In this case report, we present a patient with a giant xanthoma along the entirety of the right Achilles tendon. We discuss the pathogenesis, clinical manifestations, complications and unique surgical treatment of this rare condition. Familial hypercholesterolemia is a genetic disease that provides the best evidence for the causal role of low-density lipoprotein cholesterol in human atherosclerosis. The disease was first described by Müller in 1939 and is characterized by high cholesterol levels from birth, and the subsequent development of tendon and cutaneous xanthomas and premature atherosclerosis. Achilles tendon xanthomas are not palpable in up to 20% of those with heterozygous familial hyperlipoproteinemia.

Pathologic evaluation and analysis of the tendon specimen revealed giant tendon xanthomas caused by familial hypercholesterolemia. Within 12 weeks, patient transitioned from a posterior splint to a CAM-walker with limited weight bearing, followed by normal weight bearing to tolerance in a shoe. The patient has been walking normally for 1 month and is satisfied with outcome of the surgery. He now wants to schedule the same procedure on the contralateral limb where he has similar findings.

In this case report, we described a 44-year-old male patient with xanthomatous findings within the entire Achilles tendon. His diagnosis was made following traumatic rupture of the right Achilles tendon. The initial repair included debridement of the damaged tendon with specimen sent for pathologic analysis. His first surgical repair attempt was an end-to-end anastomosis with FHL augmentation. A standard postoperative course of non-weight bearing in a posterior splint for 6 weeks followed by partial weight bearing in a CAM-walker boot was prescribed. His recovery was unremarkable as to strength and range of motion, except that the patient continued to have pain along the entirety of the Achilles tendon long after normal healing should have taken place. The patient underwent months of physical therapy with unremitting pain. MRI imaging revealed the entire right Achilles tendon had been replaced with giant xanthoma. The patient was scheduled for surgical resection and replacement of the defective tendon with a myofascial turndown technique.


Conclusions

Xanthomas along the Achilles tendon are not a commonly reported finding and are not well recognized clinically. Most cases are discovered with advanced imaging such as MRI or contrast enhanced CT and during surgical repair with specimen sent for pathologic analysis. Awareness of this condition and treatment strategy will allow clinicians and surgeons to treat affected patients more appropriately for faster recovery and potentially effect greater functional outcomes.