

Ochronotic arthropathy of the hip with multiple joint involvement: a rare case report and literature review

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Ochronosis is a rare inherited genetic disorder that affects tyrosine and phenylalanine metabolism. It leads to the deposition of dark pigments in connective tissues and articular cartilage, which are rich in collagen. This may lead to severe joint degeneration, known as end-stage arthropathy of the joints. In such cases, arthroplasty can offer significant improvement in function. We present a case of a 52-year-old man who was diagnosed with secondary osteoarthritis in his right hip. During the surgery, ochronosis was unexpectedly discovered. An uncemented total hip arthroplasty was carried out and yielded positive results. Additionally, this particular case exhibited widespread effects on both knees and the dorso-lumbar spine. This case demonstrates that total hip arthroplasty can successfully treat severe ochronotic arthropathy, resulting in functional outcomes comparable to those seen in primary osteoarthritis. However, early diagnosis is crucial. To achieve this, healthcare professionals must be aware of the condition, maintain a high index of suspicion, and conduct a thorough clinical examination.

Keywords:

arthropathy, homogentisic acid, metabolic disorder, ochronosis, pigmentation, total hip arthroplasty

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Introduction

Alkaptonuria is a rare genetic disorder affecting phenylalanine and tyrosine metabolism. It is caused by a deficiency of homogentisate 1,2-dioxygenase enzyme, leading to the accumulation of homogentisic acid (HGA) in the body [1]. Ochronotic pigment is created by the autoxidation and polymerization of HGA. This pigment is then deposited in collagen-rich tissues, including the articular cartilage, tendon, ligaments, skin, and kidneys [2]. The irreversible binding of ochronotic pigments to collagen causes degenerative changes connective tissue structure, leading to fragile and easily breakable tissues. This ultimately leads to end-stage arthritis, known as ochronotic arthropathy [3].

Clinical features of ochronosis include blue-black or gray-blue pigmentation of the skin, cartilage of the ears, and sclera of the eyes. The urine is normal in color when freshly passed, but it rapidly darkens to assume a brownish-black color on exposure to air. Articular involvement most commonly involves the knee joint, followed by the hip, shoulders, lumbar spine, and intervertebral discs, respectively [4]. Because of the condition's rarity, awareness of the disease, a thorough clinical examination, and a high index of suspicion are required to diagnose this condition [5]. Here, we present a case of ochronotic arthropathy of the hip, which was managed with uncemented total hip arthroplasty (THA).

Case presentation

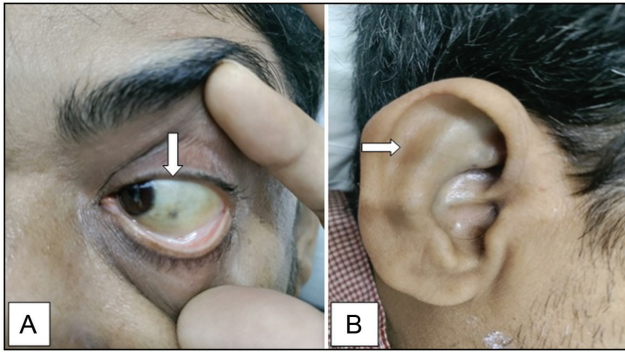
A 52-year-old male patient presented with complaints of right hip pain for the last year. It was dull, aching, gradually progressive, nonradiating, and localized to the hip region. It was relieved by taking rest and medications. He had difficulty squatting and climbing stairs. His symptoms had flared up in the past 5 months to the extent that he failed to perform daily activities.

He also complained of neck pain and low back pain for the last 5 years. In addition, both knees were painful due to arthritis with valgus deformities. There was also a history of recurrent episodes of urinary tract infection, which was managed with antibiotics.

