

A Case Series of Ochronotic Arthritis

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Abstract: Ochronotic arthropathy is a rare hereditary metabolic disease found in patients with alkaptonuria. It is associated with deposition of homogentisic acid derivatives in various connective tissues of the body. Joint involvement especially hip and knee destruction is seen. We present two cases of alkaptonuria resulting in ochronotic hip and knee arthritis treated with total hip and knee arthroplasties. Patients complaints were alleviated and no further complaints were registered, during the next follow-up.

Keywords: Alkaptonuria ochronosis, joint replacement.

CASE REPORT 1

A 58 years old female presented to the emergency department of our hospital with complaints of pain in her right hip after a simple fall, and not able to bear weight on her leg. The anteroposterior radiography of hip joint showed a fracture of neck of the femur (Figure 1). Because of a lucency at the base of femur neck, we came to the conclusion that a pathologic fracture had occurred at that region. She had no other underlying disease except a chronic low back and hip pain. Patient was taken up for Total Hip Replacement. After incision of skin and subcutaneous fat, there was a dark black lesion in the deep fascia and proximal portion of tensor fascia lata which had extended to deeper structures and joint capsule of hip joint (Figure 2) we sent the sample of the black tissue for histopathological examination which revealed ochronotic arthritis (Figure 3). Pt was advised about how to walk with crutches for two weeks following surgery (Figure 4). Patient condition was improved, she was able to walk with the help of walking aids.



Fig-1: Xray of the hip joint showing AP view



Fig-2: Excised head of femur showing black discoloration

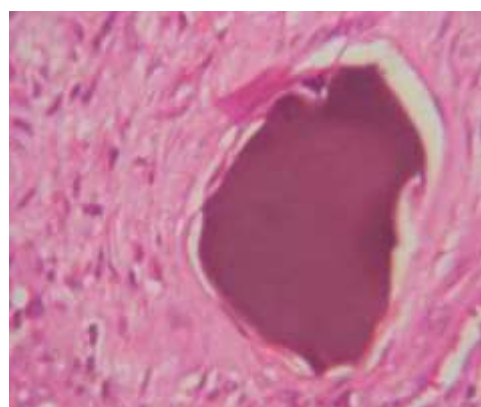


Fig-3: 40 x view showing deposition of brownish material



Fig-4: Xray of the hip joint showing acetabular cup fixed with screw

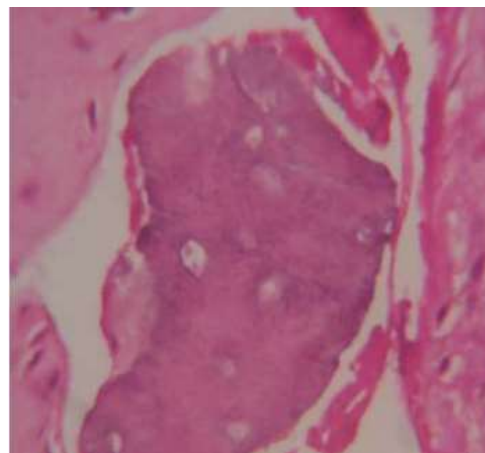


Fig-7:40 x view showing deposition of brownish amorphous material



Fig-5: Xray knee joint showing Lateral view



Fig-8: Xray showing implant in situ of the knee joint



Fig-6: Intra-op blackish discoloration over the patella

CASE REPORT 2

A 71 year old male came to Orthopaedics department with complaints of multiple joints pain and lowback ache for past 1 year. Pain was nontraumatic, progressive, dull-aching in nature, and aggravated on exertion. He had been taking nonsteroidal anti-inflammatory drugs and muscle relaxants for the last 1 year with temporary pain relief. X-rays of the cervical and lumbosacral spine showed advanced degenerative changes with intervertebral disk space narrowing and osteophyte formation. There were disk calcifications visible in the lumbosacral spine. X-rays of the knee joints showed reduced joint space, osteophyte formation with loose bodies, suggestive of osteoarthritis knees (Figure 5). Patient was taken up for total knee replacement. After incision of skin and subcutaneous fat, there was a dark black lesion in the deep fascia and patella and distal part of femur which had extended to deeper structures and joint capsule of knee joint (Figure 6). We sent the sample of the black tissue for histopathological examination which revealed ochronotic arthritis (Figure 7). Patient condition was improved; he was able to walk with the help of walking aids.

DISCUSSION

Alkaptonuria is a rare disorder with an incidence of 1:125,000 to 1:1 million worldwide [1-4].

It was one of the first conditions in which the law of mendelian recessive inheritance was proposed and one of the conditions in the group of inborn errors of metabolism. The term ochronosis was first coined by Virchow in 1866 [5] when he found pigmentation of tissues that appeared ochre, meaning yellow, when examined microscopically [6]. In alkaptonuria, ochronotic pigment is deposited in all connective tissues, especially cartilage. These pigmentary changes are termed ochronosis. Ochronosis can occur in ligaments, tendons, heart valves, the intima of blood vessels, sclera, and the skin [3]. The majority of alkaptonuria symptoms are not detected until the fourth decade [3,7-9].

CONCLUSION

The management of ochronotic arthropathy in alkaptonuria patients is usually conservative, but replacement surgery is recommended for severely affected hip and knee joints. This report describes two cases of ochronotic hip and knee arthritis treated with total hip and knee arthroplasties. Since alkaptonuria is a rare disease, it may be beneficial to pre-screen patients with degenerative disease for signs of ochronosis, so that an early diagnosis can be made and an effective conservative management can be started to improve the quality of life.

At follow-up of 6 months, both patients with total hip and knee replacement are normal. In these patients, morbidity and complications are significantly decreased by early diagnosis and management.

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