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Case Report

Radiology Section

Bilateral Spontaneous Rupture of the Achilles Tendon as an Initial Presentation of Alkaptonuria: A Case Report

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ABSTRACT

Rupture of the Achilles tendon is a common occurrence in the athletic population with a history of injury; however, a complete rupture of both Achilles tendons is uncommon. Spontaneous rupture of the bilateral Achilles tendons is an even rarer phenomenon, usually highlighting an underlying pathological condition. The authors present a 32-year-old healthy male patient with no previous known co-morbidities and radiological evidence of a complete tear of both Achilles tendons. Histopathological examination of resected tendon tissue revealed dark-coloured deposits within, and further investigations established the diagnosis of alkaptonuria. Non traumatic bilateral rupture of the Achilles tendon should raise suspicion of underlying metabolic/genetic disorders like alkaptonuria.

Keywords: Ankle joint, Grading of tendon ruptures, Phenylalanine catabolism

CASE REPORT

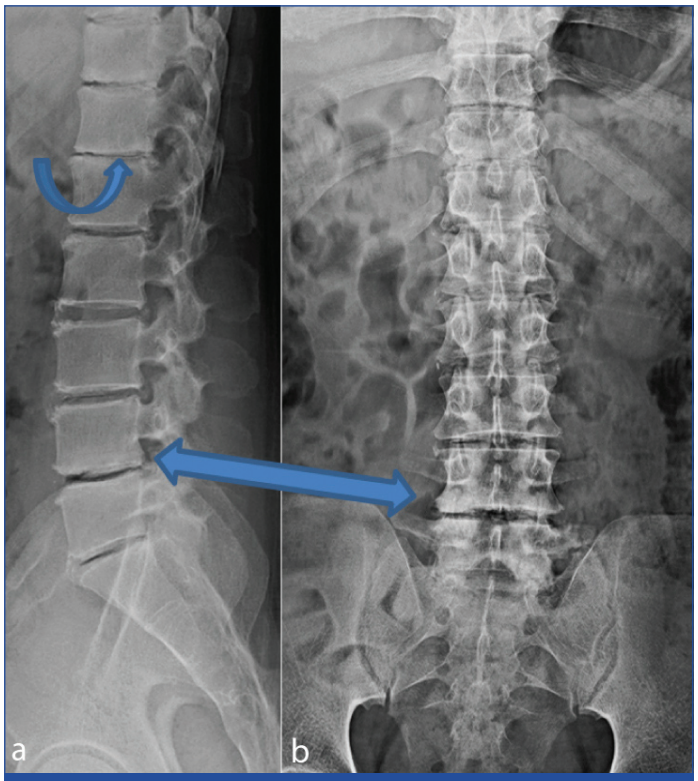
A 32-year-old healthy male patient presented to the Orthopaedic Department with a history of pain in both ankle joints, inability to bear weight in both legs, and limited movement of ankle joints for a two-month duration. He had no previous known co-morbidities. He denied a history of injury or previous intake of steroid/fluoroquinolone medication.

He developed pain and swelling around the ankle joints after climbing stairs. The patient's mother mentioned that the patient also had a past history of passing dark-coloured urine and dark staining of diapers occasionally. The patient had no family history of proven Alkaptonuria among family members.

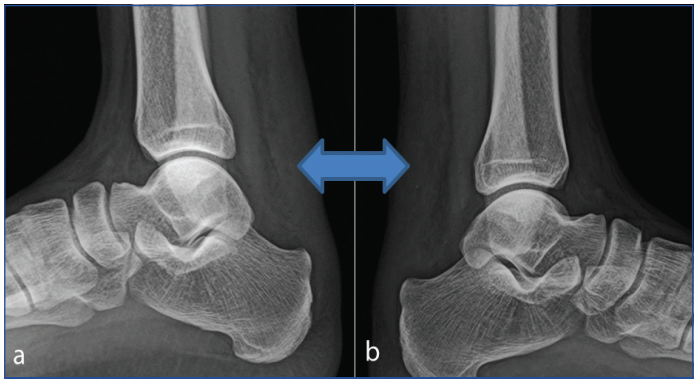
Physical examination showed no evidence of pigmentation in the eyes, ears, or skin. Biochemical examination revealed an increased level of Homogentisic Acid (HGA) in the blood and urine.

