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Case Report
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ARTHROPLASTY IN ALKAPTONURIC OCHRONOSIS.

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ABSTRACT

Ochronotic degenerative arthropathy occurs in patients with alkaptonuria. Alkaptonuria disorder is an extremely rare disease characterized by black pigmentation of various tissues (e.g., cartilage and connective tissue). Ochronotic arthropathy is a disabling disease that primarily affects the large joints. Like other metabolic diseases that involve the musculoskeletal system, care must be taken with regard to the quality of the affected bones, tendons and ligaments, and therefore the stability and survival of joint prosthesis. The following is a report of a 66-year-old man affected by several musculoskeletal manifestations of alkaptonuria with severe joints disruption, who was successfully treated with total left hip and total right knee replacements. Surgical, anesthesiological and postoperative management of these patients may require more vigilance due to the associated comorbidities of this disorder.

Keywords: *alkaptonuria; ochronotic arthopathy; total knee arthroplasty; total hip arthroplasty*

INTRODUCTION

Alkaptonuria is a rare, autosomal, recessive, inherited disease that has an estimated worldwide birth prevalence of around 1/111,000–1/1,000,000 (1–3). It is caused by a deficiency of the enzyme homogentisic acid oxidase, which causes accumulation of excess homogentisic acid, an intermediate in the catabolism of tyrosine, in the cells and bodily fluids, resulting in dysfunction involving bone, cartilage, and other connective tissues (3–5). Ochronotic arthropathy is characterized by premature progressive

degenerative change that leads to complete disruption of the axial skeleton and peripheral large joints, with an important negative influence on the health-related quality of life. We report the case of a patient with ochronotic arthropathy involving the spine, bilateral hips and knees, who underwent total arthroplasty on his left hip and right knee.

CASE REPORT

A 66-year-old man with alkaptonuria presented to our department with a medical history of lumbar, left hip, and bilateral knee pain. He had progressive limitations in daily activity for the past 6 years. No trauma was reported in his anamnesis. X-ray examination showed severe degenerative change in his left hip, with osteophytes and reduced joint space. Lumbar and knee X-rays also showed significant signs of osteoarthritis. He underwent a cementless total left hip prosthesis via the direct lateral approach.

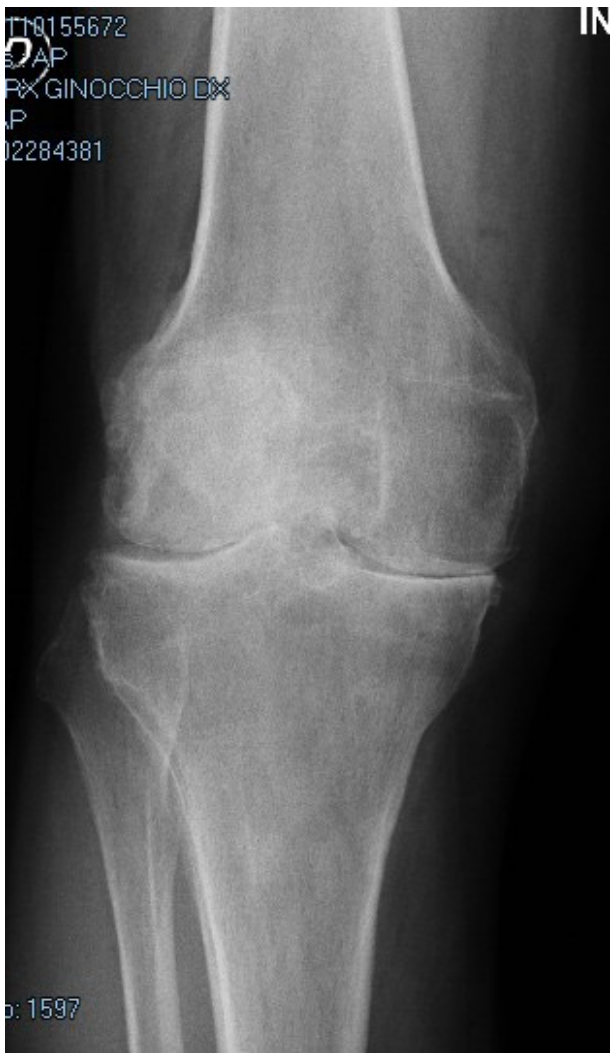

FIG 1. Knee X-ray showing degenerative change.

