

Spontaneous Achilles Tendon Rupture in Alkaptonuria

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ABSTRACT

Alkaptonuria (AKU) is a rare genetic metabolic disorder caused by deficiency of homogentisic acid oxidase (HGO) enzyme leading to accumulation of homogentisic acid (HGA) in the body. HGA excreted and oxidized through urine causes its characteristic dark discoloration; however, once accumulated in bodily connective tissues, it causes characteristic dark pigmentation (ochronosis) of tissues, and in the long-term, it leads to degeneration of cartilage and tendons. In this case report, we present a patient who has had a unilateral spontaneous atraumatic rupture of the Achilles tendon. To our knowledge, such a case has not been previously reported in the French and Bahraini literature.

Keywords: Ochronosis, Alkaptonuria, Homogentisic Acid, Achilles Tendon

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