CASE REPORT

Multiple tendon xanthomas in patient with heterozygous familial hypercholesterolaemia: sonographic and MRI findings

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SUMMARY

Tendon xanthomas are a component of familial hypercholesterolaemia, which is a hereditary disease and characterised by elevated low-density lipoprotein cholesterol plasma levels and premature coronary artery disease. Tendon xanthomas are diagnostic for heterozygous familial hypercholesterolaemia (HFH) and they mostly occur in Achilles tendon. Sonography and MRI are superior to clinical assessment and are useful in detecting tendon xanthomas. In this report, we present ultrasonographic and MRI findings of multiple tendon xanthomas in a case of HFH.

BACKGROUND

Familial hypercholesterolaemia consists of elevated low-density lipoprotein (LDL) cholesterol plasma levels, premature coronary artery disease and tendon xanthomas. The prevalence of heterozygous familial hypercholesterolaemia (HFH) is approximately 1 in 500.¹ Establishing the diagnosis in these patients as early as possible is crucial because the prophylactic treatment prevents the progression of atherosclerosis and coronary artery disease.² Tendon xanthomas are diagnostic findings for HFH and they mostly occur in Achilles tendon. When xanthoma is palpable, there is no need for further radiological evaluation. However, they are not palpable in a group of patients with HFH in whom multiple imaging techniques are necessitated.³ Sonography is an easy, reproducible and effective method for identification of xanthoma and it is superior to clinical assessment, radiography and CT. Also, MRI is a useful technique to determine abnormalities of signal, course and shape of tendons.³

In this report, we presented ultrasonographic and MRI findings of multiple tendon xanthomas in a case of HFH.

CASE PRESENTATION

A 28-year-old woman presented to the orthopaedic clinic with bilateral painful ankle swellings for 3 years. Her physical examination revealed multiple swellings on Achilles tendon regions bilaterally. There was no evidence of synovitis. The patient had been treated for these masses with non-steroidal anti-inflammatory drugs with relief of her pain, but the swelling persisted. Her family history was significant for death from myocardial infarction. In 2004, a coronary angiography was performed for chest pain and showed 60% stenosis of

left anterior descending artery. Her prior laboratory had showed hyperlipidaemia and the last LDL cholesterol and triglyceride values were 598 mg/dL and 150 mg/dL, respectively.

MRI showed a bilaterally thickened Achilles tendon and plantar fascia with focal areas of increased signal intensity on T1-weighted and T2-weighted spin-echo and short tau inversion recovery images (figures 1 and 2). In addition, MRI showed similar findings in the tendons of the left knee (not shown) and bilateral elbow and hands (figure 3). Also, bilateral thickened Achilles tendon and multiple hypoechoic focus in tendons were detected by sonography. Brain MRI was performed to investigate cerebrotendinous xanthomatosis and no abnormality was found. Furthermore, ocular examination showed no abnormality except arcus senilis. Based on the clinical and imaging findings, tendon xanthomas was diagnosed due to familial HFH.

DISCUSSION

Xanthomas define lipomatosis of tendon or tendons related to some genetically transmitted

Figure 1 T1-weighted axial spin-echo images of the right ankle showing the enlargement of Achilles tendon, focal areas with high signal intensity in it and the similar findings in the other tendons (arrows).

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Figure 2 Sagittal short tau inversion recovery images of the left ankle is demonstrating focal areas increased signal intensity in Achilles tendon and plantar fascia (arrows).

diseases commonly seen in types 2 and 3 hyperlipoproteinemias.¹ Tendon xanthoma is one of the components of HFH. Xanthomas could also be associated with non-familial hypercholesterolaemia, drug-induced hypercholesterolaemia and cerebrotendinous xanthomatosis which also include cataracts, neurological dysfunction and atherosclerosis.⁴

Xanthomas are small, mostly palpable lumps located in the skin or subcutaneous tissue that consist of cholesterol, cholesterol esters, triglycerides, phospholipids and numerous lipid-laden foamy macrophages. Previous studies showed that the most frequent site is the Achilles tendon. By the way, other frequent sites include the extensor tendons of the hands and feet.¹ Our patient had multiple xanthomas in knee, elbow and ankle bilaterally.

