Case Report

Xanthoma of Tendoachilles: Management and Follow-Up

Nabin Kumar Sahu^a Saurav Narayan Nanda^b Milan Tripathy^c
Saswat Samant^b Sumanyu Kumar Tripathy^b Ashok Kumar Gachhayat^b

^aDepartment of Orthopedics, All India Institute of Medical Sciences, Bhubaneswar, India;

^bDepartment of Orthopedics, Kalinga Institute of Medical Sciences, Bhubaneswar, India;

^cDepartment of Pathology, MKCG Medical College, Berhampur, India

Keywords

Xanthoma · Familial hypercholesterolemia · Tendoachilles · Pseudo-tumor · Flexor hallucis longus tendon augmentation

Abstract

Xanthoma is a rare pseudotumor of connective tissue, consisting of histiocytes with lipid deposits. Usually, it manifests as encapsulated lobular yellowish mass, often associated with familial hypercholesterolemia (FH). We present a case of xanthoma of tendoachilles just proximal to its insertion site, secondary to FH. The patient was a 28-year-old female presenting with a 2-year history of difficulty ambulating and complaints of swelling over the posterior aspect of her left lower leg just proximal to her heel. She had a deranged lipid profile level and was started on a lipid lowering agent for 3 weeks followed by total excision of the lesion with flexor hallucis longus tendon graft augmentation. The patient was able to bear full weight at 6 weeks and could stand on her toes by 4 months. No recurrences were noted at 1 year follow-up.

© 2021 The Author(s).
Published by S. Karger AG, Basel

Introduction

Xanthomas are rare non-neoplastic lesions that are frequently found in the tendons and synovium [1]. They appear as hard and nodular structure which is covered by skin. The common sites are the tendoachilles (TA) and the patella tendon [2]. These are most commonly seen in the 3rd decade of life with female predominance (4:3) as compared with males [3]. Usually, tendinous xanthomatosis is found to be associated with hyperlipidemia. It occurs due to a defect of the LDL receptor and is associated with familial hypercholesterolemia (FH) [4]. Histologically tendinous xanthoma contains multinucleated giant cells, histiocytes, and hemosiderin deposits. Xanthoma of TA may lead to pain, loss of function, and shoe-related problems [5].

Fig. 1. **a** Clinical picture of the swelling arising from the TA just above the insertion site. **b** Preoperative X-ray showing soft tissue shadow in the posterior aspect of distal TA. **c** MRI image of the lesion showing the high signal intensity structure along TA 2 cm proximal to the insertion site suggestive of TX. TA, tendoachilles; TX, tendon xanthoma; MRI, magnetic resonance imaging.

This lesion makes the tendon weak resulting in weakness of plantar flexion. There are no clear-cut guidelines about the management of such cases and most of the cases are managed conservatively in the literature. The graft used for reconstruction purposes was also different in the different study and there is no definite consensus. We present a case of xanthoma of left side TA just proximal to its insertion site, secondary to FH. It was managed with total excision of the diseased segment and reconstruction with autologous flexor hallucis longus (FHL) transfer.

Case

A 28-year-old female presented to the outpatient department with complaints of swelling over posterior and distal aspect of the left leg just above the heel for 2 years and pain for 2 months with difficulty on walking. It was insidious in onset without any history of trauma, gradually progressive and aggravated on walking and relieved on rest.

On examination, a firm swelling of size $6 \times 3 \times 2$ cm on the TA tendon was present 2 cm proximal to the insertion site (Fig. 1a). It was globular in shape, the skin over the swelling was smooth with the normal surrounding skin. There was no local rise of temperature, the swelling was non-tender, non-reducible and non-translucent, and non-adherent to skin. The lesion was in the tendon and was moving along the movement of the tendon. In the left ankle, terminal dorsiflexion was painful and restricted by 10° in comparison to the opposite side (30°). The patient had an antalgic gait. There was no motor weakness or sensory loss of the bilateral leg.

