

# Ochronosis of Tendo-Achilles - A Case Report and Review of Literature

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## Abstract

Ochronosis is a syndrome caused by the accumulation of homogentisic acid in connective tissues. The condition is most often associated with alkaptonuria but can occur from exogenous administration of phenol complexes like hydroquinone. Alkaptonuria is an autosomal recessive disorder of metabolism. The pathogenesis of alkaptonuria includes chronic inflammation, degeneration, and eventually osteoarthritis. Sclera, skin and heart are also affected. Ochronotic arthropathy is a rare condition found in patients with alkaptonuria. A 33-year-old gentleman presented with history of spontaneous rupture of his Achilles tendon, for which repair was done and sample was sent for histopathological evaluation. Histopathological examination revealed features of Ochronosis.

## Keywords

Ochronosis; Alkaptonuria; Joint

## Introduction

Alkaptonuria is an autosomal recessive metabolic disorder, characterized by homogentisic acid deposition in connective tissue due to deficiency in homogentisic acid oxidase, an enzyme involved in the catabolism of tyrosine and phenylalanine. In alkaptonuria, ochronotic pigment is deposited in all connective tissues, especially cartilage. These pigmentary changes are termed Ochronosis. It can affect the tendons, ligaments, sclera, heart valves, intima of blood vessels and skin [1, 2]. It was first described by Rudolf Virchow in 1865 and named after the yellowish (ocher-like) discoloration of the tissue seen on microscopic examination. Macroscopically the affected tissues appear bluish grey because of Tyndall effect [3].

A 33-year-old gentleman presented with history of spontaneous rupture of his Achilles tendon, for which repair was done and sample was sent for histopathological evaluation. Similar history was observed with his elder brother. He also had history of passing urine, which later turned into brown-black in color after an hour. Histopathological examination revealed features of Ochronosis.

## Results and Discussions

Endogenous ochronosis or alkaptonuria is an autosomal recessive disease caused by a deficiency of homogentisic acid oxidase resulting in the accumulation of homogentisic acid in tissues. Exogenous ochronosis, unlike endogenous ochronosis, does not involve cartilage, tendons, ligaments or sclera and is not associated with dark urine [4].

Exogenous ochronosis develops in case of phenol, pyrocatechin and hydroquinolone intoxication. The earliest manifestation of Ochronosis is skin pigmentation.

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Boedecker was the first person to describe about ochronotic arthropathy. Homogentisic acid causes loss of cartilage elasticity and its successive hardening. It is anticipated that homogentisic acid and its polymers disrupt the connective tissue by unfavorable impact on the proteoglycans [5].

Endogenous and exogenous ochronosis are indistinguishable on histology and the clinical history should be taken into account [4]. Most affected articular sites are para-articular tendon and hyaline cartilage of the hip, knee, shoulder, intervertebral discs, cartilage of ear and nose. The majority of exogenous lesions are found on sun-exposed areas of the body, the darker skin with brown-gray or blue-black hyperpigmentation [6].

In healthy patients, homogentisic is lacking both in blood plasma and in urine. Homogentisic plasma levels more than 6.6 pg/ml and in the urine of an average of 3.12 mmol/mmol creatinine is proofing the existence of alkaptonuria. Till date, no effective therapy exists to reduce the complications of alkaptonuria. The treatment includes conservative treatment of osteoarthritis with anti-inflammatory drugs and physical therapy. In addition, the administration of high doses of ascorbic acid (vitamin C) and dietary restriction of phenylalanine and tyrosine are often recommended. Unfortunately no benefits have been demonstrated in adults [7].

## Conclusion

Vitamin E and N-acetyl cysteine have been examined as novel potential therapies to prevent damage to articular cartilage. Ochronotic arthropathy is treated with physiotherapy, analgesia, rest, and prosthetic joint replacement when necessary. One possible hope is that nitisinone proves effective. The US Food and Drug Administration (FDA) have approved this drug for the treatment of tyrosinemia type 1. It significantly lowers the urinary excretion of HGA by inhibiting 4-hydroxyphenylpyruvate dioxygenase and, theoretically, would reduce HGA accumulation. Testing presently is assessing safety and long-term results [8].

## References

1. Smith RJ (2000) Disorders of amino acid metabolism. In: Humes HD, editor. Kelley's text-book of internal medicine 2788-2793.
2. Kontinen YT, Hoikka V, Landtman M, et al. (1989) Ochronosis: a report of a case and a review of literature. ClinExp Rheumatol 7: 435-444.

3. Findlay GH (1989) Ochronosis. Clinics in Dermatology 7:28-35.

4. Martins VM, Sousa AR, Portela Nde C, et al. (2012) Exogenous ochronosis: Case report and literature review. An Bras Dermatol 87:633-636

5. JozefRovenský, Tibor Urbánek, OľgaBoldišová, et al. (2011) Alkaptonuria and ochronosis.34-37.

6. Breer S, Amling M, Püschel K, et al. (2012) Ochronose. Rechtsmedizin 22:49-53.

7. Oberarzt CB, Thormann U, Madera N, et al. (2016) Joint Involvement in Ochronosis. Surgery Curr Res 6: 279.

8. Introne WJ, Perry MB, Troendle J, et al. (2011) A 3-year randomized therapeutic trial of nitisinone in alkaptonuria. Molecular Genetics and Metabolism 103: 307-14.

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