On general examination, pallor, cyanosis, and icterus were absent. The sclera and the pinna had patchy brownish-black discoloration (Fig. 1). The anterior hip point was tender with flexion of up to 80° and painful restriction of internal and external rotation movements. The Harris hip score was 35 points. Bilateral valgus and flexion deformities were present

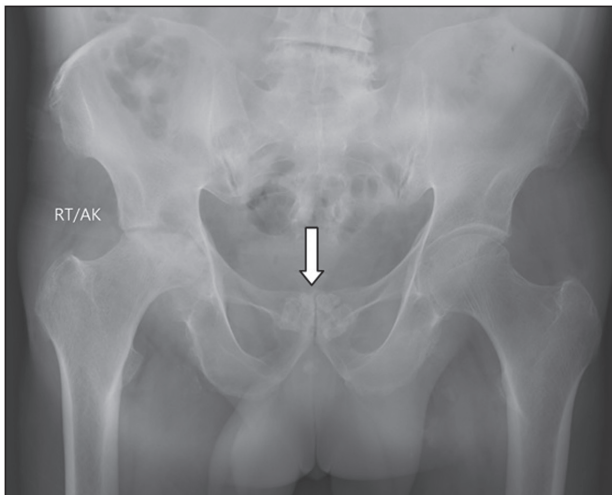
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Figure 1:



Clinical photographs showing (a) brownish-black spot noted in the sclera. (b) Brownish-black discoloration of the pinna.

Figure 2:



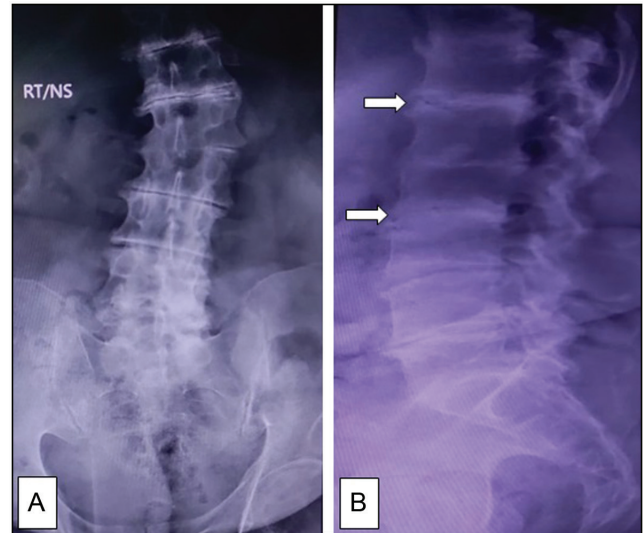
Plain radiograph showing pelvis with both hips (antero-posterior view) depicting destruction of the femoral head and subchondral sclerosis. The arrow depicts foci of calcification in the region of the urinary bladder and urethra.

in the knees. The spine was examined and found to be flexible and balanced.

Radiological examination revealed arthritic changes in both knee and hip joints. The right hip joint had decreased joint space and destruction of the femoral head with proximal migration. Chondrocalcinosis and subchondral sclerosis were also noted (Fig. 2). Also, multiple foci of calcification were noted in the region of the urinary bladder and urethra. These were later confirmed to be multiple brownish-black pigmented calculi by a cystoscopic examination that was done preoperatively.

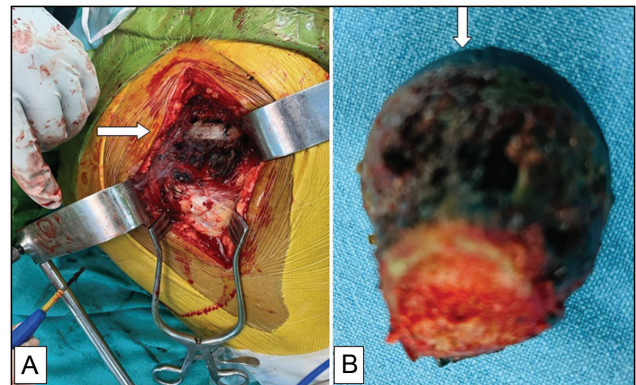
Radiograph of the lumbosacral spine revealed left lumbosacral scoliosis, mild loss of lumbar lordosis with multilevel degeneration, and calcification of the intervertebral discs. There was associated disc

Figure 3:



Plain radiograph of lumbosacral spine (a) antero-posterior view, (b) lateral views. Arrows depict intervertebral disc calcification and disc space narrowing. Osteoporosis and multiple vacuum phenomena are also noted.

