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Pathological Achilles Tendon Rupture in a Patient with Undiagnosed Alkaptonuria

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Abstract
Alkaptonuria (AKU) is a rare autosomal recessive disorder that is caused due to the deficient activity of homogentisate1, 2-dioxygenase which results in increased levels of homogentisic acid (HGA) and its oxidized product benzoquinone acetic acid (BQA), which form polymerized deposits resulting in a bluish-black discoloration of the cartilage as well as degeneration, inflammation and calcification of the tendons, ligaments and large joints and increased bone resorption. Ochronotictendinopathy most commonly affects the Achilles or patellar tendon leading to enthesopathy or spontaneous tendon ruptures. Here, we report a case of a 35-year-old male patient with a nontraumatic achilles tendon rupture without a previous diagnosis of ochronosis.

INTRODUCTION

Alkaptonuria is a rare genetic disorder due to the deficiency of the enzyme homogentisic acid oxidase, an enzyme essential for the catabolism of phenylalanine and tyrosine[1]. This deficiency results in the accumulation of homogentisic acid, which accumulates in a polymerized form in the connective tissue such as the sclera, cartilage, ligaments, tendons, skin, nail, teeth and vessel intima[2].Over the years, these accumulations give rise to a dark pigment, resulting in a condition called Ochronosis. This term describes the yellowish (resembling ochre) staining of tissues when observed under a microscope, even though to the unaided eye, the affected tissues appear bluish grey[3]. The condition presents itself in three distinct stages, with the initial stage occurring from birth, characterized by the darkening of urine. Hence the earliest clinical sign of the disorder is the tendency of the diapers to have black stains. The second stage is between the second and third decade of life, when ochronosis of the ear and sclera are the main symptoms. Final, the third stage involves the presence of large ochronotic deposits leading to biomechanical destruction of the spine, large joints, tendons, heart valves, kidney and prostate[4].Arthropathic changes occur in approximately 30% of the cases by their fourth-fifth decade of life[2]. Rupture of the Achilles tendon is reported in the literature as a rare clinical complication of patients suffering from undiagnosed alkaptonuria.

We present a case involving an adult male with no previous medical conditions. He came to our Orthopaedic clinic after experiencing an Achilles tendon rupture due to a seemingly minor injury. While performing the surgical repair, we discovered substantial ochronosis deposits within the tendon, necessitating the removal of a significant portion of the tendon. Subsequently, the tendon defect was addressed through a suture anchor technique, complemented by a procedure involving the transfer of the flexor halluces longus tendon for augmentation.

Case Presentation: A 35-year-old male, without any prior medical conditions, presented at our orthopaedic outpatient department with a chief complaint of being unable to move the left foot downwards and was experiencing pain in the back of the affected ankle for the past month. He reported a minor injury that occurred one month earlier when he twisted his ankle while descending stairs. Subsequent to this injury, he faced difficulties in both climbing stairs and walking. Upon a local examination of the affected ankle, significant swelling was observed on the posterior aspect, along with a palpable defect in the midsection of the Achilles tendon (Fig. 1). The patient displayed an inability to actively perform plantar flexion of the foot but had full range of motion for all other ankle movements, both active and passive. Additionally, he was unable to execute a single-leg heel raise on the affected side.

When the Simmonds-Thompson test was performed on the left ankle, a positive result was obtained, indicating the absence of plantar flexion of the foot when the calf muscles were squeezed. This finding suggested a discontinuity in the Achilles tendon. Subsequent MRI imaging of the affected ankle revealed a nearly complete tear of the Achilles tendon located at the posterior aspect of the ankle joint, with the proximal end retracting approximately 4cm from the calcaneum. Given the provisional diagnosis of a chronic left Achilles tendon tear dating back one month, the patient was scheduled for Achilles tendon repair involving a suture anchor and a flexor hallucislongus (FHL) tendon augmentation. Notably, during the surgical procedure, the Achilles tendon displayed an unexpected dark blue to blackish discoloration, which extended distally from the musculotendinous junction up to 4cm proximal to the calcaneum insertion site. The discolored segment of the tendon was excised and the tendon was reconstructed using a suture anchor secured to the calcaneum, along with FHL tendon augmentation and a V-Y plasty of the Achilles tendon muscle belly(Fig. 2).

