

## **Case Report**

### **Ochronosis- The Role of Total Hip Replacement: A Case Report and Review of Literature**

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**Abstract:** Alkaptonuria is a rare autosomal recessive, single-gene metabolic disease caused by the lack of the enzyme homogentisic acid oxidase and characterized by black pigmentation of cartilage and other connective tissues. 38 yr old male was admitted to the orthopedics ward with bilateral hip pain for the past 3 years and inability to walk for the past 1yr. He had also complained of brownish-black discolouration of urine when exposed to air. Physical examination revealed flexion 70°, internal rotation and extension 0°, external rotation 30° in both the hips. Radiological findings showed joint space narrowing, acetabular protrusion, sclerosis and subchondral cysts. Total hip replacement was performed on the right hip followed by the left. Histopathology showed brownish black staining of degenerated hyaline articular cartilage. Six months after the surgery, physical examination of the patient showed excellent results. There were no significant complaints. In our patient total hip replacement proved to be an excellent treatment for his ochronotic hips. Total joint arthroplasty is an effective method in the treatment of late stage osteoarthritis due to ochronosis and the technique of in cases of ochronotic arthropathy is similar to osteoarthritis due to any other etiology.

**Keywords:** Alkaptonuria, Total Hip Replacement, Homogentisic acid oxidase, Osteoarthritis

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## **INTRODUCTION**

Alkaptonuria is a rare autosomal recessive single-gene metabolic disease caused by the lack of the enzyme homogentisic acid oxidase and characterized by black pigmentation of cartilage and other connective tissues. Diet restriction is the only medical treatment available and surgery is only reserved for late stage joint disease [1].

We present one male patient, with age 38 years, who underwent staged bilateral uncemented total hip arthroplasty, for osteoarthritis caused by ochronosis. The gross pathological findings of the surgical specimens showed extensive degeneration of articular cartilage with black colored staining of the remaining articular cartilage and synovium. Microscopic examination showed brown-black pigment deposits in the synovium and the cartilage.

## **CASE REPORT**

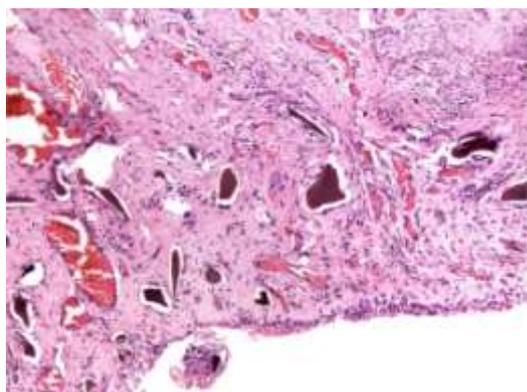
38 yr old male was admitted to the orthopedics ward with bilateral hip pain for the past 3 years and inability to walk for the past one year. He was not able to get relief with medication or physiotherapy. He had also complained of brownish-black discolouration of

urine when exposed to air. There was not any kind of significant family history. Physical examination revealed flexion 70°, internal rotation and extension 0°, external rotation 30° in both the hip joints. Examination of all other joints was grossly normal. Radiological findings showed joint space narrowing, acetabular protrusion, sclerosis and subchondral cysts. Blood cell counts and other routine investigations were normal. Rheumatoid factor and the HLA-B27 antigen were negative. The patient was planned for surgery in two stages. Initially, total hip replacement was planned on the right hip. During the surgery, there was no difficulty experienced despite the significant protrusion-acetabuli. However, there was significant blood loss intraoperatively. It was noticed that femoral head, acetabulum and joint capsule were pigmented (black) (Fig. 1). An uncemented total hip arthroplasty was performed in the same standard manner. The samples taken during the time of surgery were sent for histopathological evaluation, which showed brownish black staining of degenerated hyaline articular cartilage (Fig. 2). It also showed yellow auto fluorescence and stained positively with Schmorl's stain.

Other changes include fibrous and fibrocartilagenous repair on the articular surface; and osteophyte formation. In the synovium, chronic inflammatory cell infiltrates composed of lymphocytes and macrophages, were seen in response to the pigment deposition. The patient was diagnosed with ochronosis on the basis of clinical, radiographical and histopathological findings. Three weeks later, an uncemented total hip replacement was performed on the left hip too, with a similar intra-operative picture (Fig. 3).



**Fig. 1: Intra-operative gross picture of pigmented hip**



**Fig. 2: Microscopic examination showing brown-black pigment deposits**



**Fig. 3: Postoperative (bilateral hip replacement) X-ray**

Six months postoperatively, the patient was examined in the OPD. Physical examination showed a wellpreserved range of motion and there were no specific complaints. There was no abnormality detected on the prosthetic components on plain radiographs.

## DISCUSSION

Alkaptonuria is rare inherited genetic disorder, in which deficiency of homogentisic acid oxidase, leads to excretion of large amounts of homogentisic acid in urine and accumulation of oxidized homogentisic acid pigment in connective tissues. Usually at around 30 yrs of age, subjects develop generalized darkening of ear lobes, sclera and nose due to pigment deposition. Low back pain usually starts between 30 and 40 years of age followed by degenerative joint disease [2]. In our patient there was no history of any back pain. Pain, stiffness, and some limitation of motion of the hips, knees, and shoulders are the common complaints of ochronoticarthropathy. Small joints are usually spared. Our patient was not aware of the darkening on his eyes and ears. But, he had a history of discoloration urine on exposure to air. The disorder can be seen in family members as well although it is seen rarely. There was no specific occurrence of similar illness in the rest of the family members.

Other systems involved in ochronosis include cardiovascular and genitourinary systems; common features being, cardiac valve calcification and stenosis, coronary artery calcifications; and renal and prostatic stones. In our patient, these systems were not involved.

Ochronotic patients commonly complain of low back pain and stiffness of the spine. Calcification of multiple intervertebral discs, obliteration of the intervertebral disc spaces, sclerosis of the vertebral plates and osteoporosis of the vertebral bodies are the usual characteristic features of ochronotic spondylopathy [3-5]. In our patient, low back pain was not on the foreground. Involvement of the large peripheral joints usually occurs several years after the spinal changes. The knee is the most common site of peripheral abnormality. Other sites of involvement are the hips, shoulders, sacroiliac joints and symphysis pubis.

In the hip joint, the complaints are similar to that of any degenerative arthritis. Radiological changes seen include sclerosis, narrowing of the joint space, subchondral cysts. Osteophytic changes are not as evident as in osteoarthritis (as in our patient). Of these, the hip joint changes were the most striking. The brownish black discoloration of the articular cartilage and capsule observed during the time of surgery was typical.

There is no specific treatment for Alkaptonuria. Contrary to the previous thinking,

ascorbic acid and protein restriction are not effective in reducing homogentisic acid production [6-8]. For the joint disease, the usual anti-arthritis drugs and physiotherapy are useful in the early stage of the disease, but has little effect on the actual progression of the rate of degeneration of joints. In the late stages of the disease, joint replacements are often inevitable. In our patient, total hip replacement proved to be an excellent treatment for his ochronotic hips.

#### **CONCLUSION**

Total joint arthroplasty is an effective method in the treatment of late stage osteoarthritis due to ochronosis; and the procedure and technique of arthroplasty in cases of ochronotic arthropathy, are similar to osteoarthritis due to any other etiology.

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