Physical examination also revealed no swelling or discharging sinuses in the ankle region. The Thomson test was positive on both sides. Plantar flexion was absent, and the patient was unable to do a heel raise on both sides. Radiographs of the spine also revealed a multilevel vacuum phenomenon, reduced intervertebral disc height, and intervertebral disc calcification [Table/Fig-1a,b]. Radiographs of both ankle joints showed no bony or joint injury [Table/Fig-2a,b]. Magnetic Resonance Imaging (MRI) of both ankle joints revealed a complete tear of the Achilles tendon, with retraction of torn fibres for a length of 3.5 cm on the right-side and 4.7 cm on the left-side [Table/Fig-3a,b]. The rupture occurred at the mid-tendinous portion on both sides and also involved the bilateral calcaneal attachment site. Significant oedema was also noted within the tendon and the ruptured site [Table/Fig-3a,b]. The calcaneum and other tarsal bones showed normal marrow signal intensity. The lateral and medial collateral ligaments of the ankle joint appeared unremarkable. After preliminary investigations, the patient was scheduled for surgical repair and underwent tendinoachilles repair with gastrocnemius lengthening on the right-side [Table/Fig-4a,b] and flexor hallucis longus transfer on the left-side [Table/Fig-5a,b]. Part of the torn tendon fibres were sent for histopathological examination, which revealed Ochronosis with foreign body-type giant cell reaction and degenerative changes in adjacent fibrocollagenous and cartilaginous tissues.

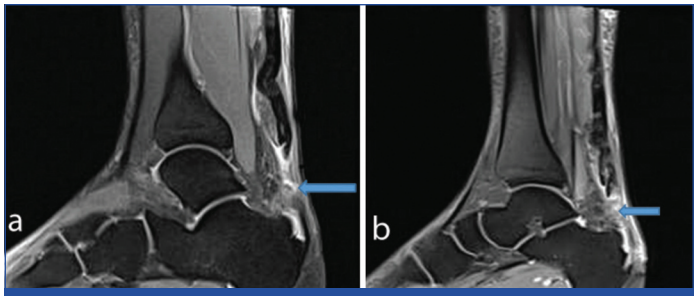
The postoperative period was uneventful, and the patient was discharged. The patient was advised to have regular follow-up.



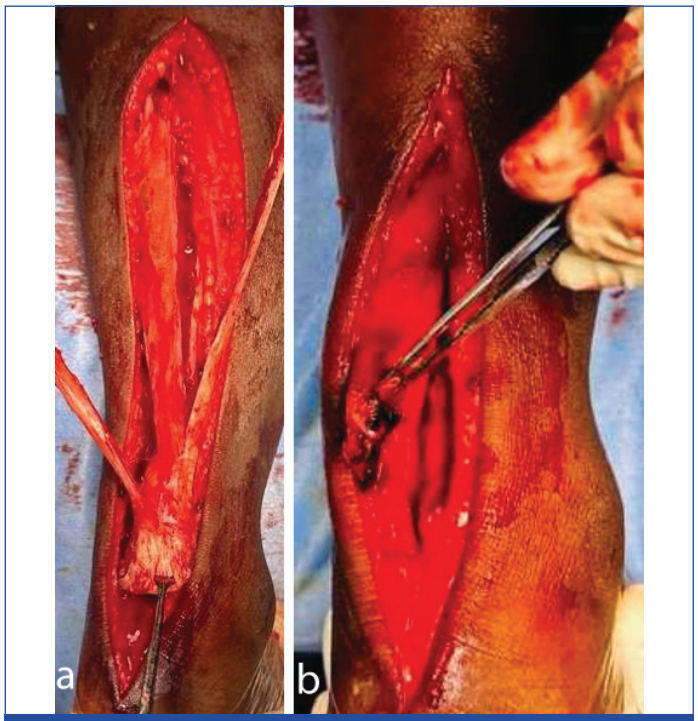
[Table/Fig-1a,b]: Lumbar spine shows multilevel intervertebral disc height reduction, vacuum phenomenon (bold arrow) and disc calcification (curved arrow).



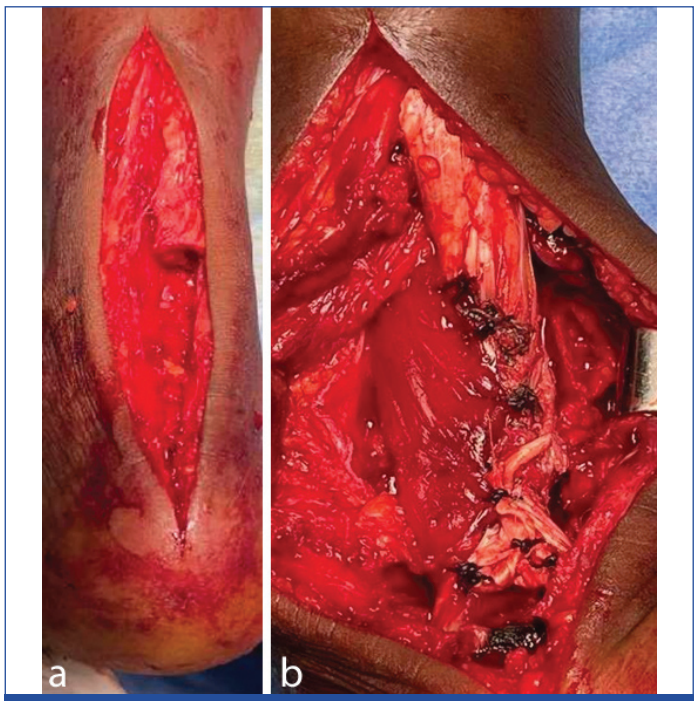
[Table/Fig-2a,b]: (Right ankle); 2b (Left ankle). Radiograph of both ankle joint lateral views showing no evidence of fracture/bony injury. Soft-tissue thickening noted in the Achilles tendon region on both sides (bold arrow).



