

A RARE CASE REPORT OF OCHRONOTIC ARTHRITIS WITH BILATERAL HIP INVOLVEMENT***Dr. Mary Lilly and Dr. Susan D. P.**

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ABSTRACT

Alkaptonuric ochronosis is a rare autosomal recessive disorder of metabolism caused due to deficiency of homogentisic acid oxidase synthetase. Alkaptonuria leads to chronic inflammation, degeneration, and finally osteoarthritis. Ochronotic arthropathy is a rare condition caused in alkaptonuric patients. This case report discusses about a 64-year-old woman with alkaptonuric ochronosis who suffered from severe degenerative arthritis of left hip with a history of right hip ochronosis. Here we describe alkaptonuric ochronosis involving bilateral hip joints. This paper also involves the understanding of pathological epidemiology of bilateral hip involvement due to ochronosis.

KEYWORDS: Ochronosis; Alkaptonuria; Bilateral hip, arthropathy, arthritis, epidemiology.**INTRODUCTION**

Alkaptonuria is an autosomal recessive disorder of metabolism caused due to deficiency of homogentisic acid oxidase synthetase which leads to homogentisic acid deposition in connective tissue. The main factor for catabolism of tyrosine and phenylalanine is homogentisic acid oxidase.

Degenerative changes in the morphologic structure of connective tissue, resulting in fragile complexes happens due to the irreversible binding of the homopolymeric oxidation products of HGA to collagen. Ochronosis is deposition of ochronotic pigment in connective tissues. Features of alkaptonuric ochronosis includes chronic inflammation, arthritis, degeneration, coronary artery disease and renal diseases. Patients with alkaptonuria tend to have a rare condition called Ochronotic arthropathy.

Knee is the most commonly affected joint followed by sacroiliac joints, lumbar intervertebral discs, hips and shoulders. Symptoms can be managed medically but there is no known cure for ochronosis. Cases with significant degenerative arthropathy are treated by joint replacement.

CASE REPORT

A 64-year-old came with the complaints of pain in the left hip which was progressive in nature and got aggravated on movements and relieved on rest since 7 months.

On local examination of the left hip, there was tenderness present over the Scarpa's triangle with no warmth or swelling. Range of movement was restricted and was painful with sensation intact. No DNVD present. Modified Schober's test was positive. Radiological findings showed severe degenerative arthritis of left hip with protrusion acetabuli. Clinically, provisional diagnosis was made as Ankylosing Spondylitis.

Total hip replacement of left hip was done and the femoral head was sent for histopathological examination.



Fig. 1: Total hip replacement done in right hip joint four years back.



Fig. 2: Total hip replacement done now in left hip joint as well.

Four years back, there was a history of low back pain and right hip pain for which total hip replacement was done on the right hip and grossly the head of femur had blackish degeneration and old records revealed that the patient underwent surgery and biopsy was done and the histopathological diagnosis was Ochronotic arthritis.

Gross Examination of Left hip: There was a specimen of femoral head with irregular external articular surface. Multiple grey white nodules and blackish discoloured patches noted at focal areas on the external surface. The specimen was decalcified. After decalcification, the cut section showed irregular articular cartilage and rest of the cut section was unremarkable. Tissue samples were fixed in 10% buffered neutral formalin and embedded in paraffin. Sections were stained with haematoxylin and eosin.

Gross appearance



Fig. 3: Femoral head with irregular external articular surface. Multiple grey white nodules and blackish discoloured batches noted at focal areas.



Fig. 4: Cut section showed irregular articular cartilage and rest of the cut section was unremarkable.

Microscopic appearance

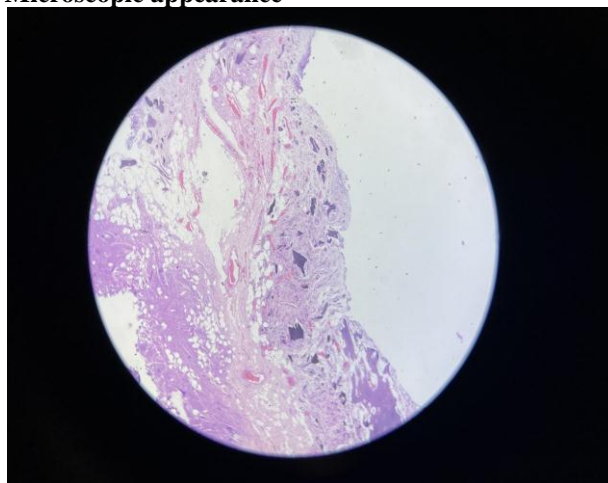


Fig. 5: Spicules of bone with brown pigmentation and acellular patches of yellowish brown on hematoxylin and eosin.