Subsequent to the surgical procedure, the limb was placed in an above-knee plaster slab with the knee in flexion and the ankle in a plantar flexed position. The resected black-pigmented portion of the tendon was sent for histopathological examination. Due to the observed blackish discoloration of the tendon, there arose a suspicion that the patient might be suffering from Alkaptonuria, prompting a reevaluation of the patient's medical condition. Upon reassessment following the procedure, it was discovered that the patient had a childhood history of dark staining of diapers during infancy, though no further investigations had been conducted at that time. Additionally, it was noted that the patient was born as a result of a consanguineous marriage.

Upon examination, it was discovered that the patient had previously unnoticed black deposits on the sclera (Fig. 3). The results of the histopathological and histochemical analysis of the biopsy specimen indicated the presence of Ochronosis in the fibro-collagenous tissue of the Achilles tendon. To confirm the diagnosis of Alkaptonuria, further testing was performed, including blood and 24-hour urine samples for metabolic screening. However, these samples tested negative and no elevated levels of homogentisic acid (HA) were detected. The rehabilitation process followed a standard treatment

plan for Achilles tendon repair, involving the gradual neutralization of a flexed above-knee slab. Three months after the surgery, the patient began partial weight-bearing and had regained near full range of motion in the left ankle, both actively and passively (Fig. 4).

RESULTS AND DISCUSSION

Achilles tendon ruptures are a common occurrence in the field of Orthopaedics, primarily stemming from traumatic events and subsequent re-ruptures, whereas atraumatic spontaneous ruptures are rare[4]. Ruptures of the Achilles tendon is an unusual clinical complication, typically observed in male patients with alkaptonuria. There have been very few reported cases of females diagnosed with Ochronosis of the tendoachillies[1], potentially owing to the fact that Alkaptonuria progresses more rapidly in males than in females. The pathological rupture of tendons due to the deposition of homogentisic acid (HA) in their structure appears to affect larger tendons such as the Quadriceps tendon and the Tendoachillies[5], with the Achilles tendon[6] rupture being the most frequently reported. Alkaptonuria typically remains asymptomatic until adulthood, and its early clinical manifestations often go unnoticed. In childhood, the initial sign is often the darkening of urine, but patients rarely report this symptom[6].

Our patient also had a history of dark diaper staining during infancy, but the doctors who attended to him at that time did not consider the possibility of alkaptonuria and did not conduct further investigations. Pigmentation in the eye sclera and

Fig. 1: Significant swelling on the posterior aspect with a palpable defect in the midsection of achilles tendon

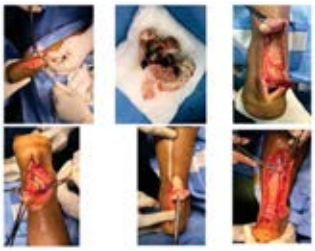

Fig. 2: FHL tendon augmentation and a V-Y plasty of the Achilles tendon muscle belly

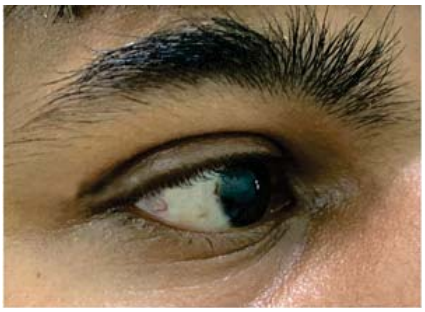

Fig. 3: Unnoticed black deposits on the sclera

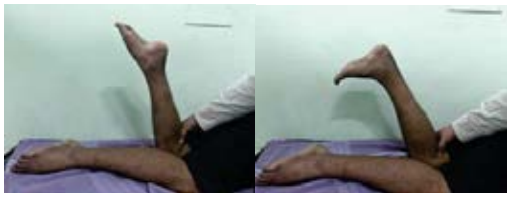

Fig. 4: Three months after the surgery, the patient began partial weight-bearing and had regained near full range of motion in the left ankle, both actively and passively

subcutaneous cartilage, such as the ear, is a common clinical sign typically observed in the second decade of life[2]. However, this sign is less apparent in individuals with dark skin, like our patient and often goes unnoticed[6]. Similar to our case, the majority of Achilles tendon ruptures associated with alkaptonuria are not diagnosed in advance and the suspicion of Ochronosis arises only when there is a dark blue-blackish macroscopic appearance of the tendons during surgery and investigations of the urine[2].