Figure 4:



Intraoperative images (a) Black-stained periarticular tissues including tensor fascia lata, tendinous insertions and capsule. (b) The extracted femoral head with its black painted articular surface.

space narrowing, osteoporosis, and multiple vacuum phenomena (Fig. 3).

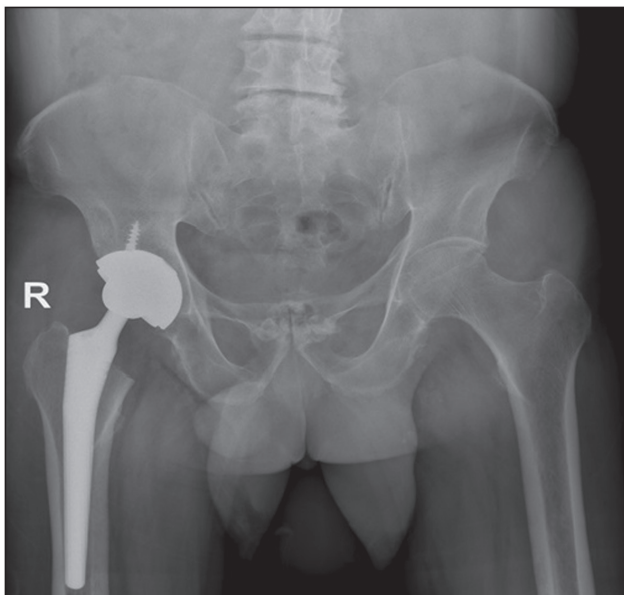
The patient underwent uncemented THA of the right hip using a modified Hardinge's lateral approach. Intraoperatively, tensor fascia lata, tendinous insertions, and periarticular tissue were stained brownish to black (Fig. 4).

The capsule was contracted and fibrosed, and the gluteus medius muscle was fragile. The acetabulum and its cartilage were also brownish-black in color. The head was extracted, and had a black painted articular surface which turned blacker over time. The pigmented

cartilaginous surface, bone, and hypertrophied synovium were sent for histopathological evaluation. Microscopic analysis of periarticular tissue, including the articular cartilage, revealed brown, banana-shaped, sickled, or rounded ochronotic bodies with homogenization, characteristic of ochronotic arthropathy (Fig. 5).

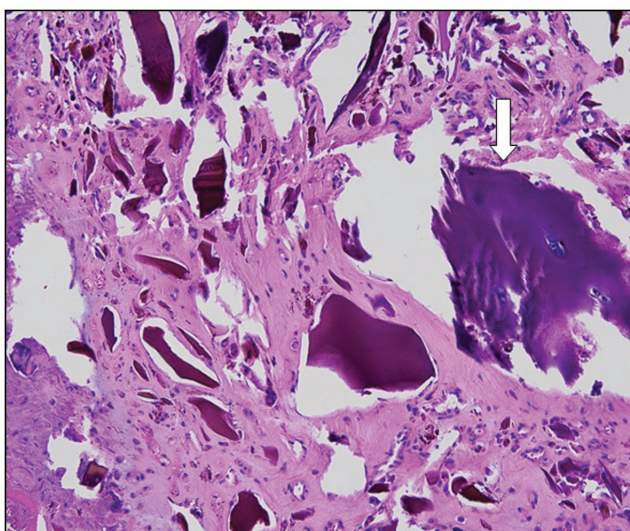
Hydroxyapatite-coated titanium acetabular cup and porous-coated uncemented femoral stem (Latitud; Meril, Vapi, Gujarat, India) were used for fixation (Fig. 6). Postoperatively, pain management included

Figure 5:



Postoperative plain radiograph (antero-posterior view) of the right hip showing an acetabular cup with a screw and a well-fixed uncemented femoral stem.

Figure 6:



Histological findings confirming ochronosis. The picture depicts banana-shaped ochronotic bodies in the articular cartilage tissue which are classical of ochronotic arthropathy.

tramadol and paracetamol. An abduction pillow was utilized, and hip precautions were explained to the patient. Aspirin 75 mg once daily was administered for DVT prophylaxis. Weight-bearing was permitted as tolerated, leading to an uneventful recovery for the patient. Sutures were removed on postoperative day 14, with home exercise and physical therapy recommended. By 3 months postsurgery, the patient had returned to baseline activity. At 6 months, gait was normal, and right hip pain was minimal, with a Harris score of 88 points (preoperative Harris hip score of 35). Regular follow-up was emphasized given to the patient for monitoring the progress in future.

