

## Case Report

# The black bone disease: a case report of ochronotic hip arthropathy

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### ABSTRACT

Ochronotic arthropathy is a rare complication in patients with alkaptonuria (AKU) that arises as a result of accumulation of ochronotic pigment in the joints. This case report presented a 70-year-old female patient with chronic pain in B/L knee and right hip with decreased range of motion. The physical and radiographic findings were agreeable with end-stage hip osteoarthritis and knee osteoarthritis. The diagnosis was done by finding of a dark capsule and femoral head during the total hip replacement. The surgical treatments significantly minimized and enhanced the range of motion (ROM). AKU normally emerges after age 40 and is normally asymptomatic till the involvement of the spine, hip, knee and shoulder joints. Therefore, orthopedic surgeons must be observant of clinical manifestations of this rare condition, before and during the surgery. Arthroplasty is an appropriate therapeutic recourse for patients suffering from ochronotic arthropathy.

**Keywords:** Arthroplasty, Alkaptonuria, Ochronosis

### INTRODUCTION

AKU is an atypical hereditary disease of the phenylalanine and tyrosine metabolism, affecting one in 100,000 to 250,000 people worldwide.<sup>1</sup> It is caused by the decreased function of the homogentisate 1,2-dioxygenase enzyme (HGD) and successive accumulation of the enzyme's substrate, homogentisic acid (HGA), in the blood and body tissues.<sup>2</sup> Autoxidation and polymerization of the HGA in collagenous tissues produce ochronotic pigment which get deposited in various tissues such as the skin, cardiovascular system, kidney, joints, ligaments, and tendons.<sup>3</sup> The orthopedic manifestation of this condition includes early-onset involvement of the lumbar spine with stiffness and pain, thickening and rupture of the tendons, muscle tearing after low-energy trauma, and ochronotic arthropathy in the knee, hip, and shoulder joints. The ochronotic deposits make the cartilage fragile and rigid, leading to degenerative changes in the axial and peripheral skeleton, usually between the ages of 40 to 50 years old.<sup>4</sup> Based on the severity of degeneration, surgical treatment may include synovectomy, arthroscopic debridement, or arthroplasty. Although ochronotic arthropathy is a rare and

late sequela in AKU, it can be rapid and aggressive. In many cases, the disease is not diagnosed until the time of arthroplasty.<sup>5</sup> In this report, we presented a patient with ochronotic arthropathy who underwent total right hip arthroplasty.

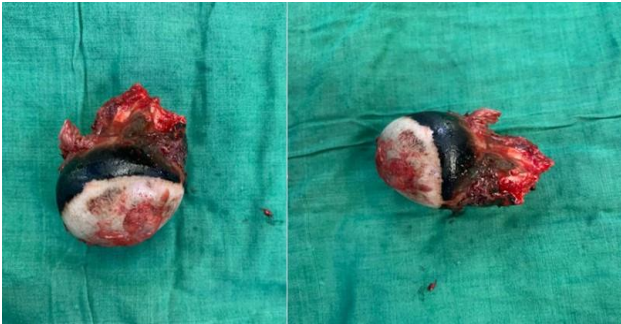
### CASE REPORT

A 70 year old woman came to our orthopaedic department with pain in the right hip and B/L knee for the last four years with no history of trauma or evidence of inflammatory arthritis. No familial history of metabolic disorders was present. In the local examination, there was decreased ROM of the right hip, and painful movement was detected. Other examinations were unremarkable. The left hip had no pain and the ROM was normal. X-ray was suggestive of the right hip and B/L knee degenerative arthritis with joint space narrowing, subchondral sclerosis, and cyst formation. Other findings were osteophyte formation and degenerative changes in the lumbar spine, scoliosis deformity, and bilateral sacroiliac joints (Figure 1). Primarily, she had taken conservative management

using analgesics and physical rehabilitation for 1 year but did not get relieved.



