OCHRONOTIC ARTHROPATHY : THE BLACK HIP CASE REPORT AND REVIEW OF THE LITERATURE

K. DOM, T. PITTEVILS

Ochronosis is the dark pigmentation of connective tissues in patients with alkaptonuria. The latter is an autosomal recessive deficit of phenylalanine and tyrosine metabolism with various clinical manifestations.
We report on a 63-year-old man with familial ochronosis, who presented with a terminal degenerative right hip joint ; his older brother had already been succesfully treated at our department by total hip replacement. The postoperative course was uneventful and satisfactory for both.

Keywords : ochronosis ; arthropathy ; alkaptonuria.
Mots-clés : ochronose ; arthropathie ; alcaptonurie.

INTRODUCTION

Alkaptonuria is a rare hereditary error of phenylalanine and tyrosine metabolism, with an incidence of one in a million births (2). It is an autosomal recessive disease, affecting males and females in an equal rate, altough there have been few cases of suspected dominant hereditary transmission (2, 3).

The gene coding for homogentisic acid oxidase, an enzyme found solely in kidney and liver tissue, is absent in alkaptonuria, which catalyses the change of homogentisic acid (HGA) to maleylacetoacetic acid (2, 3). This causes an accumulation of HGA in connective tissues where polymerisation with collagen fibres occurs, giving a melanin-like pigment (5). This brownish-black pigment stains especially the cartilage of the joints and intervertebral discs, hence the term ochronosis (= black pigment in connective tissues, which stains ochre under the microscope) (1). Its accumulation modifies the biochemical quality of the cartilage, that becomes friable and degenerates (4). The exact mechanism leading to the arthritis is still unknown.

Clinical signs become obvious during the third or fourth decade, although the metabolic error is present during the whole life. In alkaptonuria a large amount of HGA is present in the urine, which turns black upon oxygenation or alkalinisation (1). Spondylosis and ochronotic arthropathy are frequent manifestations of alkaptonuria (2).

CASE REPORT

In July 1995 a 63-year-old man presented with increasing pain in his right groin and difficulty in walking. These complaints had been increasing for one year and during the last few months his sleep at night was disturbed. Since 1975 there was a history of low back pain with stiffness. In 1989 progressive aortic valve stenosis was diagnosed and due to severe degenerative calcification of the aortic cusps, operative replacement of the aortic valve with a mechanical valve was performed in January 1992. Previously one elder brother was known with ochronosis and also had to undergo aortic valve replacement. A few months earlier this same brother was treated succesfully by total hip replacement arthroplasty.

On physical examination, pigmentation of the ears and sclerae was seen and he had hearing loss.

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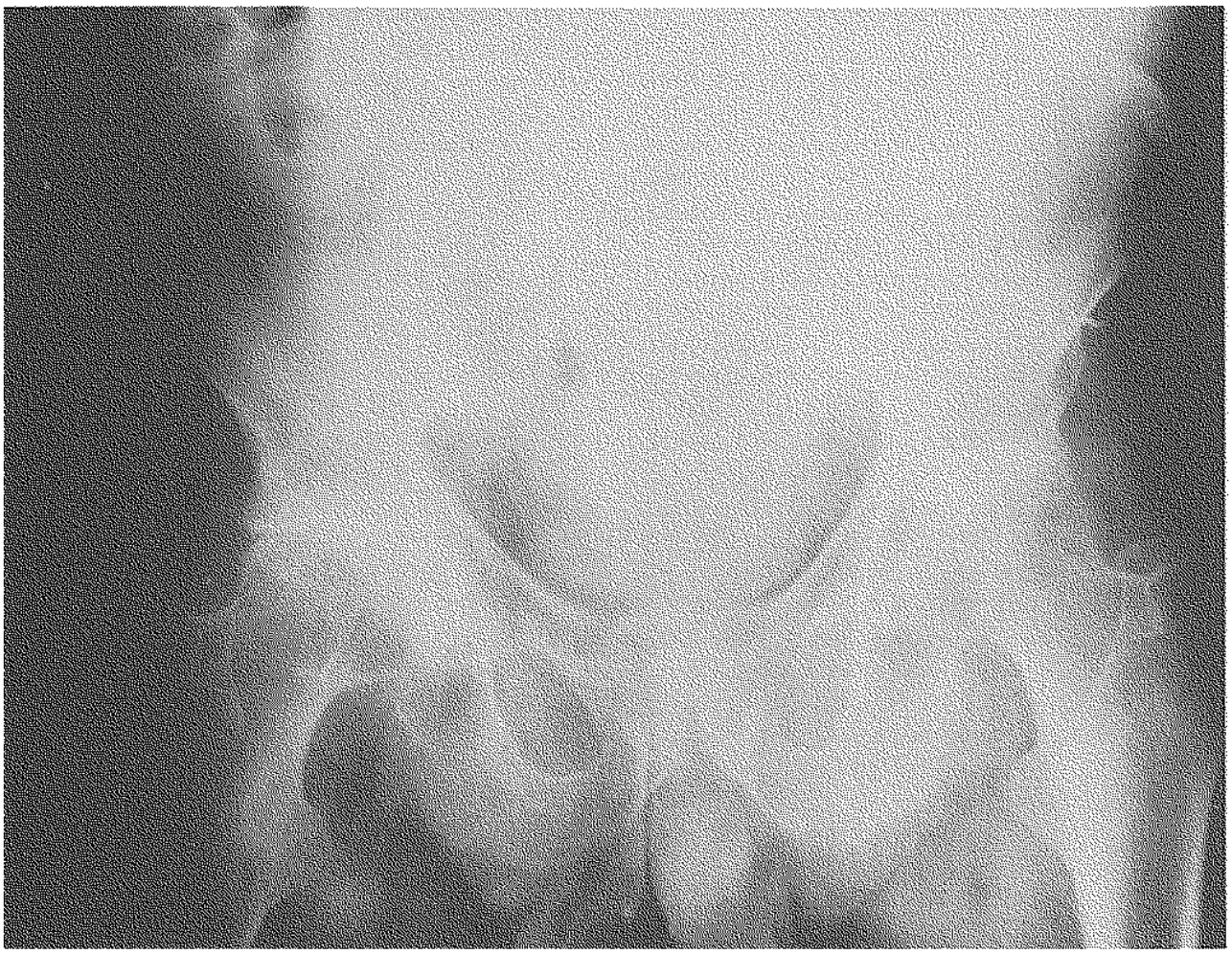