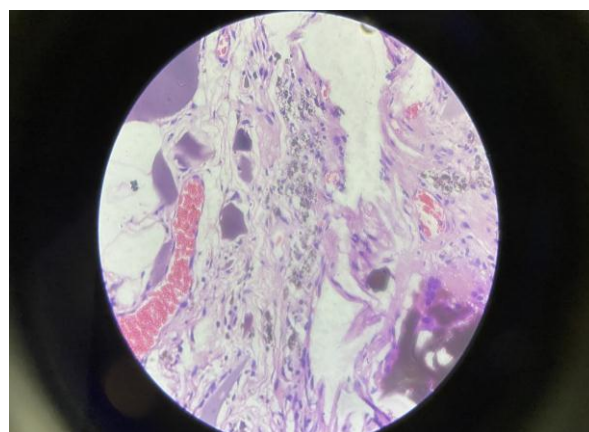


Fig 6: Spicules of bone with brown pigmentation and acellular patches of yellowish brown on hematoxylin and eosin, 400x.

Histologically, there were spicules of lamellar bone with intervening hypocellular marrow. Spicules of bone with brown pigmentation surrounded by normal lamellar bone were seen from the sections of the articular surface.

Adherent soft tissue also showed spicules of bone with brown pigmentation. Inflammation was scanty. It was reported as Ochronotic Arthritis.

DISCUSSION

Alkaptonuria affects 1 in 25,000 to 100,000 people in India and it is a rare occurrence. This is caused by deficiency in the homogentisic acid oxidase enzyme (HGAO). Homogentisic acid (HGA) degrades due to HGAO enzyme. This enzyme deficiency causes homogentisate polymers to deposit in skin, cartilage and results in urine darkening, brown black pigmentation of connective tissue, articular cartilage pathology, osteoporosis, and pathomorphologic changes in internal organs.

Pathologically, in joints, ochronotic pigment is characteristically deposited in the deeper layers of the articular cartilage and mostly in older cartilages with poor metabolism. In vertebral columns, the intervertebral discs get pigmented and eventually calcified, causing marginal osteophytosis of the vertebral bodies. In extreme cases, the vertebral column may become rigid, looking like the bamboo spine of ankylosing spondylitis. The other connective tissues of the spinal column and its joints may similarly be affected.

Histopathologically, the gross appearance of the tissue involved differs with the severity of the lesion, depending on the tissue involved and the age of the patient. The hallmark feature is tissue pigmentation, which may vary from brown to black.

Microscopically in the hyaline cartilage, there is deposition of ochre-coloured granules in all layers, but most distinctly in the deeper layers and the cartilage cells are small and pyknotic. Fibrocartilage may show areas of hyaline degeneration and calcification. Perichondrium may become thickened and pigmented. Synovium may show thickening and granule deposition, and there may be calcification and small islands of cartilage.

The majority of alkaptonuria symptoms can only be noticed after fourth decade. There is no known medical treatment for alkaptonuria.

We performed Total Hip replacement of the hip joint and the patient was given standard physical therapy with satisfactory results.

Ochronotic arthropathy is a sequel of alkaptonuria, affecting large weight-bearing joints. Ochronotic arthropathy is usually diagnosed only intraoperatively during a total joint replacement when dark synovium and cartilaginous surfaces are seen. In the present case it was revealed during the first hip replacement. Joint replacement is usually done for patients with severe degenerative arthropathy. As alkaptonuria is a metabolic disorder of the bones and joints, it affects the mechanical properties of connective tissue, especially the cartilage,

and decreases bone quality. To date, only a few case reports with short-term follow-up have described arthroplasty in ochronosis.

The female patient in our study underwent bilateral hip replacements, and no complications were encountered during the first hip replacement surgery. Her condition improved significantly, and she returned to activities of daily life without experiencing any pain. The present study demonstrates that arthroplasty can have excellent outcomes and great prosthesis survival in alkaptonuric patients with ochronotic arthritis, supporting the findings of previously published case reports.

Biochemical analysis of urine plays a very important role in preoperative diagnosis. Pathological examination plays a crucial role in the diagnosis of Ochronosis as no specific diagnosis is available in patients presenting with pure arthropathy like our case.

CONCLUSION

Since there is no effective treatment for ochronosis, only the symptoms can be alleviated. Therefore, the epidemiology of this disease is to be considered so that it would be a leading factor for pre-screening patients with this degenerative disease and help us in the diagnosis of pathology. Biochemical and pathological examination are required to confirm the diagnosis so that clinicians can treat well postoperatively. Hip replacement has shown good outcomes in ochronotic arthropathy.

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