Spontaneous rupture of Achilles tendon xanthomas in a case of familial hypercholesterolaemia

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See Online for appendix

A 70-year-old man attended our hospital reporting a 2-week history of swelling and aching of his left ankle. He did not report any recent trauma or excessive physical activity.

The patient had developed angina pectoris aged 40 years and had coronary artery bypass grafting and ten percutaneous coronary interventions; he had peripheral artery disease of the extremities of both his legs, and stenosis of the internal carotid arteries, aortic valve, renal arteries, and subclavian arteries.

30 years earlier, prior to any treatment, initial laboratory investigations had shown a low-density lipoprotein (LDL) cholesterol concentration of 7·21 mmol/L (typical level 1·81–3·62). Later, genetic analysis, using Sanger sequencing, for familial hypercholesterolemia showed the patient had a heterozygous pathogenic variant of the LDL receptor gene: c.2431A>T (p.Lys811Ter). Exploration of the family history showed a pedigree suggestive of autosomal dominant inheritance—consistent with familial hypercholesterolemia (appendix). No abnormalities were found in the APOB or PCSK9 genes. He had been prescribed atorvastatin and ezetimibe daily; he had repeated LDL apheresis over the past 30 years. Notably, before and after apheresis, his LDL cholesterol levels were 4·40 mmol/L and 1·55 mmol/L, respectively. However, his Achilles tendon had still become thickened over time. An x-ray taken 2 years ago, had shown significant thickening of the left Achilles tendon due to xanthomas caused by his severe and long-term hypercholesterolemia (figure).

On examination we found the patient to be well; his vital signs were within normal range. He had a painful, non-erythematous, soft, swollen left ankle, and the xanthomas were oedematous—very different from their previous presentations; a repeated x-ray shows clear enlargement compared with earlier imaging (figure). A Thompson test was weakly positive.

Sagittal T1-weighted MRI showed heterogeneous signal intensities with a uniform stippled appearance in the thickened tendon, which had an irregular dorsal border surrounded by high-intensity fluid (figure). In aggregate, we diagnosed spontaneous rupture of xanthomas in the Achilles tendon.

The patient had an operation to repair the damaged tendon; the surgeons reported some difficulty because of its fragility. However, the postoperative course was good, and the patient was allowed home 30 days afterwards without any noticeable disability.

Familial hypercholesterolemia is a relatively common genetic disorder. The presence of Achilles tendon xanthomas—thickening of the Achilles tendon due to the accumulation of lipid-laden macrophage—should lead to consideration of familial hypercholesterolemia. Although spontaneous rupture of xanthomas in tendons is not frequent, patients with familial hypercholesterolemia should be examined for the presence of xanthomas and monitored for progression or regression with prolonged treatment.

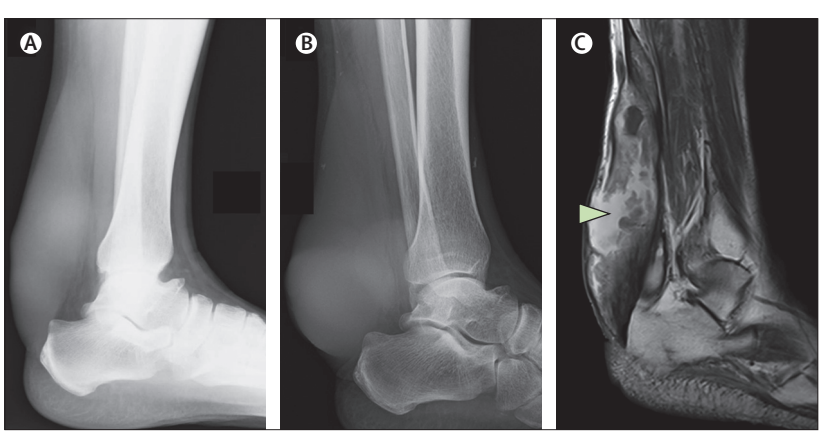

Figure: **Xanthomas in Achilles tendon rupture**
(A) An x-ray of the left ankle—done 2 years before rupture of the Achilles tendon—shows thickening of soft tissue due to xanthomas. (B) An x-ray, repeated at the time of presentation, shows clear enlargement of soft tissue. (C) A T1-weighted MRI shows heterogeneous signal intensities with a uniform stippled appearance in the thickened tendon, which had an irregular dorsal border surrounded by high-intensity fluid (arrow): indicative of partial tendon rupture.

Contributors
KM, MF, MH-S, and SY cared for the patient and collected the data. KM and MF wrote the manuscript and prepared the images. KM and MF reviewed the patient's medical history. SY reviewed the manuscript. Written consent for publication was obtained from the patient.

Declaration of interests
We declare no competing interests.