Our patient received a diagnosis of Ochronosis in the Achilles tendon solely based on the biopsy report of the Achilles tendon specimen, as both urine and blood samples tested negative. This aligns with findings in the literature, where the majority of cases involving Achilles tendon rupture associated with Ochronosis are reported to have negative results in blood and urine tests for elevated levels of homogentisic acid (HA). Instead, the diagnosis is primarily reliant on the histopathological examination of the resected specimen, revealing features of Ochronosis[3]. The optimal approach for repairing tendon ruptures with a significant gap in patients with alkaptonuria is not well-documented and remains uncertain. This uncertainty arises from the fact that the torn ends can be irregular due to extensive degeneration, making them less than ideal for suturing. The primary challenge with direct repair lies in the unpredictable gap length that remains after excising the pigmented

portion of the tendon. Various surgical techniques have been employed to address Ochronotic Achilles tendon ruptures, including direct repair, V-Y advancement and tendon transfer after removing the discolored portion of the tendon[1].

Mwafi *et al*. documented a similar case involving an adult male who experienced an Achilles tendon rupture attributed to alkaptonuria. The extensive degeneration of the tendon led to a substantial gap, necessitating the use of a Flexor HallucisLongus (FHL) tendon transfer for the repair. In cases where defects in the Achilles tendon repair are substantial, exceeding 5cm in length, the preferred approach involves the use of an autograft or allograft tendon transfer or reconstruction. The FHL tendon stands out as the most suitable choice due to its strength and considerable length. Furthermore, the FHL muscle shares similar functions with the gastrocnemius and soleus muscles, and it operates along the same axis as the Achilles tendon, ensuring that it does not disrupt the natural muscle balance of the ankle[1]. Alkaptonuria is a systemic condition and some reported cases suggest that multiple sequential ruptures of various tendons due to Ochronosis may occur over time[1]. For instance, Kumar. EGM *et al*. presented a case of an adult male who experienced sequential tendon ruptures, first involving the Achilles tendon and subsequently the Quadriceps tendon, with a one-year gap between the two incidents.The clinical manifestations, characterized by dark pigmentation in the sclera of both eyes and bilateral ear pinna, went unnoticed during the initial presentation. The patient was ultimately diagnosed with Ochronosis based on the examination of tissue samples and urine tests conducted during the most recent repair[6]. MV Nagcelwane *et al.* reported a case in which a patient initially experienced a rupture of the quadriceps tendon, followed by a rupture of the Achilles tendon several months later. Eventually, the patient suffered from tendon ruptures around the knee after a few months. These successive episodes of tendon rupture prompt the question of whether individuals with Alkaptonuria should initiate some form of prophylactic treatment after the initial tendon rupture, much like what is done in Osteoporosis following an initial fragility fracture[4].

Currently, the prophylactic measures recommended by physicians to prevent subsequent tendon ruptures in alkaptonuricochronosis involve adhering to a diet low in protein, particularly foods rich in tyrosine, such as meat, cheese and dairy products[1]. Additionally, a daily oral intake of 1gram of Vitamin C is advised, as the mild antioxidant properties of ascorbic acid aid in preventing the deposition of homogentisic acid (HA) in cartilaginous tissues[2]. Patients can also benefit from physical therapies aimed at maintaining flexibility and strengthening muscles and tendons[1].

CONCLUSION

In cases where there is a spontaneous tear of the Achilles tendon, it's important to consider the possibility of Alkaptonuria as this may serve as the initial indication for many patients that they have this condition. Repairing a substantial gap in an Achilles tendon rupture poses a significant challenge. However, as our case report illustrates, the use of FHL tendon transfer proves to be an effective method for addressing a large defect in the tendon. Following the procedure, the FHL tendon transfer enabled the patient to regain as much strength as possible in ankle plantar flexion. Three months after the procedure, the patient experienced uncomplicated healing and achieved satisfactory functional results.

While the surgical repair of Achilles tendon tears associated with Ochronosis presents challenges for orthopedic practitioners, it's crucial to emphasize the importance of prophylactic therapy for individuals with Alkaptonuria to prevent subsequent tendon ruptures later in life. This necessitates a greater focus on developing disease-modifying treatments, potentially in the form of gene or enzyme replacement therapy, to prevent the initial deposition of homogentisic acid (HA).

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