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A BLACK TENDON MAY NOT BE JUST AN AVULSION: CASE REPORT OF AN OCHRONOTIC ACHILLES TENDON INJURY IN A PATIENT WITH ALKAPTONURIA AND REVIEW OF THE LITERATURE

P. ARRIGONI[1], I. MORELLI[2], M.D. CARDANI[1], D. PRESTAMBURGO[1]

[1]Orthopaedics and Trauma Unit, ASST Ovest Milanese, Legnano Hospital, Legnano (MI), Italy
[2]Orthopaedics and Trauma Unit, ASST Nord Milano, Bassini Hospital, Cinisello Balsamo (MI), Italy

Corresponding Author
Ilaria Morelli, MD; e-mail: ilaria.morelli90@gmail.com

ABSTRACT – *Background*: Alkaptonuria is an autosomal recessive disorder caused by the accumulation of homogentisic acid (HGA) pigmented products in collagen-rich connective tissues: this leads to chronic inflammation and tissue degeneration (ochronotic arthropathy and tendon injuries).

Case Report: A 49-year-old man with a spontaneous partial Achilles tendon avulsion underwent surgery. The unexpected intraoperative findings of a black burn-like tendon stump and calcaneal tuberosity (ochronosis) led us to the diagnosis of alkaptonuria. For this patient, a combined surgical procedure of tenodesis and tendon lengthening resulted in a satisfactory outcome.

Conclusions: Only a few cases of Achilles tendon injury due to alkaptonuria are described in the literature and, to the best of our knowledge, none of them were treated with this combined surgical technique. This surgical technique used in our patient provided a very satisfactory outcome.

KEYWORDS: Alkaptonuria, Ochronosis, Achilles tendon rupture, Achilles tendon avulsion, Case report, Black tendon.

INTRODUCTION

Alkaptonuria is an autosomal recessive metabolic disorder with a prevalence ranging from 1:200,000 to 1:1,000,000 live births caused by the deficiency of the enzyme homogentisate 1,2 dioxygenase (HGD)[1]. In the absence of HGD, the excess of homogentisic acid (HGA), a metabolic product of the amino acids phenylalanine and tyrosine produced by the liver, oxidizes, forming the "ocher-like" pigment polymer. Pigment accumulation in collagen-rich connective tissues (sclera, cartilage, tendons, ligaments, skin, nails, teeth, and vessel intima) leads to a systemic disease called ochronosis. This accumulation stiffens the tissues, leading to chronic inflammation and degeneration of the joints, known as "ochronotic arthropathy" by the fourth decade of life[1,2]. We report the case of a patient with partial Achilles tendon avulsion who was diagnosed with alkaptonuria after surgical treatment.

CASE PRESENTATION

This case report has been written according to CARE guidelines[3]. The patient was informed that data concerning the case would be submitted for publication, and he provided consent.

A 49-year-old man was admitted to our hospital complaining of left ankle pain and inability to bear weight abruptly occurring after light physical activity. He suffered mild atraumatic calcaneus pain during the last three months and had a history of chronic anterior knee and lumbar pain. He was otherwise healthy, with no history of genetic disorders. He smoked five cigarettes/day, practiced moderate physical activity (running two to three times a week), and had a sedentary job. On examination, pain, swelling and a little gap were found just proximal to the Achilles tendon insertion, but he was able to plantarflex the ankle, and the Thompson squeeze test was negative. No bony avulsions were detected on radiographs. The rupture of two-thirds of the Achilles tendon, at 1.5 cm from the calcaneal insertion, was evident on dynamic ultrasound examination.

