*Case Report*

# Fracture neck of femur with alkaptonuria: A case report

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## 1. Background

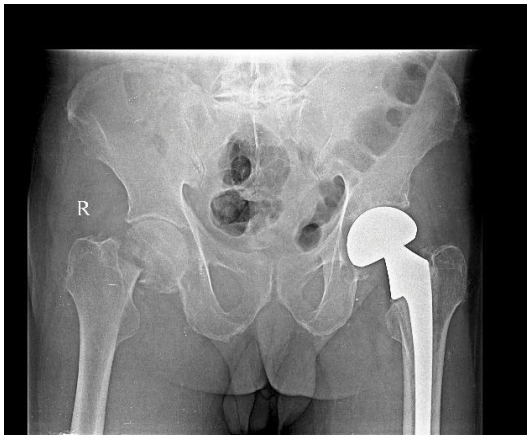
### 1.1 Abstract

Fracture neck of femur in younger age group (<60 years) poses a dilemma in terms of surgical management. The aim of treatment is early mobilization of the patient after surgery to prevent the development of complications. Major trauma is the leading cause of fracture neck of the femur. However, pathological fractures of the neck of femur have to be kept in mind. One cause of the latter is alkaptonuria, and the orthopaedic manifestations of the disease are outlined here.

## 2. Case Presentation

We present a 55-year-old male patient, who presented with acute onset of pain in his hip following a fall after tripping while walking. He had history of right hip pain since the past two years. His diabetes mellitus was well controlled on regular treatment. He had undergone a contralateral cemented bipolar hemiarthroplasty for a left sided traumatic fracture neck of femur sustained 5 years ago.

On examination, there was external rotation and shortening of the right lower limb with painful hip movements, inability to do active SLR and localised joint line tenderness. X ray showed a degenerative right hip with a fracture neck of femur [Fig 1]. Apart from the localised hip findings, the patient also had genu varum bilaterally, and minimally painful bilateral knee.




**Citation:** Manisha J Kumar, Chockalingam. Fracture neck of femur with alkaptonuria: A case report. Kauverian Med J., 2025;2(4):16-20.

Academic Editor: Dr. Venkita S. Suresh

ISSN: 2584-1572 (Online)

**Fig (1):** Pre-operative radiograph showing fracture neck of femur right side with hemiarthroplasty on left side

### 2.1 Management

The patient underwent a cemented total hip replacement under GA for the fracture neck of femur. Regional anaesthesia was attempted but was not successful. Hence, his surgery was performed under general anaesthesia.

Intraoperative findings were as follows. There was blackish pigmentation of the tensor fascia lata, the capsule and the tissue surrounding the hip and the labrum. The head and the acetabular cartilage was completely blackened [Fig 2]. A standard cemented total hip replacement was performed. Wound was closed over a drain, which was removed after 48 hr. Routine post-operative rehabilitation was done following the procedure including walker assisted weight bearing on the first postoperative day. He was discharged having had an uneventful recovery.

**Fig (2)**. Head of femur when removed from the patient showed complete black discolouration

### 2.2 Discussion

Fracture neck of femur is a common, serious, life-threatening injury especially in the elderly population, with an incidence of 1/1000 population [9]. Femoral neck fractures in the elderly population is generally secondary to osteoporosis with low energy trauma. Apart from senile osteoporosis, osteomalacia is also known to be a cause of pathological fracture neck of the femur. Insufficiency fractures may occur in patients, without an apparent history of trauma. These fractures may occur in pre-existing diseases causing an alteration in the quality of bone or due to external factors like bisphosphonates that may cause a change in bone quality. While all these causes may be the aetiological factors for femoral neck fractures in the elderly age group, the incidence of the fractures in young adults is also rising due to the higher incidence in road traffic accidents and trauma.

The treatment for fracture neck of femur is largely based on the age of the patient and the time from injury to the presentation of the patient at the hospital. In older patients and patients with rheumatoid arthritis, arthrosis or pathological fractures, surgeons agree that the best modality of treatment is arthroplasty. The choice between hemiarthroplasty and total joint replacement is influenced by many factors. AAOS in 2014 published guidelines that recommended a total hip replacement in patients above the age of 65 years [10]. However, there is no consensus on the treatment of femoral neck fractures

in younger patients with a displaced fracture. In younger patients, conservative management with fixation using screws may be opted for. However, with this line of management, there is an increased risk of revision surgery. If however, there has been a considerable lag between the time of occurrence of the fracture and the time of surgery, the risk of avascular necrosis rises, and joint replacement may be the choice of the surgeon.

Therefore, apart from the age of the patient and the time of presentation, the aetiology of the fracture is also an important factor in determining the line of management for the patient.

Alkaptonuria is one such disorder that may cause pathological fracture of the neck of femur. It is a rare autosomal recessive metabolic condition which affects the metabolism of phenylalanine and tyrosine. It occurs due to the deficiency of homogentisate dioxygenase leading to the accumulation of homogentisic acid. On exposure to air, HGA undergoes a process called ochronosis, causing oxidation and polymerization of the homogentisic acid that gives a black colour to urine and the tissues it accumulates in [1]. The pigment gives a macroscopic appearance of black colour, while it has an ochre colour on unstained HPE slides giving it the name ochronosis [5].