Discussion

In ochronotic arthropathy, the presence of HGA can affect not only the knees but also other joints that have collagen-rich connective tissues [6]. This could explain why our patient has a medical history of cervical spondylosis, low back pain, and involvement of multiple joints. It is worth noting that individuals with ochronotic arthropathy experience chronic joint pain at an earlier age (between 30 and 40 years) compared to primary osteoarthritis, which typically occurs between 50 and 60 years of age [7]. This suggests that this metabolic disorder is linked to accelerated degeneration of cartilage. Additionally, these patients' average age for arthroplasty is 53 years [7]. In our study, the patient also underwent THA at the age of 52 years.

Table 1 presents a summary of the published studies on arthroplasty in ochronotic arthropathy. In a short-term follow-up of 6 months, Yap San Min *et al.* [10] concluded that THA in ochronotic arthropathy produces excellent results in terms of Harris hip score. Ilyas *et al.* [15] reported a case involving staged bilateral THA and total knee arthroplasty (TKA) and found significant improvement in Harris hip score and range of motion after 12 years. Bhattacharjee *et al.* [8] concluded from a case report that the use of dual mobility cups in THA for ochronotic arthropathy is a potential option, but since our case did not have the specific indications (like severe hip dysplasia, neuromuscular and spino-pelvic imbalance) for their use, we chose not to use them.

The role of capsular excision during THA is still a subject of debate. Cebesoy *et al.* [19] suggested removing the contracted anterior capsule to prevent dislocation, while Fernando *et al.* [18] recommended complete removal of the capsule to prevent disease recurrence. Pachore *et al.* [16] published their results of arthroplasty in a case series of 12 ochronotic hips and advised against

Table 1: Review details of the published case reports on total joint replacements in ochronotic arthropathy

Serial numbers	References	Journal	Year	Number of cases	Joint involved	Sex	Surgery	Outcomes assessment	Remarks
1.	Bhattacharjee <i>et al.</i> [8]	<i>J Orthop Case Rep</i>	2023	1	Hip	M	THA	HHS and ROM	Dual mobility cup in THA is a potential option in ochronotic arthropathy
2.	Liu <i>et al.</i> [7]	<i>Front Med (Lausanne)</i>	2023	1	Hip and knee	M	THA and TKA	VAS, FJS, HHS and KSS	Reduction in pain and improved satisfaction levels was present after TKA and THA
3.	Arora <i>et al.</i> [9]	<i>Cureus</i>	2023	2	Knee	M	TKA	ROM	TKA is useful in midterm 5 year follow-up
4.	Yap San Min <i>et al.</i> [10]	<i>World J Clin Cases</i>	2023	1	Both hips	M	THA	HHS and ROM	THA useful in ochronotic arthropathy. 6 month follow-up
5.	Kang <i>et al.</i> [11]	<i>J Bone Metab</i>	2023	1	Both knees	M	TKA	ROM	Successful outcome even in bilateral TKA
6.	Lazaro <i>et al.</i> [12]	<i>Cureus</i>	2023	1	Knee	M	TKA	ROM	Bilateral patellar tendon rupture following TKA in ochronotic arthropathy patient. A single-stage revision TKA with direct repair of the extensor mechanism was performed
7.	Butterfield <i>et al.</i> [13]	<i>J Surg Case Rep</i>	2022	1	Shoulder	M	RSA	Shoulder ROM	Reverse shoulder arthroplasty is an effective procedure for alleviation of joint pain and mobility improvement in patients with ochronotic arthropathy
8.	Al Dosari <i>et al.</i> [14]	<i>International Journal of Surgery Case Reports (IJSCR)</i>	2020	1	Knee	M	TKA	KOOS-PS score	Robotic TKA in ochronotic arthropathy gives good results. good functional results, pain improvement and patient satisfaction at 1 year
9.	Ilyas <i>et al.</i> [15]	<i>Arthroplasty Today</i>	2020	1	Hip and knee	M	Staged b/I THA and TKA	HHS and ROM	12 year follow-up
10.	Pachore <i>et al.</i> [16]	<i>Arthroplasty</i>	2019	10	Hip	M/F	THA	HHS and ROM	3-24 year follow up
11.	Lee <i>et al.</i> [17]	<i>Case Rep Orthop</i>	2019	1	Knee	M	TKA	OKS, KSKS and ROM	Improvement in OKS, KSKS and ROM with 2 year follow up
12.	Fernando <i>et al.</i> [18]	<i>J Orthop Case Reports</i>	2018	1	Hip	F	THA	HHS and ROM	Complete capsulotomy is useful
13.	Harun <i>et al.</i> [3]	<i>International Journal of Surgery Case Reports</i>	2014	2	Hip and knee	M	THA, TKA	ROM	Joint replacement gives excellent outcome in patients with degenerative ochronotic arthritis
14.	Cebesoy <i>et al.</i> [19]	<i>The American Journal of Case Reports</i>	2014	1	Hip	M	THA	HHS, ROM	Capsulotomy should preferably be done during THA in ochronotic arthritis
15.	Spencer <i>et al.</i> [20]	<i>Acta OrthopaedicaScandinavica, 2004</i>	2004	11 joints on 3 patients	Hip, knee, shoulder	M/F	THA, TKA	ROM	Follow-up of 12 years concluded that the survival rate of prosthesis is comparable to that of Arthroplasty done for other cases