The choice between conservative and surgical treatment was offered to the patient, who ultimately chose surgery. Routine preoperative blood tests, chest X-rays, and electrocardiograms were within normal limits. Under spinal anesthesia, the Achilles tendon was exposed through a medial approach. Intraoperatively, the lateral third of the tendon was found intact, and the medial two-thirds detached from the calcaneal tuberosity. The tendon stump was almost black with a burn-like appearance, and the calcaneal tuberosity looked grey and poorly vascularized (Figure 1). The distal medial part of the tendon (1.5 cm) was resected and sent for histopathological analysis, and the calcaneal tuberosity was scraped, reaching the healthy bleeding tissue. A Krakow non-absorbable suture was used to reattach the tendon stump to the calcaneal tuberosity through transosseous stitches. Finally, the right tension of the tendon was restored by performing a Z-lengthening of the medial side, based on the intact lateral third, and the stumps were sutured together and to the lateral third using intratendinous absorbable suture (Figure 2).

The intraoperative burn-like appearance of the tendon suggested examining the patient for alkaptonuria. The histopathology report described degenerative tendon changes and rupture compatible with ochronosis. Dark nodules were noted on the ears, and spondyloarthropathy was detected on lumbar radiographs. The patient's urine exposed to light was observed to change color. Genetic counseling was requested, and the clinical diagnosis of alkaptonuria was confirmed by urine chromatography.

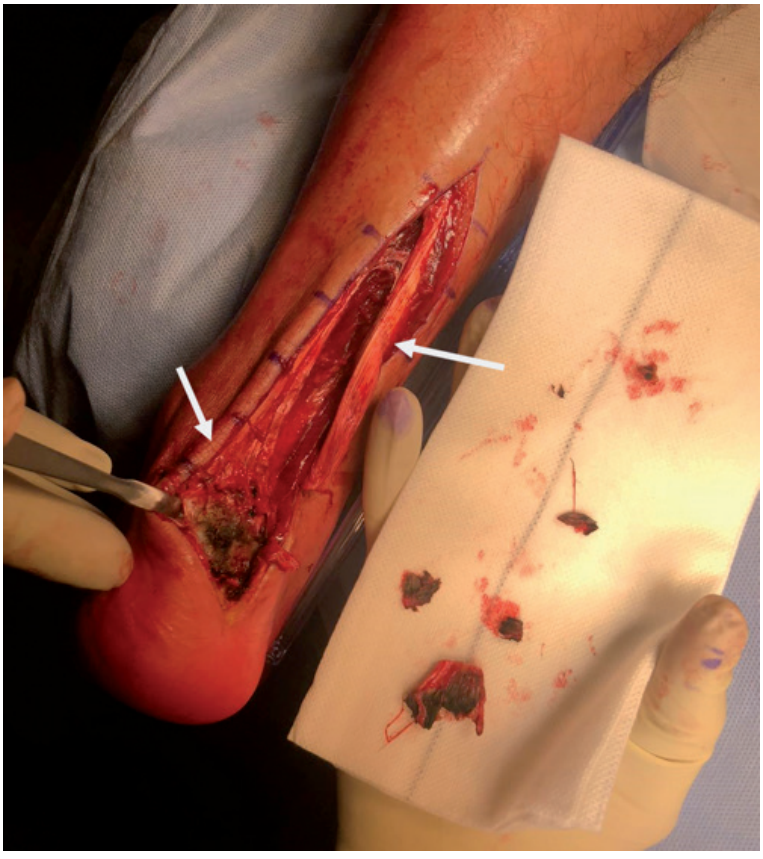

Figure 1. The black appearance of the calcaneal tuberosity and the distal Achilles tendon resected. The Achilles tendon is longitudinally divided, and the lateral third (left arrow) is still attached to the calcaneal tuberosity, while the medial part is avulsed (right arrow).

