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**Olgu Sunumu**

# OSTEOCHONDRAL FRACTURE OF TIBIAL EMINENTIA DUE TO OCHRONOSIS

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**Abstract**

**Introduction**: Ochronosis is a rare homogentisic acid oxidase enzyme deficiency resulting in accumulation of homogentisic acid in connective tissues which results in dysfunction and degeneration. Degeneration of the weight bearing joints like knee and hip has been reported to be involved mostly in the literature. Although degenerative arthritis is the most common pathology in this literature series, mechanical limitation was the first symptom on admission in our case.

**Patient and Methods**: 49 years old male patient has admitted to our clinic with sudden knee pain and limitation of flexion. During the arthroscopy of the knee joint brownish pigmentation of all connective tissues has been found. Degeneration of the medial meniscus and osteochondral fragment of medial tibia eminentia were major pathological findings during surgery for which excision of the fragment and partial meniscectomy were applied. All mechanical symptoms were ceased at the first follow up with almost totally pain relief.

**Results**: Brownish-black discoloration of all soft tissues and also menisci was detectable. All chondral surfaces and menisci were degenerated and brittle which was incompatible with patient's age. On the anterior aspect of the tibia plateau an osteochondral fragment of eminentia and chondral defect of tibia plateau has been detected.

**Conclusion**: Ochronosis causing early degenerative arthritis can be diagnosed sometimes very late, even during arthroplasty surgery in some cases. To prevent early degeneration of the joints due to ochronosis, detection of initial mechanical symptoms and awareness of differential diagnosis of the ochronosis must be kept in mind.

**Key Words**: Arthroscopy, Alkaptonuria, Ochronosis, Knee.

**Tibial Eminentia'nın Okronozise Bağlı Osteokondral Kırığı**

**Özet**

**Giriş**: Alkaptonuri homogentisik asit oksidaz enzim eksikliğine bağlı otozomal resesif bir hastalıktır. Homogentisik asit yıkılamaz ve siyahımsı bir pigment yumuşak dokularda özellikle de kıkırdak dokularda birikerek okronozis denilen dejeneratif değişikliklere yol açar. Bu dejeneratif değişiklikler özellikle yük taşıyan kalça ve diz gibi eklemlerde 4. ve 5. onlu yaşlardan başlayan erken dönem osteoartrite yol açar ve bu hastalar genelde dejeneratif artritlere bağlı şikâyetler ile polikliniklere başvurmaktadırlar. Saf mekanik semptomlarla başvuru yok denecek kadar azdır. Bizim sunduğumuz vakada hastamız dizde kilitlenme ve bükmede zorluk gibi sadece mekanik semptomlar ile başvurmuştur.

**Hasta ve Yöntem**: 49 yaşında erkek hasta dizde ağrı ve bükmede zorluk ile polikliniğe başvurdu. Fizik muayene ve MRG sonuçlarına dayanarak hastaya yapılan diz artroskopisinde tüm dokularda kahverengimsi siyahımsı renk değişikliği izlendi. Medial menisküste dejenerasyon ve medial tibia eminensiada osteokondral kırık fragmanı izlendi. Fragman eksize edilerek patolojiye gönderildi. Medial menisküs arka boynuzdaki dejeneratif yırtık için parsiyel menissektomi uygulandı. Hastanın ameliyat sonrası erken dönem kontrolünde mekanik şikayetleri tamamen geçtiği ve diz ağrısının tama yakın azaldığı tespit edildi.

**Bulgular**: Hastamızın eklem çevresi tüm yumuşak dokularında ve menisküslerde siyahımsı renk değişiklikleri göze çarpmaktaydı. Tüm menisküslerde ve kıkırdak yüzeylerde yaşla uyumsuz dejenerasyon ve kırılganlık mevcuttu. Özellikle tibia plato anteriorunda osteokondral fragman ve kıkırdak defekti izlendi. Medial menisküs arka boynuzunda yırtık mevcuttu.

**Sonuç**: Okronozisin genç yaşlarda başlayan eklem dejenerasyonuna yol açtığı ve hastaların bazılarının artroplasti cerrahisi esnasında tanı aldığı bilinmektedir. Ancak sadece mekanik semptomları olan hastalarda da ileride eklem dejenerasyonuna yol açacağı öngörülen değişikliklerin başlangıcı açısından okronozis tanısı akıllarda tutulmalıdır.

**Anahtar Kelimeler**: Artroskopi, Alkaptonuri, Okronozis, Diz

## Introduction

Alkaptonuria is an autosomal recessive metabolic disease which is caused by homogentisic acid oxidase enzyme deficiency in tyrosine and phenylalanine catabolism. Homogentisic acid (HGA) accumulates progressively in the connective tissues which results in discoloration called ochronosis[1]. This brownish-black discoloration can occur in tendons, ligaments, sclera, heart valves, the intimae of blood vessels and on the skin. Although multiple systems have been affected by deposition of ochronotic pigment, most important

complication of alkaptonuria is ochronotic arthropathy[1,2]. Spine and large joints, especially knee, shoulder and hip are mostly involved[3-5]. Patients are asymptomatic until the 4th and 5th decades of life and first admission is generally because of arthritis[2,6]. We are reporting a case with osteochondral fracture of medial tibia eminentia. Our patient had only mechanical symptoms rather than generalized arthritis which is unusual in ochronosis.

