

SPONTANEOUS ACHILLES TENDON RUPTURE - A CASE OF OCHRONOSIS

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Abstract :

Alkaptonuria is a rare autosomal recessive disorder characterised by the absence of homogentisic acid oxidase and the subsequent accumulation of homogentisic acid, a metabolic product of the aromatic aminoacids phenylalanine and tyrosine; which is deposited in articular cartilages, intervertebral discs, sclera, tympanic membrane, tendons and ligaments leading to their degeneration. Here we describe a case of spontaneous rupture of Achilles tendon^{1,2,3} due to Ochronosis.

Keywords: Alkaptonuria, Ochronosis, Achilles tendon.

Introduction:

The term Ochronosis was first coined by Rudolf Virchow in 1865 and is now used to refer to the musculoskeletal manifestation of alkaptonuria. It is so named after the yellowish (ochre-like) discolouration of the tissues on microscopic examination, however: macroscopically the affected tissues appear bluish-grey. Ochronotic enthesopathy⁴ usually develops after the fourth decade.

Case report:

A 46 year old grandmother presented to our casualty 6 hours after she developed sudden onset of giving way, weakness and mild pain in the back of the right ankle while climbing stairs at her house. On examination she had diffuse swelling over the back of the ankle with a palpable defect over the Achilles tendon proximal to its insertion. There was no external injuries. The discontinuity of the Achilles tendon was confirmed clinically by the inability to

stand on her toes and by the Thompson "squeeze" test and radiologically by the obliteration of the superior corner of the Kager's triangle in a lateral view radiograph of the ankle.

She was operated on the same day under spinal anaesthesia. There was no difficulty in administering spinal anaesthesia. The severed stumps of the tendon were found to have dark pigmentation and were highly indurated almost to the consistency of rubber. The tendon was repaired with prolene, and the limb was immobilised with the ankle in equinus of 20° in an above knee plaster cast for 4 weeks. At 4 weeks the cast was removed. The surgical wound had healed and sutures were removed and a below knee cast with ankle in equinus for 4 weeks was reapplied. After 4 weeks the cast was removed and reapplied with the ankle in neutral position for 4 more weeks and non weight bearing crutch walking was continued. After 4 weeks the cast was removed and she was advised to walk bearing full weight without crutches.

She was reviewed again at 17 weeks post operative period. She had no complaints and was able to walk, squat cross legged and perform her daily activities. On examination the surgical wound had healed. The Achilles tendon was not tender with normal overlying skin. Ankle movements were painless with 0-25° of plantarflexion and 0-20° of dorsiflexion as compared to 0-40° of plantarflexion and 0-25° of dorsiflexion of the contralateral side.

She was reviewed again at 72 weeks post operative period.

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She had no complaints and was able to walk, squat cross legged and perform her daily activities. Ankle movements were painless and equal on both sides with 0-40° of plantarflexion and 0-25° of dorsiflexion, with an improvement in plantarflexion.

Retrospectively, radiographs of her thoraco-lumbar spine showed narrowing of disc spaces with calcification of discs and sclerosis of vertebral margins. She also had dark pigmentation of sclera of both eyes and pinna of both ears which started developing after the age of 40 years, which are progressively getting worse. She did not have any joint or spine abnormalities.

Histopathological examination of a resected sample of the tendon revealed yellow brown pigment (ochre) deposited in the chondrocytes and the tendon. Features suggested possibility of ochronosis. All investigations of blood were within normal limits. 24 hour urine sample turned to a dark colour and was negative for homogentisic acid.

She had similar spontaneous rupture of the contralateral Achilles tendon 4 years prior to this, which was repaired surgically at another hospital. It has healed uneventually except for a tendon stitch granuloma probably caused by prolene suture used to repair the tendon.

She gives a history of visiting hospital at around the age of 4-5 years for dark staining of clothes. All tests done then including 24 hour urine tests were negative.

Her parents and their parents did not have any similar complaints. Her parents were unrelated prior to marriage (not a consanguinous marriage). Her sister aged 37 years has complaints of dark staining of the clothes but she does not have dark pigmentation of her sclera of eyes or pinna of ears. Her sister has no history of spontaneous rupture of tendons or joint or spine involvement. She has four children 2 girls and 2 boys who are all unaffected. However two grandchildren, one boy and a girl; children of one of her daughters have a history of dark staining of clothes but there is no sign of dark pigmentation of sclera of eyes or pinna of ears or spontaneous rupture of tendons or joint or spine involvement as of now in either of the grandchildren.

Discussion:

Alkaptonuria is a rare autosomal recessive disorder with an incidence of 1:1000000. This results in dysfunction of homogentisic acid oxidase, which is an enzyme involved in the metabolism of the aromatic aminoacids phenylalanine and tyrosine⁵.

The patients are usually asymptomatic till the third decade. By the second decade bluish grey pigmentation may appear in the sclera, cartilage of the ear, in the teeth, buccal mucosa and in the nails or skin⁶.

These manifestations are caused by the irreversible binding of homogentisic acid in the fibrillary collagens of connective tissues by polymerisation and oxidation⁷. This inhibits collagen cross-linking leading to reduction of the structural integrity of collagen, thus increasing the likelihood of spontaneous rupture.

Our patient had characteristic radiological changes of the spine⁷ but did not have any spine or joint symptoms. She also had characteristic dark staining of her clothes, dark pigmentation of her sclera of eyes and pinna of ears and her urine turned dark in colour upon standing. The diagnosis was retrospective. There was extensive degenerative changes in the tendon extending beyond the cut ends. The trivial nature of the injury leading to the rupture at this instance and also of the contralateral tendon 4 years prior indicates that spontaneous rupture of Achilles tendon is an important clinical feature of ochronosis. In our patient the tendon healed well after primary repair.

With no specific treatment for ochronosis, dietary reduction of phenylalanine and tyrosine with an administration of vitamin C upto 1g/day is recommended. Vitamin C due to its antioxidant property retards the conversion of homogentisic acid to the polymeric material. Nitisinone⁸ is an experimental drug which inhibits the production of homogentisic acid.



Fig. 1 : Disc



Fig. 2 : Ankle



Fig. 3 : Urine



Fig. 4 : Pinna



Fig. 5 : Tendon

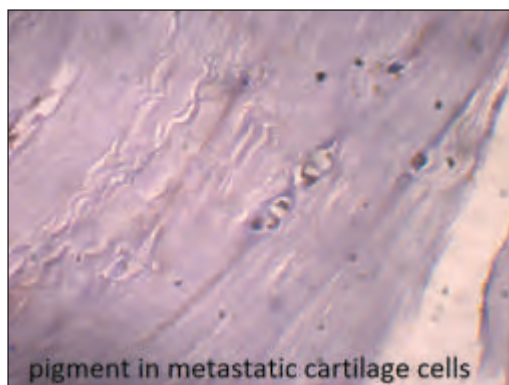


Fig. 6 : Pigment in metaplastic cartilage cells



Fig. 7 : Foot

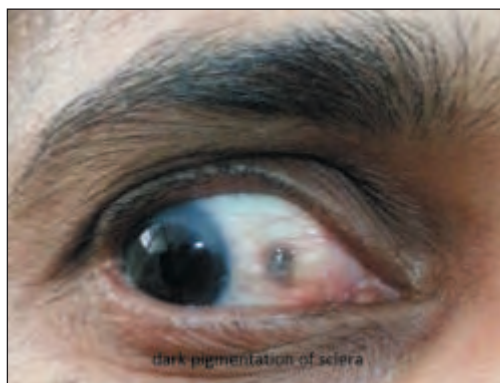


Fig. 8 : Sclera

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