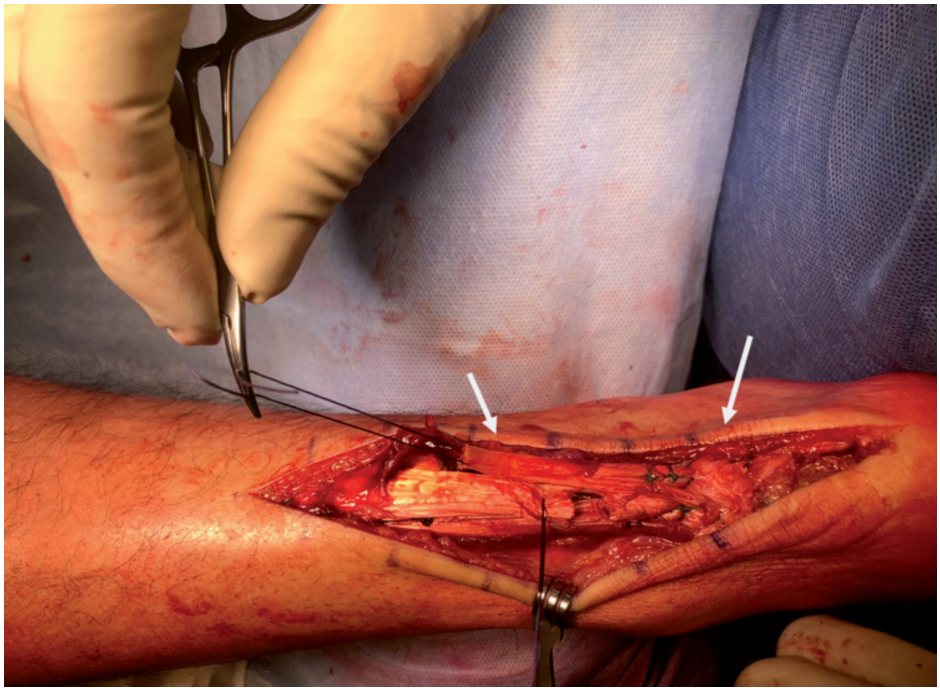

Figure 2. Z-lengthening of the medial part of the Achilles tendon (left arrow) after tenodesis to the calcaneal tuberosity (right arrow).

The ankle was kept in mild plantar flexion with a cast for two weeks and then in a neutral position for another four weeks. Weight-bearing was allowed in a cast one month after surgery. Physical therapy was started six weeks postoperatively, progressing from isometric to concentric and, finally, eccentric strengthening of the gastrocnemius-soleus complex. At four months, the patient was able to perform a symmetrical toe rising, recovering the ankle's full range of motion, and at six months, he was back to moderate physical activity as before the injury.

After nine months, the MRI showed a physiologically thickened healthy tendon (Figure 3).

At the one-year follow-up, the patient had no complaints of pain or activity limitation.

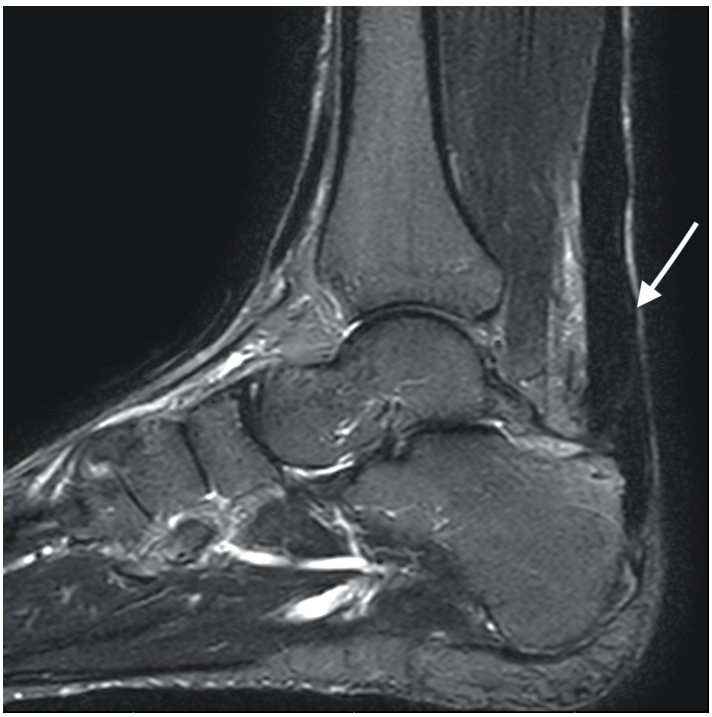

Figure 3. STIR_longTE MRI sequence shows a healthy Achilles tendon nine months after surgery (white arrow).

