Rev esp cir ortop traumatol. 2010;**54(6)**:396-398

Revista Española de Cirugía Ortopédica y Traumatología

www.elsevier.es/rot

NOTA CLÍNICA

Ochronotic arthropathy: A presentation of 2 cases

L. Fontao-Fernández*, M.J. Ferreirós-Conde and J. Otero-Villar

Orthopaedic Surgery and Traumatology Department, Hospital do Meixoeiro, University of Vigo Hospital Complex, Vigo, Spain

Received on March 21st, 2010; accepted on August 5th, 2010

KEYWORDS
Ochronosis;
Arthropathy;
Knee;
Hip

Abstract
Introduction: Alkaptonuria is a hereditary metabolic disease, characterised by the triad: dark urine, connective tissue pigmentation (ochronosis) and degenerative arthritis of the weight-bearing joints.
Clinical cases: We present two cases of ochronotic arthropathy treated by arthroplasty.
Conclusions: Given that there is currently no medical treatment that can halt or reverse this metabolic process, arthroplasties, are, at present, a good therapeutic option in cases that have progressed to joint involvement.

PALABRAS CLAVE
Ocronosis;
Artropatía;
Rodilla;
Cadera

Artropatía ocronótica: a propósito de 2 casos

Resumen
Introducción: La alcaptonuria es una enfermedad metabólica hereditaria caracterizada por la tríada: orina oscura, pigmentación del tejido conectivo (ocronosis) y artritis degenerativa de las articulaciones de carga.
Casos clínicos: Presentamos dos casos de artropatía ocronótica tratados mediante artroplastia.
Conclusiones: Dado que en la actualidad no existe ningún tratamiento médico que pueda frenar o revertir este proceso metabólico, las artroplastias son hasta el momento una buena opción terapéutica en casos evolucionados de afectación articular.

*Corresponding author.
E-mail: lorena_fsc@hotmail.com (L. Fontao-Fernández).

1888-4415/$ - see front matter

Introduction

Alkaptonuria is a hereditary metabolic disease with a hallmark triad consisting of dark urine, pigmentation of the connective tissue (ochronosis), and degenerative arthritis of the weight-bearing joints.[1,2] It is autosomal recessive, is almost equally distributed between both sexes, and its incidence varies between one in every million to one in every ten million inhabitants. Mutations in the homogentisic gene (HGO), located on the long arm of chromosome 3, produce a metabolic disorder characterized by the partial or complete absence of homogentisic acid oxidase, the enzyme in charge of catabolizing tyrosine and phenaylalanine.[1-3] This causes serum levels of this acid to rise, accumulating in the connective tissues with polymerization of collagen fibres and, through oxidation, a blackish coffee-coloured pigment is formed. This pigment is deposited and stains the cartilage of the joints and periarticular tissues in particular. Other locations include the intervertebral discs, the cartilage of the ear, the sclera, the tympanum, heart valves, or the larynx, and its excretion in urine is characteristic.[2,3,10]

Case report 1

Seventy-one year old female previously treated with total arthroplasty of the left hip in 1999 who is diagnosed with right coxarthrosis, for which she underwent surgery in 2002. During the arthroplasty of the right hip, black pigmentation of the cartilage of the femoral head is observed and this, together with the pigmentation of sclera of the eyes and ear cartilages, is cause to suspect alkaptonuria. A specimen is sent to the pathology department; a radiological study is conducted of the skeleton, and a 24-hour urinary homogentisic acid quantification is carried out, confirming the microscopic diagnosis. Joint discs in the dorsolumbar spine present calcification and alkaptonuria is detected. Over the course of the following years, se develops bilateral gonarthrosis, causing her to undergo total arthroplasty of the left knee in March, 2007. During surgery, once again dark staining of the joint cartilage and periarticular tissues is observed (fig. 1).

Figure 1 Intra-operative image of ochronotic arthropathy of the knee. Involvement of the lumbar intervertebral discs. Pigmented sclera.