The demonstration of tendon xanthomas is helpful in diagnosing HFH. However, they are not palpable in up to 20% of individuals with HFH who are more than 20 years old.³ Hence, clinical assessment of these tendons unfortunately lacks sensitivity. Thus, multiple radiological modalities have been investigated as ways to identify these xanthomas such as plain radiography, CT, MRI and sonography.^{5–7} Although they were palpable in our patient, xanthomas were not diagnosed and she was treated with anti-inflammatory drugs. However, the sonography and MRI demonstrated xanthomas and the diagnosis was confirmed.

Sonography is more widely available, less expensive and an easy technique for the identification of xanthomatosis and is superior to clinical assessment. Also, it has been reported to be an effective

method in the investigation of tendon xanthomas.^{1–3} Normal sonographic appearance of the Achilles tendon is characterised by multiple parallel linear echoes that give a fibrillar pattern. Achilles tendon xanthomas have been described either as hypoechoic nodules or as having a diffusely heterogeneous echo pattern at sonography.¹ These features are not specific and partial tendon tears or tendinitis may present a similar appearance and may cause wrong diagnosis.⁷ In our patient, sonography revealed multiple hypoechoic focus in tendons of knee, elbow and ankle.

Other techniques in investigating tendon xanthomas include plain radiography and CT. They could demonstrate the enlargement of Achilles tendon. Unfortunately, detecting internal structural abnormalities before tendon enlargement occurs has not been successful.^{5–6}

MRI of the tendon xanthomas demonstrates morphological and signal intensity abnormalities. When a xanthoma is present, the normally flat or concave margins of tendons may change and may have a convex appearance on axial images. Also, the tendons tend to have higher signal intensity on T1-weighted and T2-weighted spin-echo images than normal tendons do.⁸ Similarly, we have found tendon abnormalities on T1-weighted and T2-weighted spin-echo images. In addition, a speckled or reticulated appearance on axial MRIs, with or without enlargement, is the characteristic feature of xanthomas.³

Sonography was easier and quicker than MRI in detecting Achilles tendon xanthomas, but MRI was shown to be helpful in investigating not only Achilles tendon, but also the other tendons at the same time. Differential diagnosis of tendon xanthoma includes Achilles tendinosis particularly with hypoxic degeneration or some rheumatological conditions characterised by involvement of enthesis points.

Surgical treatment of Achilles tendon xanthomas is rare. It is only indicated when the xanthomas are extremely large, painful and cause problems of mobility. The excision of tumoral nodule or subtotal resection is the opportunity of the surgical treatment. When severe or diffuse infiltration of tendon is involved, the best methods of treatment are total resection and reconstruction of the defect.⁹

In conclusion, demonstration of tendon xanthomas is helpful in the diagnosis of HFH. When xanthoma is clinically apparent, there is no need to perform radiological evaluation. When tendon xanthomas are not easily palpable, radiological investigation is essential to identify tendon abnormalities. Sonography is widely used to diagnose xanthomas and superior to clinical assessment. MRI is another radiological technique that shows tendon abnormalities and depicts xanthomas in individuals with heterozygous familial HFH.

Figure 3 T1-weighted axial spin-echo images of the right hand is showing focal areas increased signal intensity both in extensor and flexor tendons (arrows).

Learning points

- ▶ Xanthoma is mostly the diagnosis of palpable soft tissue swellings occur in tendons often in Achilles.
- ▶ Tendon xanthoma can be a component of a serious inherited clinic syndrome named as heterozygous familial hypercholesterolaemia, which is including high plasma low-density protein cholesterol levels as well as early atherosclerosis.
- ▶ Differential diagnosis of a tendon tumour can be made by sonography and MRI should be helpful in not only depicting the focuses of xanthomas but determining underlying tendon anomalies.
- ▶ Diagnosis of a xanthoma should prevent a serious condition resulting as a heart attack.

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