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Ochronotic Arthropathy Presenting as Bilateral Hip Osteoarthritis with Right Femoral Head Necrosis: A Case Report

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Abstract: ***Alkaptonuria is an autosomal recessive metabolic disorder caused by homogentisate 1,2-dioxygenase deficiency. It leads to the accumulation of homogentisic acid in connective tissues, resulting in a condition known as ochronosis. We present a case of a 50-year-old female with a known history of ochronosis who presented with bilateral hip pain and walking difficulty. She reported a history of passing black-colored urine since childhood. Imaging revealed bilateral hip osteoarthritis with right femoral head necrosis, bilateral acetabular ischemia, and protrusion acetabuli. The patient underwent a right-sided total hip replacement. Histopathology confirmed the presence of pigmented ochronotic bodies and a foreign body giant cell reaction. Joint replacement remains a highly effective treatment for end-stage ochronotic arthropathy.***

Keywords: Alkaptonuria, Ochronosis, Ochronotic Arthropathy, Total Hip Replacement

1. Introduction

Alkaptonuria is a rare, autosomal recessive metabolic disease affecting tyrosine and phenylalanine metabolism. It occurs due to a deficiency in the homogentisate 1,2-dioxygenase enzyme [1]. This enzymatic deficiency leads to the accumulation of homogentisic acid (HGA) and its oxidized products in collagenous structures [2]. The classic clinical triad of the disease includes the excretion of excess HGA causing dark urine upon standing, dark pigmentation of connective tissues known as ochronosis, and rapidly progressive joint arthritis known as ochronotic arthropathy [1].

Ochronotic arthropathy most commonly involves the spine and large weight-bearing joints, such as the knees and hips. The accumulated pigment weakens the cartilage, making it brittle and susceptible to mechanical failure, which eventually leads to severe degenerative changes [2]. Because the condition is rare, the diagnosis is sometimes delayed or discovered incidentally during surgery when black intra-articular pigmentation is observed [3,4]. This report details a case of severe bilateral hip ochronotic arthropathy complicated by right femoral head necrosis, which was managed surgically with a total hip replacement.

2. Case Report

A 50-year-old female patient presented to the clinic with chief complaints of pain in her bilateral hip joints and difficulty in walking. Upon clinical history taking, she reported the passage of black-colored urine since childhood. The patient was already a known case of ochronosis.

Radiological investigations were performed to evaluate the extent of the joint disease. X-ray of the bilateral hips showed severe degenerative changes along with the disappearance of the joint space. Magnetic Resonance Imaging (MRI) demonstrated bilateral hip arthritis accompanied by aseptic necrosis of the right femoral head, edema of the left femoral head, and bilateral acetabular ischemia. The radiological impression concluded bilateral hip osteoarthritis with necrosis of the right femoral head. Correlating the clinical and radiological findings, the final diagnosis was established as bilateral hip osteoarthritis with protrusion acetabuli in a known case of ochronosis.

To manage the severe joint destruction, the patient underwent surgical intervention consisting of a right-sided total hip replacement.

Specimens retrieved during the surgery were sent for microscopic evaluation. Microscopy revealed multiple tissue fragments lined by synovial epithelium showing subepithelial capillary proliferation, hemorrhage, and a mild mononuclear infiltrate [Figure 1]. Pigmented ochronotic bodies were identified within the subepithelial tissue [Figure 2] . These were associated with aggregates of pigment-laden macrophages and a foreign body giant cell reaction [Figure 3]. Pigmentation was also notably present within the cartilage and fibrous connective tissue [Figure 4]. Dead bony spicules were observed in the specimen . The resected femoral head revealed occasional bony trabeculae separated by

fibroadipose tissue, along with focal hematopoietic marrow. Furthermore, occasional bony trabeculae and cartilage fragments exhibited characteristic brown-black discoloration

