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Case Report

Knee osteoarthrosis secondary to ochronosis – clinical case☆,☆☆

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ABSTRACT

Alkaptonuria is a rare metabolic disease in which a deficiency of the enzyme homogentisate dioxygenase causes an accumulation of homogentisic acid. Ochronosis consists of excessive deposition of homogentisic acid in the connective tissue and presents as a chestnut brown or black pigmentation. With aging, the accumulation of pigments from homogentisic acid in the joints causes osteoarthrosis. There is no specific treatment for the disease and the approach is symptomatic. Arthroplasty is the solution for severe cases of osteoarthrosis caused by this pathological condition and presents results comparable to those from patients with primary osteoarthrosis. Here, the case of a 67-year-old patient who underwent several arthroplasty procedures because of osteoarthrosis caused by this rare pathological condition is presented. The last surgical intervention consisted of total right knee arthroplasty.

Osteoartrose do joelho secundária a ocronose – Caso clínico

Palavras-chave:
Alcaptonúria
Ocronose
Joelho
Artroplastia

RESUMO

A alcaptonúria é uma doença metabólica rara em que a deficiência da enzima ácido homogentísico-oxidase provoca uma acumulação de ácido homogentísico. A ocronose consiste na deposição excessiva de ácido homogentísico no tecido conjuntivo e apresenta-se como uma pigmentação acastanhada ou preta. Com o envelhecimento, a acumulação de pigmentos de ácido homogentísico nas articulações provoca osteoartrose. Não existe um tratamento específico para a doença e a abordagem é sintomática. A artroplastia é a solução

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para casos graves de osteoartrose causada por essa patologia e apresenta resultados comparáveis aos doentes com osteoartrose primária. Os autores apresentam o caso de um doente de 67 anos submetido a várias artroplastias, em virtude da osteoartrose causada por essa rara patologia. A última intervenção cirúrgica foi uma artroplastia total do joelho direito.

Introduction

Alkaptonuria is a rare recessive autosomal metabolic disease caused by absence of the enzyme homogentisic oxidase. This enzyme is responsible for degradation of homogentisic acid, which is an intermediate product from metabolism of the amino acids tyrosine and phenylalanine. If this enzyme is defective, this leads to accumulation of homogentisic acid in tissues and blood.

The incidence of alkaptonuria is less than one in one million.[1]

Over time, the deposits of homogentisic acid accumulate in the tissues and present as a dark pigmentation. This condition is called ochronosis and it may affect not only the musculoskeletal system but also the cardiovascular and genitourinary systems, the sclera and the skin.[2,3]

Most of the symptoms of alkaptonuria are only observed starting in the fourth or fifth decade of life,[4] except for the appearance of dark urine, which is detected during childhood, resulting from excretion and oxidation of homogentisic acid.

Alkaptonuria causes progressive ochronotic arthropathy of the major joints that are subject to weight-bearing. The knee is the joint that is most affected, followed by the hip.[4–6]

The treatment for the disease is symptomatic and total arthroplasty is the preferred treatment in severe cases of osteoarthrosis.[7,8]

Clinical case

The patient was a 67-year-old man whose diagnosis of alkaptonuria had been made at the age of 40 years. The first signs of the disease were darkening of the urine and appearance of dark pigments in the sclera, ears and first interdigital crease of the left hand (Fig. 1A–D). There were no other relevant antecedents or any family history of the disease.

At the age of 60 years, the patient underwent a surgical intervention to extract a bladder stone of large dimensions (Fig. 2).

Joint complaints arose some years later and initially affected the left hip, followed by the left knee and lastly the right knee.

The patient was referred for an orthopedics consultation and the first surgical intervention was total arthroplasty of the

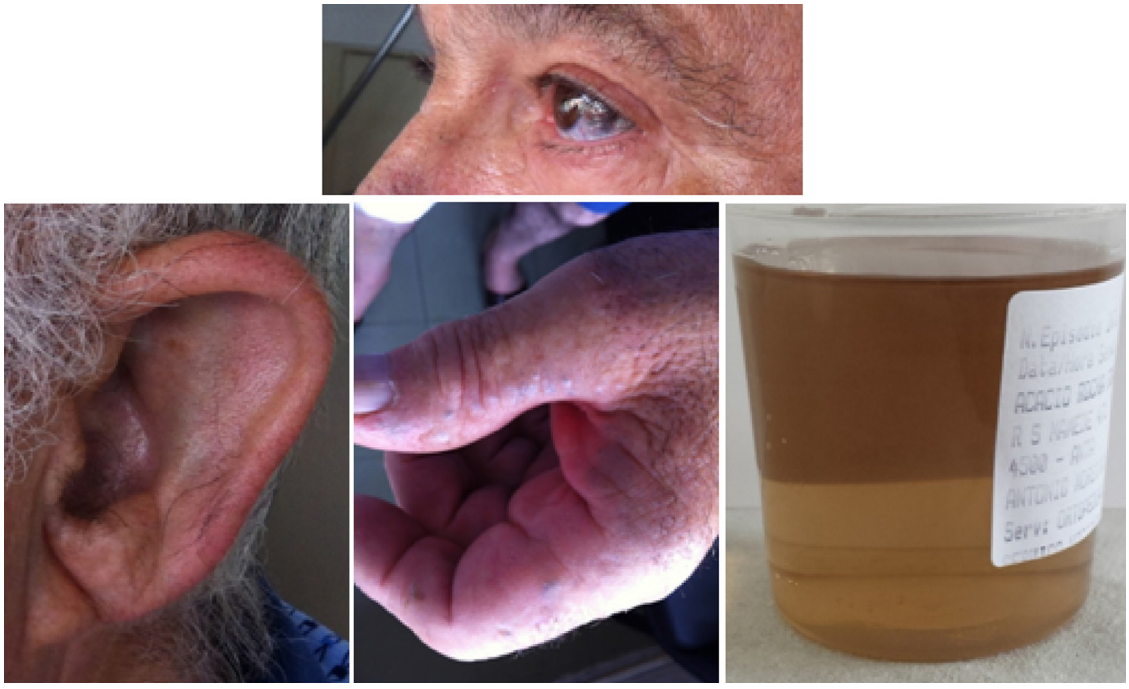

Fig. 1 – Dark pigment in the sclera, ears and first interdigital crease of the left hand. The last figure demonstrates the darkened appearance of the urine.