DISCUSSION

Only a few reports[4-7] describe Achilles tendon rupture or avulsion in alkaptonuria. In this case, the partial avulsion of the tendon from the calcaneal tuberosity has been reported. The literature[4-7] shows similar outcomes between surgical and conservative treatment of Achilles tendon lesions except for a higher risk of re-rupture in the conservative cases. Ultrasound is useful to determine complete *vs.* partial ruptures; MRI is usually requested in equivocal cases and for chronic ruptures[8,9]. In this case, given the partial lesion, a choice of conservative treatment was offered to the patient. However, the young age and the increased re-rupture rate favored a surgical approach.

The most relevant surgical finding was the burn-like, black, avascular aspect of the tendon stump and the calcaneal tuberosity. Because of those findings, the excision of the distal tendon and the calcaneal tuberosity to healthy bleeding tissue was preferred. As the diagnosis was still unknown at the time of surgery, a transosseous suture was preferred to metallic anchors to allow a better acquisition of MRI images in case of need. The Z-lengthening of the reattached tendon was used to restore the right tension based on the intact third of the tendon. A precautionary rehabilitation protocol starting after six weeks of immobilization was preferred because of the surgical technique used and the intraoperative findings, as during surgery, the healing potential of the suspected ochronotic tendon was unpredictable. Nevertheless, the rehabilitative protocol led to a good functional outcome within the expected time. An MRI at the 9th month showed continuity of the tendon without insertional alterations besides the physiological thickening. The resection of the degenerate Achilles tendon segment and the suture of normal, vascularized tissues showed a good healing potential. The patient returned to sport at the pre-injury level after six months: this result is in line with the return-to-play time range reported by a recent systematic review[10] regarding Achilles tendon ruptures treated operatively.

Alkaptonuria is usually asymptomatic during childhood: the only sign can be the black color of the soiled diapers. The diagnosis is often delayed until adulthood, when signs and symptoms become more evident (Table 1)[6]. The main symptoms are joint and back pain due to premature osteoarthritis. Knees are the most frequently affected joints, followed by hips[6]. Tendon thickening and calcification are also frequent and may lead to spontaneous ruptures. The appearance of these lesions can be the first clue suggesting the underlying systemic disease, leading the patient to genetic counseling.

There is no definitive treatment for ochronosis. Phenylalanine and tyrosine diet restriction and daily uptake of ascorbic acid at a dose of 1 g have been used[1]. The antioxidant nature of vitamin C can delay the conversion of homogentisate to the polymeric material and its deposition in connective tissues.

Table 1. Alkaptonuria manifestations by age.

Alkaptonuria manifestation	Age at presentation (years)	Frequency
Black urine exposed to open air	Birth	Frequent
Gray pigmented sclerae and pinnae of the ears, color change of skin and teeth	20-40	Frequent
Tendon and muscle ruptures, thickened Achilles tendon, bursa effusion	30-50	Frequent
Degenerative joint disease, vertebral disk calcification, vertebral fusion, bamboo aspect of the spine, osteopenia and fractures	30-50	Frequent
Decreased respiratory reserve and restrictive lung disease caused by impaired thoracic mobility	>40	Rare
Aortic and mitral valve calcification and murmur, aortic stenosis, hearth failure, coronaropathy	>40	Rare
Kidney stones, sialolithiasis, gallstones, prostatic stones	>40	Rare
Hearing loss, tinnitus, diplopia, peripheral neuropathy, stroke	>40	Rare

However, those treatments were not shown to be effective in clinical studies[1]. There is some evidence[1] that the herbicide nitisinone may be effective in the treatment of alkaptonuria, inhibiting the enzyme 4-hysroxyphenylpyruvate dioxygenase, but further studies are needed.