X-ray of heel showed soft tissue shadow without the involvement of bone (Fig. 1b). Ultrasonography of the swelling showed a homogenous hypoechoic lesion inside the TA. The magnetic resonance imaging of the left ankle showed swelling (hyperintensity structure) of $6 \times 3 \times 2$ cm, arising from TA (Fig. 1c) suggestive of tendon xanthoma (TX). After clinical and radiological examination, the provisional diagnosis was made as TX with differential diagnosis as cholesterol accumulation, tendonitis, peritendinitis or bursitis, trauma, nodules from rheumatic arthritis, or gout tophi. The patient was advised for general medicine consultation for further investigation and medical management because of TX. Her Total cholesterol level was 361 mg/dL, and the total LDL-C level was 277 mg/dL as per the laboratory report. Her mother was having hyperlipidemia and under treatment for the same. She was diagnosed as

Fig. 2. **a** Microscopic section showing fibrocartilaginous tissue infiltrated by foamy histiocytes areas of cholesterol cleft surrounded by foreign body giant cells. **b** Immediate postoperative X-ray showing immobilization of ankle in plantar flexion with dorsal slab.

heterozygous FH and she was put on atorvastatin 20 mg once daily as suggested by the concern physician.

Her LDL-C level was 198 mg/dL after 3 weeks of atorvastatin therapy. There was no reduction in the size of the swelling after 3 weeks. There was a pain due to the mechanical block on the dorsiflexion of the foot and the patient was uncomfortable during walking. The swelling was intervened surgically due to persistent pain and gait disturbance. Intraoperatively, full-thickness involvement was found and the total excision of the affected part was done through the posterior approach and sent for histopathological examination. In the neutral position of the ankle, a gap of about 6 cm was created due to excision of the affected part and that was reconstructed by harvesting short FHL graft and transfixing with bioabsorbable interference screw through the calcaneum (Fig. 2b). Histopathological examination confirms our diagnosis of TX (Fig. 2a). Macroscopically, it showed cut section which was firm, solid, and greyish-yellow in color. On the microscopic section, fibrocartilaginous tissue infiltrated by foamy histiocytes was seen. There were large areas of cholesterol cleft surrounded by foreign body giant cells.

A below-knee splint was applied dorsally in plantar flexion position for 2 weeks. The dressing was done on the 2nd and 5th day, which was clean and healthy, and the suture was removed on day 14th. A boot cast in plantar flexion was applied for the next 2 weeks followed by in neutral position for 2 weeks with partial weight mobilization with a walker. The patient was allowed for full weight-bearing mobilization after 6 weeks of surgery. The patient was able to stand on her toe at 4 months. The patient was followed up every month and at the end of 1 year, there was no recurrence.

Discussion

Xanthoma is defined as pseudotumors of connective tissue, consisting of histiocytes with lipid deposits, out of which lipid constitutes around 33% of dry weight and collagen constitutes 24% of dry weight [6]. These tumors manifest as encapsulated lobular masses which is yellow in appearance. Most often, TX associated with FH [7]. The size of the lesion increases with age [8]. As per existing literature, among whites, 1/500 are heterozygous for FH and

1/1,000,000 are homozygous [9, 10]; however, in most countries, a larger population is not even diagnosed [11]. The extent of underdiagnosis and undertreatment of individuals in the general population with FH is largely unknown, and there was no uniform registry maintained in each region [12].

Plain X-ray is usually the first imaging modality showing the soft tissue component (thickened TA with or without associated calcification) [13, 14]. CT scan can be useful to measure the thickness of the TA [15–17]. Ultrasonography is the investigation of choice, and it can be able to detect focal hypoechoic areas of lipid deposit inside the normal hyperechogenic tendon along with the measurement of tendon diameter [18–20]. Magnetic resonance imaging scans can be useful in delineating collagen fiber (low signal intensity structure) from infiltrating foam cells and inflammatory cells (high signal intensity structure) [21].