Edgar Reid first described this condition as a clinical curiosity rather than a phenomenon with and morbid interest [2]. Since then, alkaptonuria has been shown to cause a disorder with multisystem involvement. It is known to affect various organs namely kidneys, major vessels, heart, soft tissues including tendons and cartilage. The clinical manifestations are dark coloured urine on exposure to air, staining of the clothes due to the pigment in sweat, and it is deposited in the patients' skin, teeth, nails, pinnae, sclera and buccal mucosa, larynx, tympanic membrane and tendons. Renal stones, renal failure, aortic valve stenosis and orthopaedic predominance of severe arthritis and spondyloarthropathy, tendon rupture are all signs of a more serious disorder.

While the disorder may itself be congenital, Patients with alkaptonuria may be asymptomatic till the 4th decade of life, when it largely manifests as arthropathy. The pigment has a greater affinity for the hyaline cartilage of the large joints and the intervertebral discs. With advancing age there is deposition of homogentisic acid and its oxidised products extracellularly on the surface of collagen fibres as well as intracellularly in the fibroblasts. With impregnation of the deposit, the tissue tends to become friable and brittle. As a result, severe degenerative changes are seen in the large joints and the thoracic and lumbar spine [3]. Synovial reaction is common, however, pathological fracture neck of femur is quite uncommon.

Ochronotic arthropathy typically involves the intervertebral discs and gives it a characteristic appearance on radiographs. However, peripheral arthropathy has no such imaging peculiarities, leading to a missed diagnosis. The picture obtained radiologically generally does not correlate to the clinical symptoms of the patient.

**3. Orthopaedic manifestations**

The commonest involvement is that of the spine – patient may present with spondylosis; it is describing as the most common 'first sign' of ochronosis [5]. Clinically, the patient presents with chronic back pain with loss of lumbar lordosis with an increase in thoracic kyphosis. It may also lead to loss of height and decreased lumbar flexion. It differs from other degenerative disorders in that the symptoms are more thoraco lumbar than lumbosacral and the lack of involvement of the sacroiliac joint (differentiates it from ankylosing spondylitis). It is a progressive disease in which there is a calcification

or ossification of the disc which is pathgnomonic on radiography, with typical narrowing of the disc spaces and a variable degree of fusion of the adjacent vertebrae and small osteophyte formation and occasional reports of a prolapsed intervertebral disc.

Peripheral arthropathy usually involves the larger joints and commonly, the small joints are spared. The high affinity of the pigment to the proteoglycans of the hyaline cartilage may increase the fragility of the tissue leading to early degenerative changes. Premature large joint arthritis may develop in the middle aged patient between 30 and 50 years of age. It typically affects the hips, knees and shoulders. Radiographically, there is loss of joint space and subchondral sclerosis and osteophytes are not as evident as osteoarthritis. Calcification may also be seen in the menisci when the knee is involved. While there is no definitive medical management of the disease, patients may present with painful debilitating joints that require arthroplasty for severe degenerative arthritis. While there is some concern regarding the decrease in quality of bone and connective tissue due to the disease, Studies have shown that there is no difference in the longevity of the joint post replacement in these patients [6].

Connective tissue involvement is seen with pigmentation of sclera and ear cartilage. However, orthopaedically, it leads to tendon related findings. The accumulation of homogentisic acid in the connective tissue inhibits collagen cross linking and thereby reduction in the structural integrity of collagen. The most often reported finding is that of thickening of the Achilles tendon, with involvement of upto 50% of the patients. Spontaneous ruptures or ruptures with low energy trauma may be seen in these patients, with Achilles and patellar tendons being commonly involved. Tendons generally rupture where they have been weakened by the deposition of the pigment – which is seen intra operatively as blackish discoloration at the ruptured ends. The repair of the tendon has to be done after the tendon has been debrided to healthy tendon. The tendons once repaired, heal well after debridement and primary repair.

**4. Anticipated difficulties during surgery**

Anaesthetic problems - A thorough evaluation of the type and severity of systemic dysfunction is essential before administration of anesthesia [7]. Cardiac problems due to calcified and stenotic valves may be detrimental while considering general anaesthesia. The accumulation of homogentisate may also lead to the development of renal calculi and therefore renal failure. The cartilage of the airway and respiratory system too may be affected in ochronosis - heavy deposition of the pigmentinthe laryngeal, tracheal, and bronchial cartilages may result in hoarseness, dysphagia, and difficult airway management [7]. Restrictive pulmonary disease may also be present in ochronotic fibrosis of the costal cartilages. Homogentisic acid may cause damage to the dura and arachnoid membrane and also narrowing of the disk space and spine fusion would make the regional technique unsuccessful.

Difficult arthroplasty procedure – reports have suggested that the thickening of the capsule may lead to difficulties while attempting to cut the capsule during arthrotomy, and a saw blade may need to be used to substitute the scalpel; difficulty in dislocating the patella during TKR due to stiffened and attenuated patellar tendons. In the post operative phase, ochronosis may affect the mechanical properties of the bone and connective tissue because of which early implant failure maybe seen.

## 5. Conclusion

Fracture neck of femur is a very common fracture causing considerable morbidity and mortality in young and old patients. Apart from trauma, various other causes, like alkaptonuria have to be borne in mind. Alkaptonuria is one such cause of a pathological neck of femur fracture with various other orthopaedic manifestations.

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