F, female; FJS, forgotten joint score; HHS, Harris hip score; KOOS-PS, knee injury and osteoarthritis outcome score physical function short form; KSKS, knee society knee score; KSS, knee society score; M, male; OKS, Oxford knee score; ROM, range of motion; RSA, reverse shoulder arthroplasty; THA, total hip arthroplasty; TKA, total knee arthroplasty; VAS, visual analog scale.

total capsulectomy due to increased blood loss, instead advocating for its complete closure. In our practice, we routinely perform subtotal capsulectomy in all THA

procedures, including this case, as we believe that the contracted pathological capsule may impede adequate acetabular exposure and satisfactory cup placement.

Pachore *et al.* [16] suggested the use of screws with the cup in uncemented THA due to the sclerosis of the acetabular rim. They observed that uncemented THA in ochronotic hips provided superior pain relief, functional restoration, and no radiological evidence of loosening, instability, or subsidence.

In their study, Spencer *et al.* [20] performed 11 different arthroplasties in three ochronotic patients and followed up for 12 years. They concluded that the survival rate of the prosthesis in ochronotic patients is comparable to that in patients undergoing arthroplasty for other indications. Arora *et al.* [9] conducted a 5-year follow-up after TKA in their patients and reported positive outcomes and also found that bilateral TKA in these patients yielded good results [11]. Harun *et al.* [3] performed both cemented TKA and uncemented THA in a patient with ochronosis and concluded that arthroplasty provides excellent outcomes for patients with degenerative arthritis caused by ochronosis.

Short-term follow-up studies have shown favorable outcomes of THA and TKA in patients with ochronotic arthropathy. This condition can be an unexpected finding during surgery, as in our case. Current literature suggests that THA can be an effective intervention for pain relief and functional restoration. However, due to the limited number of cases, it is necessary to accumulate and pool more cases and conduct long-term follow-up studies. Additionally, further research is needed to establish optimal management strategies, including exploring the genetic basis of the disorder for potential targeted therapies.

Comprehensive patient care for ochronotic arthropathy requires a multidisciplinary approach involving orthopedic surgeons and metabolic specialists. Our case report highlights the importance of clinicians being familiar with this rare condition. Therefore, accurate and detailed history taking, including personal and family histories and a meticulous physical examination, are crucial.

Conclusion

There is a lack of extensive published case series or clinical guidelines for managing ochronotic hip arthropathy, making it a rare condition. At present, there is no known definitive cure for ochronosis, and the primary focus of management is to provide relief from symptoms and improve functionality. In severe cases of ochronotic arthropathy, arthroplasty is considered a treatment option to restore joint function and alleviate pain. This particular case report indicates that THA can be performed safely in severe ochronotic arthropathy without further complications or difficulties.

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Conflicts of interest

There are no conflicts of interest.

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