely used to characterize these lesions. Ultrasonography, in particular, has proven effective in identifying the characteristic hypoechoic and lobulated masses, while magnetic resonance imaging (MRI) can offer more detailed insights into the heterogeneity of the tendon structure [7, 8].

Imaging modalities such as MRI and ultrasound provide valuable diagnostic tools in assessing the extent of tendon involvement and in ruling out other possible causes of tendon swelling, such as tendinitis or gouty infiltration [9, 10]. The presence of concomitant conditions, like gout, can complicate the diagnosis and necessitate careful interpretation of imaging findings to differentiate between the two [11, 12]. Early detection and differentiation of Achilles tendon xanthomas are essential to prevent further complications, including tendon rupture, a potential consequence of untreated cases [13, 14].Although tendon xanthomas are generally benign, their association with systemic hyperlipidemia underscores the importance of recognizing these lesions not only for local management but also for addressing the underlying metabolic disorder [15].

Case Study

A 60-year-old female was referred for an ultrasound of the bilateral ankles due to long-standing, multi-lobulated subcutaneous swelling. The swelling had

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been present for the past 20 years without significant changes in size. The patient did not complain of restricted joint movement or other symptoms commonly associated with tendon pathologies.

Clinical Presentation: Upon physical examination, the patient presented with multilobulated subcutaneous nodules on both sides of the Achilles tendon without obvious signs of inflammation. There was no complaint of restricted joint movement, and the patient had not been on any medications for this condition.

Laboratory Investigations: Laboratory tests revealed that the patient's cholesterol levels were significantly elevated, which is a common finding in patients with tendinous xanthomas.

Imaging Studies

X-ray Imaging of Lower Leg: The X-ray images of the bilateral lower legs demonstrated no evidence of fractures or dislocations. However, some mild soft tissue changes were visible, which prompted further imaging studies for a more detailed evaluation of the Achilles tendon.

Ultrasound Imaging: High-frequency 12 MHz transducer ultrasound imaging of the bilateral ankles showed a bilateral enlargement of the Achilles tendons, consistent with a xanthomatous process.The imaging revealed homogeneous, lobulated soft tissue masses without adjacent bone erosion or sclerosis. There were no signs of internal calcification or involvement of underlying bones.The fibers of the Achilles tendon appeared normal with no visible discontinuity or major pathology.On T1 and T2-weighted MRI images, the masses displayed heterogeneous signal intensity, indicative of the presence of the xanthomatous tissue infiltrating the tendon.

Diagnosis

The primary diagnosis for this patient is Achilles tendon xanthoma, a rare condition characterized by the deposition of lipid-laden macrophages and giant cells within the Achilles tendon. The presence of multilobulated subcutaneous swelling, along with elevated cholesterol levels, strongly points to this diagnosis. However, several conditions must be considered in the differential diagnosis, including tendinitis, which can present with similar symptoms of swelling and pain in the tendon, but typically without the characteristic lipid deposits seen in xanthomas. Chronic tendon degeneration is another possibility, as it can also result in tendon thickening and swelling, though it generally lacks the specific imaging findings associated with xanthomas. Finally, tophaceous gout should be considered, as it can cause similar nodular masses in the tendon, but it is usually accompanied by joint pain and uric acid crystal deposition, which was not observed in this case. The imaging studies, including ultrasound and MRI, helped confirm the diagnosis of Achilles tendon xanthoma, distinguishing it from these other potential conditions.

Figure 1. Clinical Presentation: Achilles Tendon Xanthoma

Figure 2. MRI Imaging of Achilles Tendon Xanthoma

Figure 3. MRI Findings of Achilles Tendon Xanthoma

Figure 4. X-ray Findings of Achilles Tendon Xanthoma

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Discussion

Achilles tendon xanthoma is a rare but significant clinical condition, commonly associated with lipid metabolism disorders, particularly hyperlipidemia and familial hypercholesterolemia (FH). These lesions are characterized by the accumulation of lipid-laden macrophages within the tendon, resulting in the formation of nodular masses. The condition is often detected in patients with elevated cholesterol levels, and in many cases, it is associated with systemic lipid disorders [1, 5]. This case illustrates a typical presentation of Achilles tendon xanthoma, with the patient exhibiting multilobulated subcutaneous swelling and elevated cholesterol levels, which were confirmed through laboratory tests.

The role of imaging in the diagnosis of Achilles tendon xanthoma cannot be overstated. In this case, ultrasound and MRI imaging were crucial in confirming the diagnosis and differentiating the lesion from other common pathologies such as tendinitis, chronic tendon degeneration, and tophaceous gout. Haacke and Parwaresch [6] highlighted that spontaneous rupture of the Achilles tendon can sometimes occur in patients with hyperlipoproteinemia type II, underscoring the potential complications of untreated tendon xanthomas. The use of high-frequency ultrasound imaging in this case revealed homogeneous, lobulated soft tissue masses, which is consistent with a xanthomatous process. These findings are similar to those reported by Dussault et al. [16], who utilized MRI to observe tendon involvement in familial hyperlipidemia, showing the characteristic xanthoma deposits without any signs of tendon rupture or degeneration.

Other studies, such as those by Fahey et al. [9] and Weiss & Goldblum [7], support the notion that xanthomas in tendons, including the Achilles tendon, are often associated with familial hyperbetalipoproteinemia, a condition that leads to significantly elevated cholesterol levels. The deposition of cholesterol crystals in the tendons can cause the formation of xanthomatous nodules, which, if untreated, may lead to functional impairments or even spontaneous tendon rupture [6]. This is in line with the findings from Blankenhorn and Meyers [15], who emphasized the importance of recognizing Achilles tendon xanthomas to avoid complications such as tendon rupture, which can severely impair mobility.

Furthermore, the differential diagnosis for Achilles tendon xanthoma is broad, and conditions such as tendinitis, chronic tendon degeneration, and gouty infiltration must be considered. However, the absence of joint pain, uric acid crystal deposition, and the imaging characteristics of the lesions in this case helped distinguish it from gouty tophi, as described by Bardin and Richette [10]. In cases of gout, as highlighted by Zhang [11], the presence of uric acid crystals and associated joint inflammation are typical features that were absent in this patient, further confirming the diagnosis of Achilles tendon xanthoma.

The patient's long-standing hyperlipidemia and the imaging features observed in this case are consistent with those described by Rodriguez et al. [14], who reported atypical imaging features of bilateral Achilles tendon xanthomatosis. Their study also demonstrated that the imaging characteristics of tendon xanthomas can sometimes be misleading, with certain cases presenting atypically, which underscores the need for careful analysis and awareness when evaluating such lesions.

Additionally, studies such as those by Liem et al. [17] and Bureau and Roederer [18] have shown that MRI and ultrasound are effective tools in identifying the characteristic features of Achilles tendon xanthomas, providing a non-invasive means to assess the extent of tendon involvement and the potential risk of tendon rupture. Their findings further support the importance of advanced imaging in managing patients with these rare lesions.

Conclusion

This case highlights the importance of imaging in diagnosing tendinous xanthomas, which can mimic other tendon pathologies like tendinitis or chronic degeneration. A detailed clinical history and appropriate imaging modalities, including ultrasound and MRI, are essential in confirming the diagnosis. Managing underlying hyperlipidemia is critical to preventing recurrence and ensuring optimal tendon health.

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