A review of the treatments and rehabilitation protocols in the previously reported cases of ochronotic Achilles tendon rupture is reported in Table 2[4-7,11,12]. The literature shows highly variable immobilization time (ranging from 5 to 9 weeks) and periods in a plantarflexed cast (from 3 to 6 weeks)[4-7,11,12]. Furthermore, the rehabilitation protocols and follow-up time before the patients reached the final range of motion varied as well[4-7,11,12]. A shared protocol is still missing when Achilles tendon ruptures are due to ochronosis, maybe due to the common belief that this metabolic disorder may impair the tendon healing potential. Nevertheless, in this case, the resection of the degenerate Achilles tendon segment and the suture of normal, vascularized tissues showed an almost physiologic healing time. The Z-lengthening restored the physiological length without excessive tension to the distal insertion.

CONCLUSIONS

Alkaptonuria is a genetic disorder usually diagnosed during adulthood. In this case, the unusual intraoperative findings of a black tendon after an apparently common Achilles tendon avulsion suggested the diagnosis. The treatment performed allowed the patient to return to his preoperative level of sports activity.

Table 2. Achilles' tendon rupture management in patients with alkaptonuria (review of the literature).

First author and year	Treatment	Immobilization time	Rehabilitation protocol and follow up
Alajoulin et al[11], 2015	Surgery (excision of frayed tendon ends and V-Y inverted tenoplasty to restore tendon length)	6 weeks (cast in plantarflexion)	Ankle motion. Walking without aids at 12 weeks. Dorsiflexion 0°-20°, plantarflexion 0°-25°
Ando et al[4], 2004	Surgery Right tendon: modified White and Kraynick Repair + pull-out wire + repair reinforcement with peroneus brevis tendon. Left tendon: reconstruction with anchoring system + reinforcement with peroneus brevis tendon	3 weeks with cast in plantarflexion, 2 weeks with neutral cast after pull-out wire removal	Full weight bearing at 6 weeks from each tendon repair. At 7 months follow up Right ankle range of motion: dorsiflexion 0°-15°, plantarflexion 0°-40°. Left ankle range of motion: dorsiflexion 0°-10°, plantarflexion 0°-40°
Baca et al[5], 2019	Surgery (excision of frayed ends and Lindholm's double facial flap technique)	9 weeks	Not reported. Ankle range of motion (dorsiflexion 0°-20°, full plantarflexion) gained at 30 months follow up
Jiang et al[6], 2019	Surgery (excision of necrotic tendon and long flexor tendon transposition)	6 weeks (cast in plantarflexion)	Ankle motion. Walking without aids at 16 weeks. Dorsiflexion 0°-15°, plantarflexion 0°-20°
Tanoglu et al[7], 2018	Surgery (excision of the distal part of tendon stump and reinsertion with titanium anchors)	4 weeks with a long leg cast in plantarflexion and 30° knee flexion, 2 weeks with a walking cast	Active ankle motion exercises. Full dorsiflexion and plantarflexion gained at 16 months follow up
Wu et al[12], 2018	Surgery (not specified)	Not reported	Not reported

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Authors' Contributions

PA, IM, MDC, and DP contributed to the conception and design of the study, acquisition and interpretation of data; PA drafted the article; PA, IM, MDC, and DP contributed to the revisions of the manuscript; DP supervised the work; PA, IM, MDC, and DP contributed to validation and final approval of the version of the article to be published.

ORCID ID

Ilaria Morelli: 0000-0001-5719-0739

Conflict of Interest

The authors declare that they have no conflict of interest to disclose.

Data Availability

All data generated or analyzed during this study are included in this published article.

Ethics Approval

Not applicable.

AI Disclosure

No AI was used for this study.

Informed Consent

The patient was informed that data concerning the case would be submitted for publication, and he provided consent. No patient's identifying information was included in this article. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation and with the Helsinki Declaration of 1975, as revised in 2013.

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