[Table/Fig-3]: a) (right leg); b) (left leg) Sag Proton-density Fat-suppressed (PDFS) images showing complete rupture (Bold arrow) of Achilles tendon with retraction of torn proximal fibers on both sides.



[Table/Fig-4]: Showing a) ruptured and b) repaired; Achilles tendon on right-side.



[Table/Fig-5a,b]: Showing a) traumatised and b) repaired Achilles tendon on left-side.

DISCUSSION

Alkaptonuria or Ochronosis is one of the rare inborn errors of metabolism, an autosomal recessive disorder, with an incidence of 1 in 250,000 to 1 in 1,000,000 in most populations [1]. Most patients appear asymptomatic during childhood and until the fourth decade of life. A few may notice a blackish discolouration of urine after exposure to air and black discolouration of a diaper, which is the earliest manifestation of this disease and is rarely reported by patients [1,2]. Most of these patients remain asymptomatic until adulthood. Alkaptonuria or Ochronosis is due to a mutation on Chromosome 3q21-q23. This condition is characterised by the accumulation of HGA due to deficient Homogentisate 1,2-Dioxygenase (HGD), an enzyme that converts homogentisate to 4-maleylacetoacetate and is involved in the catabolism of phenylalanine and tyrosine amino acids. Excessive HGA is oxidised to benzoquinones, which polymerise and form a dark pigment [2-5]. An increased concentration leads to the deposition of Homogenistic acid in body parts like the sclera, skin, and earlobe, causing pigmentations of the earlobe, sclera, and teeth. Deposition of HGA in the kidney and prostate can cause renal and prostatic calculi [6]. The excretion of HGA through urine causes the dark discolouration of urine and dark staining of a diaper in the case of neonates. Deposition into the articular cartilage, ligaments, and tendons leads to progressive arthropathy and weakening of tendons, which further leads to deformity and rupture, respectively [7]. Among the tendons, involvement of the Achilles tendon, patellar tendon, and quadriceps tendon have been reported in the literature [8].

In the present case, the patient was generally asymptomatic, although he had a few episodes of dark discolouration of urine and dark staining of diapers, and was not diagnosed until the incident of bilateral rupture of the Achilles tendons.

Mwafi N et al., reported a 42-year-old Jordanian male patient with a history of spontaneous unilateral rupture of the Achilles tendon. This patient had a family history of Alkaptonuria, as his parents were related (Consanguineous marriage), and his brother and sister were also diagnosed with Alkaptonuria [9].

AlShenow A et al., reported a 52-year-old female, a full-blown known case of Alkaptonuria, who presented with unilateral non traumatic rupture of the Achilles tendon. This patient also had a chronic history of back pain, pigmentation in the skin, eye, and earlobe. The patient underwent surgical repair of the ruptured Achilles tendon [10].

There is no definite treatment for Alkaptonuria. Dietary restrictions like lower protein intake, particularly tyrosine and phenylalanine, are recommended. Some physicians advise intake of Vitamin C (ascorbic acid) at a dose of 0.5 to 1.0 gm/day, which is believed to prevent the oxidation of HGA. Non steroidal anti inflammatory drugs could be used for symptomatic relief. Glucosamine and chondroitin sulfate supplements are helpful in slowing down the degeneration of joint cartilage [11,12].

A low dose of nitisinone reduces the formation of HGA, thereby reducing the concentration of HGA in the blood and urine and slowing the progressive disease course of Alkaptonuria [13,14]. Increasing awareness and early detection of Alkaptonuria, along with frequent follow-up, could reduce the pace of disease progression and prevent the development of complications.

CONCLUSION(S)

Alkaptonuria is a rare metabolic disorder that remains asymptomatic in the early part of life and presents as multisystem involvement with complications at a later stage. Early diagnosis, increased awareness, and screening among family members will lead to early detection of this condition. Available treatments could delay disease progression and prevent complications.

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