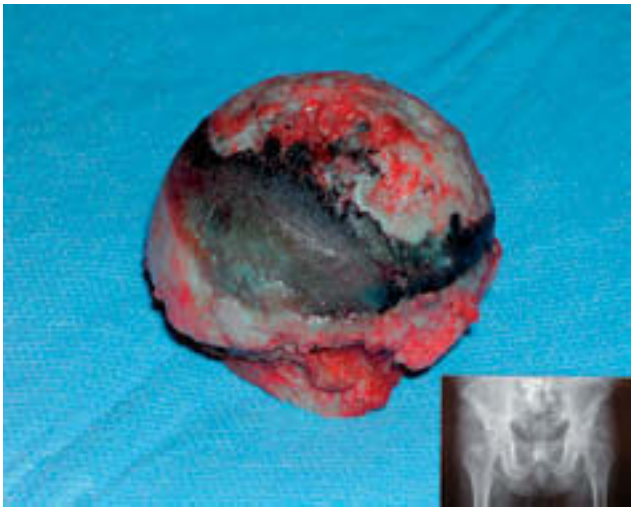

Figure 2 Ochronotic arthropathy of the hip: macroscopic appearance of the femoral head and AP x-ray of both hips.

Case report 2

Sixty-five year old male diagnosed with alkaptonuria and right coxarthrosis, for which he was treated with total hip arthroplasty, at which time the pigmentation characteristic of the disease was observed in said joint. As in the previous case, he presented calcification of intervertebral discs, although no external signs of the disease were seen (fig. 2).

Results

No complications with stability in the fixation of the implants have been seen thus far.

Discussion

The accumulation of homogentisic acid in the cartilage modifies its consistency and makes it more friable, which accelerates its degeneration and leads to arthritis, affecting multiple joints.

The differential diagnosis of this illness can include osteoarthritis, ankylosing spondylitis, rheumatoid arthritis, and chondrocalcinosis. The calcifications of the vertebral discs may be similar to those seen in hyperparathyroidism, amyloidosis, haemochromatosis, diffuse idiopathic skeletal hyperostosis, or vertebral arthrodesis.

The course and prognosis of this disease depends on the severity of the alkaptonuria. At present, there is no medical treatment capable of halting or reversing this metabolic process.[3,5-7] We agree with other authors with respect to the satisfactory evolution of arthroplasties in this illness, the only treatment alternative at present in advanced cases of articular involvement.[4,5,7-9]

References

1. Shimizu I, Hamada T, Khalpey Z, Miyanishi K, Hara T. Ochronotic Arthropaty: pathological evidence of acute destruction of the hip joint. Clin Rheumatol. 2007;26:1189-91.
2. O'Brain WM, La Du BN, Bunim JJ. Biochemical, pathologic and clinical aspects of alkaptonuria, ochronosis and ochronotic arthropathy. Review of world literature (1584-1962). Am J Med. 1963;34:813-38.
3. Mannoni A, Selvi E, Lorenzini S, Giorgi M, Airó P, Cammelli D, et al. Alkaptonuria, ochronosis and ochronotic arthropathy. Semin Arthritis Rheum. 2004;33:239-48.
4. Aydogdu S, Cullu E, Ozsoy MH, Sur H. Cementless Total Knee Arthroplasty in Ochronotic Arthropathy: a case report with 4-year follow-up. J Arthroplasty. 2000;15:539-43.
5. Miyazaki T, Kokubo Y, Kobayaschi S, Yayama T, Sato R, Uchida K, et al. Ochronotic arthropathy requiring bilateral khip and knee joint arthroplasty: case study. Japanese Journal of Rheumatism and Joint Surgery. 2007;25:489-96.
6. Laskar FH, Sargison KD. Ochronotic Arthropathy, A Review with Four Cases Report. J Bone Joint Surg Br. 1970;52-B:653-66.
7. Spencer JMF, Gibbons CLH, Sharp RJ, Carr AJ, Athanasou NA. Arthroplasty for ochronotic artritis. Acta Orthop Scand. 2004;75:355-8.
8. Araki K, Sudo A, Hasegawa M, Uchida A. Devastating ochronotic arthropathy with successful bilateral hip and knee arthroplasties. J Clinical Rheumatol. 2009;15:138-40.
9. Carrier DA, Harris CM. Bilateral hip and bilateral knee arthroplasties in a patient with ochronotic arthropathy. Orthop Rev. 1990;11:1005-9.
10. Emel E, Karagoz F, Aydin H, Hacisalihoglu S, Seyithanoglu MH. Alkaptonuria with lumbar disc herniation: a report of two cases. Spine. 2000;25(16):2141-4.