Fig. 1. — Ochronotic arthropathy of the right hip with almost absent joint space, subchondral sclerosis, cystic changes and periarticular osteophytosis.

There was a general rigidity of the spine with flattening of the lumbar lordosis and dorsal hyperkyphosis. The right hip was markedly limited and painful in rotation, with an extension deficit of 25 degrees. Pathological cardiac murmers were absent.

A lateral radiograph of the lumbar spine showed the typical calcification and narrowing of the intervertebral discs, with minimal osteophytosis of the vertebral bodies and degenerative apophyseal joints. Computed tomography confirmed these findings without any evidence of spinal stenosis. An anteroposterior x ray of the pelvis (Fig. 1) revealed a destructive ochronotic arthropathy of the right hip. The sacroiliac joints (Fig. 1) were narrowed with osteophytosis and subchondral sclerosis, but not fused.

Preoperative laboratory findings, including a routine urinalysis were normal. The patient was taken to surgery for total hip replacement arthroplasty. Peroperatively upon opening the hip capsule and dislocating the joint, a blackening was noted both in the remaining cartilage of the acetabulum and the femoral head.

Macroscopic examination of the femoral head showed the border of black cartilage. Histological examination study revealed black pigmented cartilage together with typical detritus synovitis.

The postoperative course was uneventful and one year later the patient had no complaints.

DISCUSSION

Alkaptonuria often goes unrecognized until middle age when degenerative joint disease becomes disabling in the majority of cases (3). Spondylosis is the most common first sign of ochronosis (2). However, the diagnosis of ochronosis should be obvious, as the radiographic appearance of the spine is pathognomonic : calcification or ossification of the intervertebral discs, which is absent in all other diseases that can cause spondylosis (ankylosing spondylitis, rheumatoid arthritis, osteoarthritis, psoriatic arthritis) (2). There is a typical narrowing of the disc spaces and the vertebral osteophytes have the tendency to be rather small (4). Usually the apophyseal facet joints and the sacroiliac joints are not fused (3, 4). Arthritis mainly affects the large peripheral joints, causing painful limitation of movement of hips, knees and shoulders, which often occurs many years after spondylosis started (2). Deposition of calcium pyrophosphate crystals with acute periods of pseudogout is possible in ochronosis (4, 5). The small joints of hands and feet are left undisturbed (6). In men, arthritis appears to have an earlier onset and a more severe course than in women (1).

Other clinical signs, besides ochronotic arthropathy, are pigmentation of the ear lobes, sclera, nose, axilla and groin. The cerumen is black and the tympanic membranes can also appear discolored associated with a hearing impairment (3). Pigmentation (which is situated intra- and extracellularly) of tendons, ligaments, tracheal rings, heart valves also occurs and lesser deposition is present in the endocardium, intima and viscera. Further complications involve aortic valve calcification and stenosis. The latter was present in our patient and his elder brother, even before severe arthritis showed up. Occasionally the genitourinary tract obstructs by ochronotic calculi.

In early infancy, the characteristic dark staining of the napkins moistened with the urine, is sometimes the first element of diagnosis (2). Our patient never noticed discoloration of the urine, as it darkens only after several hours on oxygenation and even later if acid urine or high ascorbic acid concentrations are present. Homogentisic acid in urine can be identified by different tests : addition of ferric chloride shows a purple black color ; addition of aqueous silver nitrate solution (and a dilute ammoniumhydroxide solution) turns the urine immediately black ; treatment with Benedict's reagent yields a brown color. More specific identification of HGA in urine can be achieved by paper chromatography, enzymatic assay or spectrophotometric determination (3).

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SAMENVATTING

K. DOM, T. PITTEVILS. Ochronose : de zwarte heup.

Ochronose betekent de donkere verkleuring van bindweefsel bij patiënten die lijden aan alkaptonurie. Dit laatste is een autosomaal recessief defect van het phenylalanine en tyrosine metabolisme, waarbij zich een gevariëerd klinisch beeld manifesteert.
We melden het geval van een 63-jarige man met familiale ochronose, die consulteerde met een terminaal degeneratief rechter heupgewricht. Zijn oudere broer werd succesvol behandeld met een totale vervangingsarthroplastie. Het postoperatieve verloop was zonder complicaties en bevredigend voor beiden.

RÉSUMÉ

K. DOM, T. PITTEVILS. L'arthropathie ochronique : la hanche noire.

L'ochronose signifie la coloration foncée du collagène chez des patients qui souffrent d'alcaptonurie. Cette dernière est une anomalie autosomique récessive du métabolisme de la phénylalanine et de la tyrosine, se manifestant par un tableau clinique varié.

Nous présentons le cas d'un homme âgé de 63 ans, atteint d'ochronose familiale. Il consultait pour une articulation coxo-fémorale dégénérative au stade terminal. Son frère aîné avait déjà été traité avec succès par arthroplastie totale de la hanche. L'évolution post-opératoire fut sans complications et satisfaisante pour chacun d'entre eux.