The initial modality of treatment for TX is usually with hypolipidemic medications (statins) which may help in the regression of the disease [22–30]. A newer drug-like Ezetimibe with a diet low in sterol content is also showing the promising result. Estrogen therapy has also shown to have some impact on the decrease of the tendon size [31]. In advanced cases, surgical resection is required when the patient presented with mass with pain and gait disturbance [32]. The rate of recurrence is 12–15% if partial resection is done [32]. Total resection of the tendon from musculotendinous junction and calcaneum is recommended to decrease the recurrence [33]. Use of various tendons to supplement the gap after excision has been advised by various authors. The tensor fascia lata, FHL, gracilis, and semitendinosus are the usual options for reconstruction [34]. Dinçel et al. [35] recommend the use of quadriceps tendon graft with bony fragments when the defect is large. FHL transfer was described by Pearce et al. [36]. Reconstruction with peroneus brevis was proposed by Gallant et al. [37]. Scagenelli et al. [38] have found the reconstruction by using allograft satisfactory in their study. Dincel et al. [35] have shown good results in bilateral xanthoma excision and reconstruction done by quadriceps tendon with bone fragment in 1 side and other side reconstruction with FHL transfer in different stages. FHL tendon has definite advantages in the Achilles tendon reconstruction than other existing options as (1) it is close to the Achilles tendon; easy to harvest, (2) the FHL muscle has the same function as the triceps surae; same excursion, (3) it has adequate strength; hypertrophy of FHL occurs to bear the strength of TA, and (4) it has the same axis of moving with the Achilles tendon [39]. There is a 2–8% chance of re-rupture of Achilles tendon treated surgically as a complication [40–43].

In this case, we have done total excision of xanthoma, and reconstruction was done with FHL transfer at 1 stage. There was no recurrence till 1 year of follow-up period with very good functional outcomes. She was able to stand, walk independently at a 6-week postoperative period. Power was adequate, and she was able to stand on her toes at 4 months after the surgery. We believe the total excision of the tumor mass with reconstruction with FHL autograft is a valid option with a low recurrence rate and good outcome.

Conclusion

Xanthoma is a rare pseudotumors of connective tissue, consisting of histiocytes with lipid deposits. Most often, TX associated with FH, which can be established with clinical examination, biochemical tests, and radiological imaging. Although statins are the drug of choice in such conditions, in the advanced stage, when it is painful and associated with gait disturbance, surgical intervention is needed. The total excision of the lesion followed by augmentation with FHL tendon graft is a valid option with good functional outcome.

Statement of Ethics

Consent was taken from the patient at the time of surgery. The patient gave her written informed consent to publish their case (including publication of images). The study protocols have been approved by the committee on human research of Kalinga Institute of Medical Sciences, Bhubaneswar, Odisha, India.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

Funding Sources

The authors did not receive any funding.

Author Contributions

S.N.N., S.S., and S.K.T. were the operating surgeons. Data were collected by S.S. and A.K.G. N.K.S. and S.N.N. wrote up the paper. M.T. did the histopathological study for diagnosis. All authors read the manuscript and agreed on the content.

References

- 1 Brodsky JW, Beischer AD, Anat D, East C, Soltero E, Tint GS, et al. Cerebrotendinous xanthomatosis: a rare cause of bilateral Achilles tendon swelling and ataxia. A case report. *J Bone Joint Surg Am*. 2006;88:1340–4.
- 2 Bude RO, Adler RS, Bassett DR. Diagnosis of Achilles tendon xanthoma in patients with heterozygous familial hypercholesterolemia: MR vs sonography. *AJR Am J Roentgenol*. 1994;162:913–7.
- 3 Fahey JJ, Stark HH, Donovan WF, Drennan DB. Xanthoma of the Achilles endon: seven cases with familial hyperbetalipoproteinemia. *J Bone Joint Surg Am*. 1973;55:1197–211.
- 4 Yamakawa K, Yanagi H, Saku K, Sasaki J, Okafuji T, Shimakura Y, et al. Family studies of the LDL receptor gene of relatively severe hereditary hypercholesterolemia associated with Achilles tendon xanthomas. *Human Genetics*. 1991;86:445–9.
- 5 Carranza-Bencano A, Fernández-Centeno M, Leal-Cerro A, Duque-Jimeno V, Gomez-Arroyo JA, Zurita-Gutierrez M. Xanthomas of the Achilles tendon: report of a bilateral case and review of the literature. *Foot Ankle Int*. 1999;20:314–6.
- 6 Tall AR, Small DM, Lees RS. Interaction of collagen with the lipids of tendon xanthomata. *J Clin Invest*. 1978;62:836–46.
- 7 Bhattacharyya AK, Preacher AB, Connor WE. Ectopic xanthomas in familial (type II) hypercholesterolemia. *Atherosclerosis*. 1980;37:319–23.
- 8 Myant NB, Slack J. Type II hyperlipoproteinaemia. *Clin Endocrinol Metab*. 1973;2:81–109.
- 9 Goldstein JK, Hobbs HH, Brown MS. Familial hypercholesterolemia. In: Scriver CR, Beaudet AL, Sly WS, Valle D, editors. *The metabolic & molecular bases of inherited disease*. 8th ed. New York: McGraw-Hill; 2001. p. 2863–913.
- 10 Austin MA, Hutter CM, Zimmern RL, Humphries SE. Genetic causes of monogenic heterozygous familial hypercholesterolemia: a HuGE prevalence review. *Am J Epidemiol*. 2004;160:407–20.
- 11 Marks D, Thorogood M, Neil HA, Humphries SE. A review on the diagnosis, natural history, and treatment of familial hypercholesterolaemia. *Atherosclerosis*. 2003;168:1–14.
- 12 Nordestgaard BG, Chapman MJ, Humphries SE, Ginsberg HN, Masana L, Descamps OS, et al. Familial hypercholesterolaemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease: consensus statement of the European atherosclerosis society. *Eur Heart J*. 2013;34:3478–90.
- 13 Blomqvist G. Xanthoma of the tendo achilles. *Acta Radiol*. 1962;57:45–8.