**Case Report**

A 49 year old man has admitted to our hospital complaining left knee pain and limited flexion. He hasn't report any trauma nor any overuse. Especially when extending his knee joint, he had felt limitation and popping. In physical examination tenderness on the lateral knee joint space and crepitation of patellofemoral joint have been defined. Rotational tests were positive in both medial and lateral side. The only abnormal laboratory results were elevation of alkalen phosphatase and HAV (Hepatitis A virus) antibody levels. In direct radiographic examination minimal narrowing of the joint space has been defined. In MRI sections chondral changes in lateral femoral condyle and grade 3 degeneration of medial meniscus have been reported. Osteochondral fracture has been defined also in MRI sections in the anterior aspect of tibia plateau (Figure 1).

Figure 1: An MRI coronal section showing osteochondral fragment of tibia eminentia.

By direct examination of the knee at arthroscopic surgery, diffuse discoloration of all cartilaginous tissues, synovium and menisci has been noted (Figure 2). Femoral condyle and tibia plateau on the medial side were degenerated and grade 3-4 chondromalacia has been noted.

On the anterior aspect of the medial eminentia a 2,5x1,5x0,5 cm osteochondral fragment which is also darkly brown colored has been noted and excised during surgery.

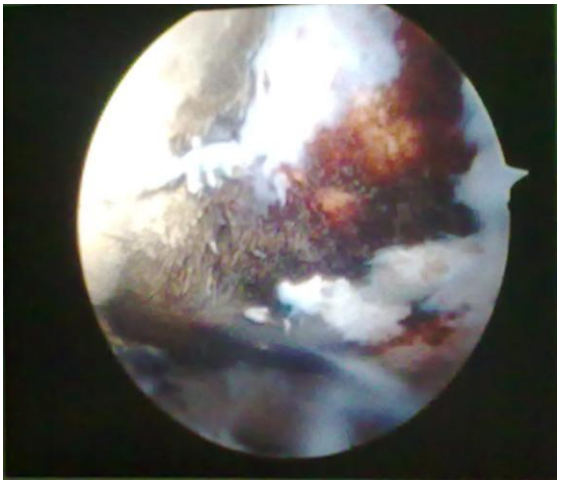

Figure 2: Brownish-black discoloration and degeneration of the cartilage.

The fragment was round shaped and mobile (Figure 3). Posterior horn of the medial meniscus was also degenerated and torn which then has been partially resected. Without any complication patient has been controlled at 15th day and sutures have been removed. His all mechanical complains were ended in the postoperative period with minimal pain during walking. The ROM of the knee was unlimited and painless.

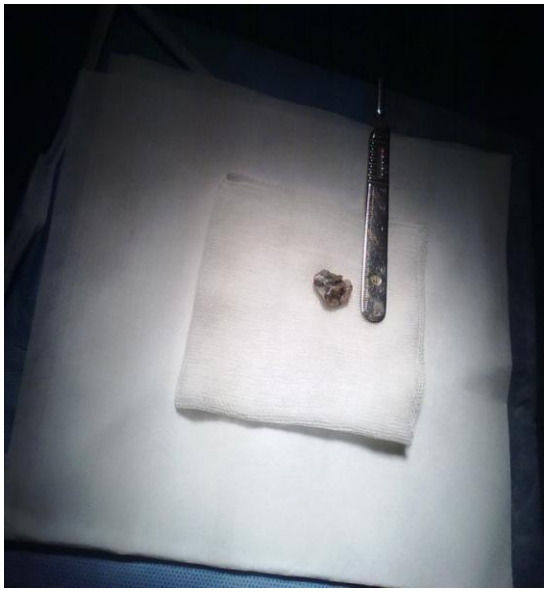

Figure 3: Excised osteochondral fragment of tibia eminentia which is round shaped and discoloured.

**Discussion**

Alkaptonuria is a rare metabolic disease (1/250000) which is inherited autosomal recessively[1]. Absence of homogentisic acid oxidase enzyme in phenylalanine and tyrosine metabolism causes homogentisic acid accumulation in all connective tissues but especially in the cartilage. Due to this accumulation of homogentisic acid, pigmentation called ochronosis occurs in affected tissues. Accumulation of this pigment causes degenerative changes mainly in the cartilage of large joints like knee, hip, shoulder and also intervertebral discs, ribs and ear. The clinical presentation with vertebral changes like spondylosis and canal stenosis is also not rare in this disease[6]. Degeneration of the joints occuring in the knee, shoulder and hip are the leading causes for admission after 4th decade of life. The first symptoms are mainly swelling and pain of the joint. In the knees affected by ochronosis, cartilage becomes more brittle and fragile. Its mechanical strength also decreases against strain and stress. Therefore chondral fissures and fractures resulting in degradation and separation of cartilage from underlying bone can be seen[5-7]. Also small fragments of cartilage can also adhere to the synovial membrane and cause fibrosis and chondromatosis. As a general rule with degenerative symptoms of osteoarthritis all differential diagnosis including metabolic deposition disorders also should be kept in mind. But with this case we also should keep in mind that ochronosis can appear in younger patients with mechanical symptoms only. Meniscal tissues and osteochondral junctions can also be the first affected tissues in an active individual causing admission to the orthopedic clinics[8]. Therefore not only the elderly arthritic patients but also younger and more active people can also be affected by ochronosis which will be generally misdiagnosed or ignored for treatment.

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