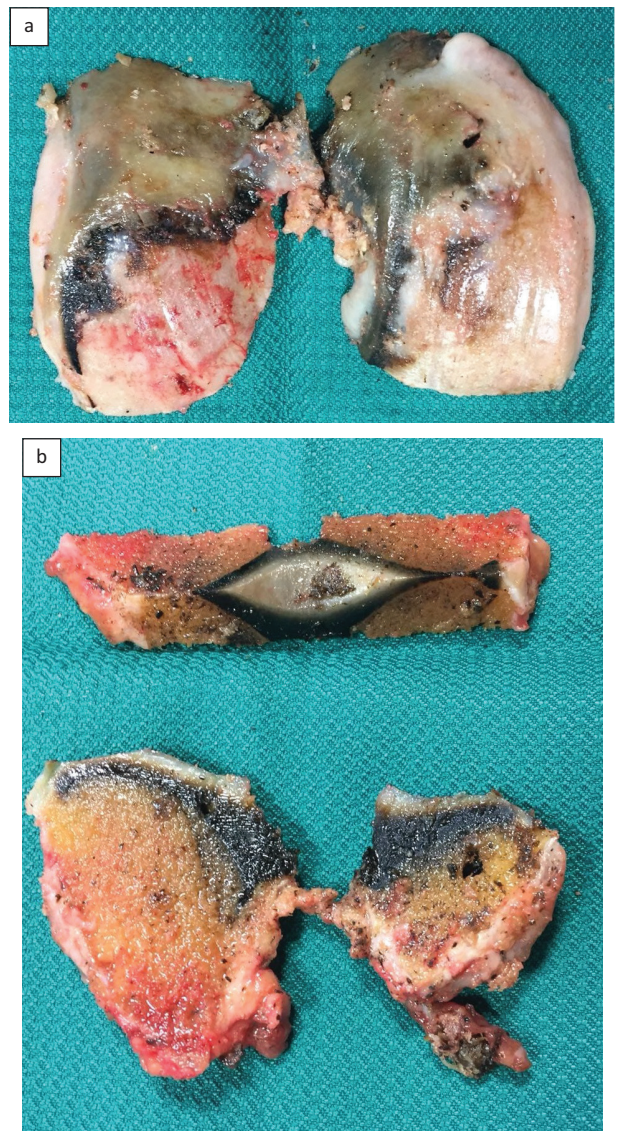

FIG 2. (a, b). Surgical image: The disease involving bone and cartilage.

J Popul Ther Clin Pharmacol Vol 26(2):e20–e24; July 23, 2019.

The joint presented severe acetabular and femoral degenerative changes. Postoperatively, partial weight-bearing with crutches was permitted during the first week.

One year later, the patient had recovered from the total hip arthroplasty, but was admitted to our department due to constant and progressive pain in the right knee, which had not improved with conservative treatment (e.g., oral analgesics, nonsteroidal anti-inflammatory drugs, and physiotherapy).

Examination of the patient revealed a painful right knee with limited movement. X-rays of the right knee showed severe degenerative change (Figure 1). A cemented posterior stabilized total knee arthroplasty was performed on the patient's right knee with his consent.

A standard midline incision was used. The subcutaneous and synovial tissues presented an area of black pigmentation; no pathological extensor mechanism alteration was found, but blackened articular cartilage and bone of the femur, tibia and patella were observed (Figures 2a, 2b, 3). The cemented prosthesis was very lightly impacted.

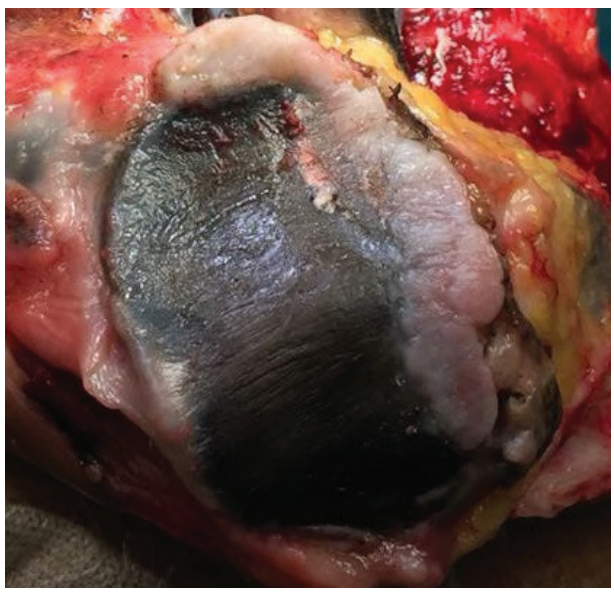

FIG 3. The typical black color of the patellar cartilage surfaces.

There were no difficulties or complications during the surgery. Postoperatively, the patient had an increased loss of blood totaling 1,100 ml and he needed one unit of blood transfusion. We did not use postoperative blood salvage as is customary. The operated joint was passively mobilized on the second postoperative day, and partial weight-bearing with crutches was permitted on the third day. Postoperative wound healing was normal.