- 14 Blankenhorn DH, Meyers HI. Radiographic determination of Achilles tendon xanthoma size. *Metabolism*. 1969;18:882–6.
- 15 Steinmetz A, Schmitt W, Schuler P, Kleinsorge F, Schneider J, Kaffarnik H. Ultrasonography of achilles tendons in primary hypercholesterolemia. Comparison with computed tomography. *Atherosclerosis*. 1988; 74:231–9.
- 16 Kolovou G, Daskalova D, Mastorakou I, Anagnostopoulou K, Cokkinos DV. Regression of Achilles tendon xanthomas evaluated by CT scan after hypolipidemic treatment with simvastatin. A case report. *Angiology*. 2004;55:335–9.
- 17 Durrington PN, Adams JE, Beastall MD. The assessment of Achilles tendon size in primary hypercholesterol-aemia by computed tomography. *Atherosclerosis*. 1982 Dec;45(3):345–58.
- 18 Bude RO, Adler RS, Bassett DR. Diagnosis of Achilles tendon xanthoma in patients with heterozygous familial hypercholesterolemia: MR vs sonography. *Am J Roentgenol*. 1994;162:913–7.
- 19 Tuthill TA, Rubin JM, Fowlkes JB. Frequency analysis of echo texture in tendon. *Ultrasound Med Biol*. 1999;25: 959–68.
- 20 Ebeling T, Farin P, Pyorala K. Ultrasonography in the detection of Achilles tendon xanthomata in heterozygous familial hypercholesterolemia. *Atherosclerosis*. 1992;97:217–28.
- 21 Dussault RG, Kaplan PA, Roederer G. MR imaging of Achilles tendon in patients with familial hyperlipidemia: comparison with plain films, physical examination, and patients with traumatic tendon lesions. *Am J Roentgenol*. 1995;164:403–7.
- 22 Regnstrom J, Walldius G, Nilsson S, Elinder LS, Johansson J, Molgaard J, et al. The effect of probucol on low density lipoprotein oxidation and femoral atherosclerosis. *Atherosclerosis*. 1996;125:217–29.
- 23 Kajinami K, Nishitsuji M, Takeda Y, Shimizu M, Koizumi J, Mabuchi H. Long-term probucol treatment results in regression of xanthomas, but in progression of coronary atherosclerosis in a heterozygous patient with familial hypercholesterolemia. *Atherosclerosis*. 1996;120:181–7.
- 24 Yamamoto A, Matsuzawa Y, Kishino B. Effects of probucol on homozygous cases of familial hypercholesterolemia. *Atherosclerosis*. 1983;48:157–66.
- 25 Nakamura T, Ueyama Y, Funahashi T, Yamashita S, Takemura KK, Kubo M, et al. Non-macrophage-related accumulation of cholesterol during probucol treatment in familial hypercholesterolemia: report of two cases. *Atherosclerosis*. 1992;92:193–202.
- 26 Franceschini G, Sirtori M, Vaccarino V, Gianfranceschi G, Rezzonico L, Chiesa G, et al. Mechanisms of HDL reduction after probucol. Changes in HDL subfractions and increased reverse cholesteryl ester transfer. *Arteriosclerosis*. 1989;9:462–9.
- 27 Rouffy J, Chanu B, Bakir R, Goy-Loefer J, Djian F, Monsuez JJ, et al. Changes in lipid and lipoprotein levels and Achilles tendon diameters and indices in familial hypercholesterolaemic patients with tendinous xanthomatosis treated by diet and bezafibrate for 2 years. *Curr Med Res Opin*. 1988;11:123–32.
- 28 Muller DP, Pavlou C, Whitelaw AG, McLintock D. The effect of pregnancy and two different contraceptive pills on serum lipids and lipoproteins in a woman with a type III hyperlipoproteinaemia pattern. *Br J Obstet Gynaecol*. 1978;85:127–33.