**Figure 1: Initial radiograph (A) pelvis (B) knee joint (C) L-S spine.**



**Figure 2: Removed black femoral head with thick capsule.**



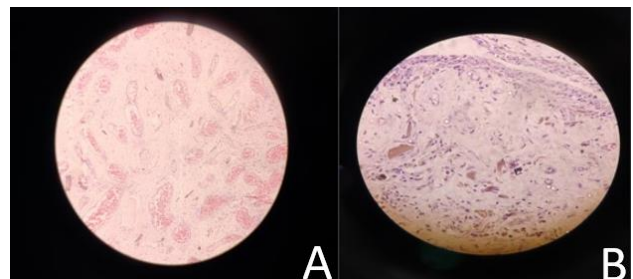
**Figure 3: Follow-up X-ray of pelvis A-P view with total hip replacement right (uncemented).**

In the clinical examination of the right hip, we found limited ROM and pain, especially in rotational movements. Radiographical suggestive of severe degenerative arthritis was present in the right hip and B/L knee. During the surgery through a directly posterior approach, we noticed that the hip capsule was thick, contracted, and black with severe pericapsular adhesions. After capsulotomy, in the absence of infection and abnormal preoperative evaluations with increased erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP), we decided to proceed with arthroplasty. Due to the severe adhesions and contracture, we had to perform an extensive soft tissue release to have the desired exposure. An osteotomy was performed in the femoral neck and the black femoral head was removed (Figure 2). By the same posterior approach, femoral and acetabular bone components were inserted without any complications (Figure 3).

After the surgery, the patient was examined carefully for other symptoms of the disease. At this time, multiple pigmentations were found in the eyes, ears, and hands (Figure 4).



**Figure 4: Dark pigmentation of (A) palm of hand (B and C) ears lobes (D) sclera of eyes.**



**Figure 5: Histopathology slide under (A) 10x (B) 40x.**

In the histological study with hematoxylin and eosin (H and E), multiple sections examined form containers A and B grey white soft tissue pieces show multiple fragments of

articulated cartilage along with hypertrophied synovial tissue. Sub-synovial tissue showed marked chronic inflammatory cell infiltration along with lymphocytes, plasma cells, and foreign body giant cell reaction. There were multiple areas of the new bone formation known as osteochondral bodies identified. The surrounding stroma showed hemosiderin and chronic pigment laden macrophages, inflammatory cell infiltrates, and neovascularization (Figure 5).

Her pain significantly decreased after the surgery. After 2 years of follow-up, the range of motion was improved and acceptable in the right hips without any pain or discomfort.

## DISCUSSION

Ochronotic arthropathy is a late complication of AKU that leads to cartilage damage and degenerative changes. It is asymptomatic in most cases until the fourth decade of life, and normally the disease is diagnosed by intraoperative findings.<sup>6</sup> Degenerative joint diseases are usually managed conservatively in the early stages and surgically in severe cases.<sup>7</sup> Our patient was managed with bilateral total hip arthroplasty due to mechanical pain and degenerative changes in both hips. Ochronosis can lead to cardiovascular and renal diseases, putting patients with AKU at a higher risk of general anaesthesia and surgical complications. Furthermore, in our experience, this condition can be confused with infection intra-operatively. Thus, it is important to diagnose this condition before the surgery and conduct the appropriate preoperative evaluations to avoid complications. Surgeons must be vigilant in the general examination for clinical clues such as pigmentation in the eyes, ears, hands, and skin folds, which were present in our case, and urine color which turned black when exposed to air.<sup>8,9</sup> During the surgery, we observed soft tissue and capsular contracture in our patient, which was in line with other reports.<sup>10</sup> It seemed that this finding could be an important intraoperative clue to guide the surgeon. Additionally, extra caution should be taken while providing care for these patients since this condition may cause osteoporosis and the patients may be vulnerable to intraoperative or postoperative fractures.<sup>11</sup> Due to the severity of the degenerative changes, we managed our patient successfully with total hip arthroplasty. In accordance with our results, Couto et al showed that arthroplasty was a safe and effective option to treat ochronotic arthritis.<sup>12</sup> Similarly, Pachore et al reported an excellent outcome for total hip arthroplasty in a patient with a severe degenerative joint disorder secondary to ochronosis. They also mentioned that joint replacement in ochronotic arthritis was associated with prosthetic survival comparable to that of patients with primary osteoarthritis. They highlighted the fact that orthopaedic surgeons should expect an atypical arthropathy when they see darkened cartilage during surgery.<sup>13</sup> Although this rare condition had been discussed in a few case reports, the long-term outcomes of joint arthroplasty in ochronotic arthritis should be evaluated in further studies.

## CONCLUSION

Ochronotic arthropathy is a very rare disorder that can be potentially misdiagnosed as osteoarthritis in patients with joint pain and radiographic joint space narrowing. We believe that a total hip arthroplasty is a good option for patients with severe degenerative changes due to ochronotic arthritis.

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