At the 12-month follow-up, the patient was very satisfied, with a good range of motion (0–115°) and a pain-free knee. X-ray examination showed correct positioning of the components (Figure 4), and the patient could perform the activities of daily life with no pain, swelling, or any other limitations.

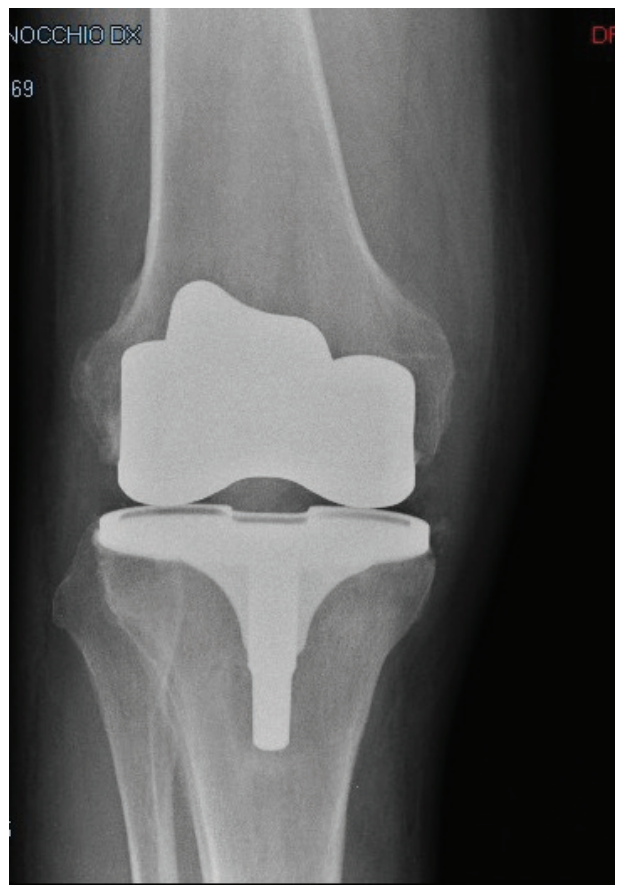

FIG 4. Knee replacement.

J Popul Ther Clin Pharmacol Vol 26(2):e20–e24; July 23, 2019.

DISCUSSION

Alkaptonuria is an extremely rare autosomal, recessive, inherited disease, associated with metabolic dysfunction that involves bone, cartilage, and other connective tissues. These tissues are characterized by homogentisic acid deposit with dark pigmentation caused by a deficiency in the enzyme homogentisic acid oxidase, which is present in the liver and kidney (4–6).

Alkaptonuric ochronosis causes bluish-black discoloration and weakness of some connective tissues. It is an uncommon cause of progressive arthropathy and is in most cases diagnosed only from intraoperative findings. This disease could potentially be misdiagnosed using conventional imagery, which may appear as primary osteoarthritis, unless assessed by thorough the patient's history analysis and physical examination (6–9).

After the third decade of life, the renal clearance of homogentisic acid decreases, and the musculoskeletal manifestations of alkaptonuric ochronosis progressively appear. The disease is more severe in male patients than in female patients (6–8). The most common musculoskeletal localizations of ochronotic disease are the spine (intervertebral disc calcification and narrowing of the intervertebral space) and the large peripheral joints (hip, knee, and shoulder). Progressive chondral damage is observed in almost all patients as they grow older (10). The manifestation is mainly found in the large joints and infrequently in the small joints of the hands. The main symptoms include pain, swelling, and limited range of motion, leading over time to a total inability to use the affected joint.

Treatment for ochronotic degenerative arthropathy could be started as for degenerative osteoarthritis, with therapies such as analgesic drugs, nonsteroidal anti-inflammatory drugs (NSAIDs), rest, physiotherapy, and local injections.

In case of severe arthropathies, if conservative treatment are not effective, the only alternative is the joint replacement under general or spinal anesthesia (11–14); care must be taken with regard to the weakness of the bone, and the altered strength and resistance of tendons and ligaments with high risk of intraoperative complications affecting the stability and survival of joint prosthesis. There are a few, rare case reports describing joint arthroplasty in the literature (14–19). No complications or other adverse events have been reported (12–17). Most of the recently published studies report excellent outcomes for joint replacement surgery (13).

CONCLUSIONS

In accordance with the literature, our case report demonstrates that total hip and total knee arthroplasty has an excellent outcome in patients with severe degenerative arthropathy, secondary to ochronosis, with survival comparable to prosthesis in patients with osteoarthritis (17, 20, 21).

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J Popul Ther Clin Pharmacol Vol 26(2):e20–e24; July 23, 2019.