- 29 Illingworth DR, Cope R, Bacon SP. Regression of tendon xanthomas in patients with familial hypercholesterolemia treated with lovastatin. *South Med J*. 1990;83:1053–7.
- 30 Lind S, Olsson AG, Eriksson M, Rudling M, Eggertsen G, Angelin B. Autosomal recessive hypercholesterolaemia: normalization of plasma LDL cholesterol by ezetimibe in combination with statin treatment. *J Intern Med*. 2004;256:406–12.
- 31 Mabuchi H, Sakai Y, Watanabe A, Haba T, Koizumi J, Takeda R. Normalization of low-density lipoprotein levels and disappearance of xanthomas during pregnancy in a woman with heterozygous familial hypercholesterolemia. *Metabolism*. 1985;34:309–15.
- 32 Friedman MS. Xanthoma of the Achilles tendon. *J Bone Joint Surg Am*. 1947;29:760–6.
- 33 Tomita T, Ochi T, Fushimi H, Matsuzawa Y, Ono K. Reconstruction of the Achilles tendon for xanthoma: findings at operative re-exploration. A case report. *J Bone Joint Surg Am*. 1994;76:444–7.
- 34 Boopalan PR, Jepegnanam TS, Titus VT, Prasad SY, Chittaranjan SB. Open infected Achilles tendon injury: reconstruction of tendon with fascia lata graft and soft tissue cover with a reverse flow sural flap. *Foot Ankle Surg*. 2008;14:96–9.
- 35 Dinçel YM, Arıkan Y, Özer D, Basılgan S. Reconstruction of the Achilles tendon using quadriceps tendon graft in bilateral xanthomas secondary to familial hypercholesterolemia: a case report. *Eklem Hastalik Cerrahisi*. 2018;29(2):117–22.
- 36 Pearce CJ, Sexton S, Gerrard D, Hatrick A, Solan M. Successful treatment of a chronically infected and necrotic tendo Achillis in a diabetic with excision, flexor hallucis longus transfer and split-skin grafting. *J Bone Joint Surg Br*. 2008;90:186–8.
- 37 Gallant GG, Massie C, Turco VJ. Assessment of eversion and plantar flexion strength after repair of Achilles tendon rupture using peroneus brevis tendon transfer. *Am J Orthop*. 1995;24:257–61.
- 38 Scagnelli R, Bianco G, Imarisio D. Cadaver bone-tendon graft for xanthomatosis of the tendo Achillis. *J Bone Joint Surg Br*. 2009;91:968–71.
- 39 Rahm S, Spross C, Gerber F, Farshad M, Buck FM, Espinosa N. Operative treatment of chronic irreparable achilles tendon ruptures with large exor hallucis longus tendon transfers. *Foot Ankle Int*. 2013 Aug;34(8): 1100–10.

- 40 Chan JY, Elliott AJ, Ellis SJ. Reconstruction of Achilles rerupture with peroneus longus tendon transfer. [Foot Ankle Int](#). 2013;34(6):898–903.
- 41 García-Germán D, Rubio-Quevedo R, Lopez-Goenaga J, Martin-Guinea J. Achilles tendon recurrent rupture following surgical repair: report on two cases. [Foot Ankle Surg](#). 2009;15(3):152–4.
- 42 Pajala A, Kangas J, Ohtonen P, Leppilahti J. Rerupture and deep infection following treatment of total Achilles tendon rupture. [J Bone Joint Surg Am](#). 2002;84(11):2016–21.
- 43 Rettig AC. Potential risk of rerupture in primary Achilles tendon repair in athletes younger than 30 years of age. [Am J Sports Med](#). 2005;33(1):119–23.