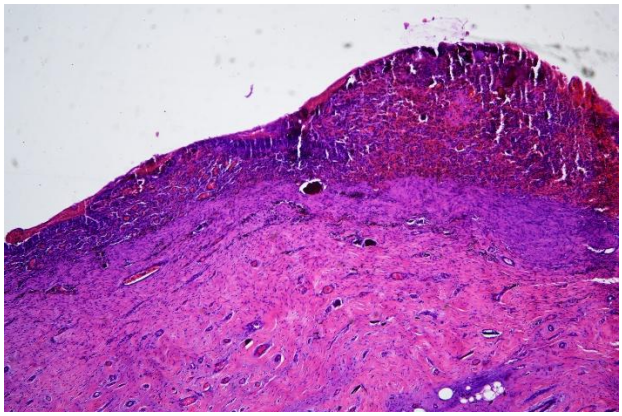

Figure 1

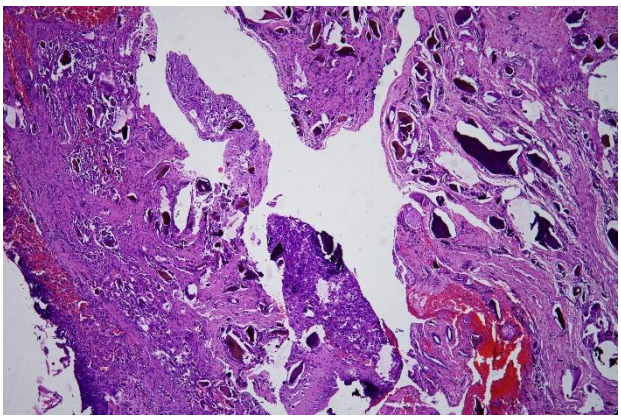

Figure 2

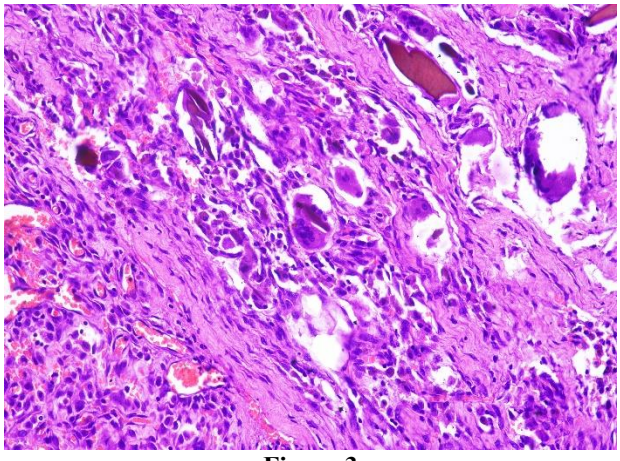

Figure 3

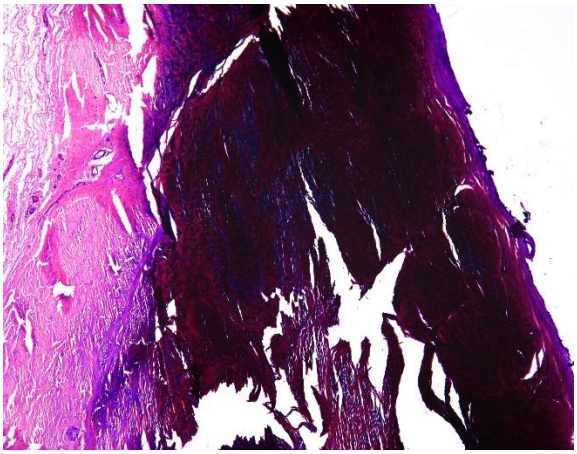

Figure 4

3. Discussion

Ochronosis is the tissue manifestation of alkaptonuria, characterized by the deposition of bluish-black or brown-yellow polymerized HGA pigments in connective tissues, particularly affecting hyaline cartilage [2,3]. Patients typically have a long-standing history of dark urine since childhood, which was a prominent historical feature in our patient [5].

The deposition of the ochronotic pigment profoundly alters the mechanical properties of the cartilage, rendering it brittle and significantly reducing its resistance to mechanical strain [2]. This degradation cascade results in advanced osteoarthritis, typically affecting large weight-bearing joints like the hips [3,4]. In our case, the patient developed severe bilateral hip osteoarthritis complicated by aseptic necrosis of the right femoral head and protrusion acetabuli.

Histopathologically, ochronosis is characterized by the presence of ochre or yellow-brown pigment deposited within collagen bundles and cartilage [6]. Our microscopic findings were entirely consistent with this presentation, demonstrating pigmented ochronotic bodies, distinct brown-black discoloration of the cartilage and bone trabeculae, and a robust foreign body giant cell reaction. The inflammatory response, characterized by macrophage aggregation and giant cell reactions around the deposited pigment, contributes further to ongoing joint destruction [4].

Advanced ochronotic arthropathy of the hip is a severely disabling condition with extremely limited non-surgical treatment options [2,3]. Conservative management generally relies on symptomatic relief using analgesics, as there is no definitive cure to reverse the structural damage once it has occurred [7]. For end-stage disease, such as the severe joint destruction and femoral head necrosis observed in this patient, joint replacement surgery remains the most effective intervention [2]. The existing literature supports that total joint arthroplasty provides excellent pain relief and functional restoration in patients suffering from advanced ochronotic arthropathy [2,3].

4. Conclusion

Ochronotic arthropathy is a rare, progressive, and highly debilitating complication of alkaptonuria [1]. The diagnosis should be strongly suspected in patients presenting with a history of dark urine and severe, early-onset joint degeneration [5]. Histopathological examination revealing characteristic pigmented bodies and foreign body giant cell reactions confirms the diagnosis. Total hip replacement provides a highly successful surgical solution for managing end-stage ochronotic arthritis associated with femoral head necrosis [2].

References

[1] Jirel A, Paul N, Kaganur R, Gopurathingal A, George J. Ochronotic hip arthropathy – A case report. Journal of Orthopaedic Case Reports. 2022;12(10):14-17.
[2] Salem KH, Elmoghazy AD. Ochronotic arthropathy: skeletal manifestations and orthopaedic treatment. EFORT Open Reviews. 2025; 10: 75-81.
[3] Yap San Min N, Rafi U, Wang J, He B, Fan L. Ochronotic arthropathy of bilateral hip joints: A case report. World Journal of Clinical Cases. 2023;11(1):210-217.
[4] Nair AV, Kiran V, Rajan A, Bharadwaj MS, Daware MA, Khan PS. Ochronotic Arthropathy of the Shoulder – A Rare Case Report. Journal of Orthopaedic Case Reports. 2025;15(1):90-93.
[5] Erhan SŞ, Keser SH, Gülsever K, Şensu S. Ochronotic Arthropathy: A Report of Two Cases. South. Clin. Ist. Euras. 2020;31(1):80-83.
[6] Vallonthaiel AG, Kakkar A, Singh MK, Ramam M. Ochronosis with subtle histological findings. Indian Journal of Dermatology, Venereology, and Leprology. 2015;81(6):623-624.
[7] de Lima DA, de Paiva DL, Bastos YRC, de Aquino Filho JC, Clazzer R, de Lima LL. Ochronotic Arthropathy and Alkaptonuria (Ochronosis): Case Report. Rev Bras Ortop. 2024;59(S2): e220-e223.