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Case Report / Olgu Sunumu

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Sacroiliac joint involvement in ochronotic spondylosis

Okronotik spondilozda sakroiliak eklem tutulumu

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Abstract

The musculoskeletal involvement can be seen in alkaptonuria, which is an autosomal recessive disease. Homogentisic acid levels elevate during the disease. The accumulation of homogentisic acid in the body tissues and the formation of pigment is termed ochronosis. We present a 51-year-old male patient with intraoperative ochronotic arthropathy of the knees and bilateral punctate brown pigmentation on the sclera. Spine movements in the lumbar and cervical movements were limited. Movements of both hips were limited and painful in all directions. On the sacroiliac joint graphy, severe narrowing, irregularity, and erosions were detected in the bilateral coxofemoral areas, especially on the right, which might suggest grade 3 sacroiliitis. When the patient's urine was mixed with sodium hydroxide, the urine became alkaline and its color turned black.

Keywords: Ochronosis, sacroiliac joint, ankylosing spondylitis

Öz

Otozomal resesif bir hastalık olan alkaptonüride kas-iskelet sistemi tutulumu görülebilir. Hastalıkta homogentisik asit düzeyi yükselmiştir. Homogentisik asitin bağ dokularında birikmesi ve pigment oluşumuna okronozis denir. Bu yazıda, dizlerinde intraoperatif okronotik artropatisi saptanan ve sklerasında bilateral punktat kahverengi pigmentasyonu olan 51 yaşında bir erkek hastayı sunuyoruz. Hastanın lomber ve servikal bölgelerde omurga hareketleri sınırlıydı. Her iki kalça hareketleri kısıtlı ve her yöne ağrılıydı. Sakroiliak eklem grafisinde özellikle sağda bilateral koksofemoral alanlarda şiddetli daralma, düzensizlik ve erozyonlar saptandı ve bu da grade 3 sakroiliiti düşündürebilirdi. Hastanın idrarına sodyum hidroksit solüsyonu eklenip alkali hale getirildiğinde idrarın rengi siyaha döndü.

Anahtar Kelimeler: Okronozis, sakroiliak eklem, ankilozan spondilit

Introduction

Homogentisic acid (HGA) oxidase enzyme deficiency is seen in ochronosis, which is an autosomal recessive disease. This deficiency causes HGA accumulation in the connective tissue.[1,2] Brown-black pigment deposition is seen in connective tissues, especially in the cartilage, and less commonly in the skin and sclera.[3] Musculoskeletal involvement, including axial (intervertebral discs of the lumbar spine) and peripheral joint involvement, is one of the important causes of morbidity. Treatment is usually conservative. Ascorbic acid and dietary restrictions of foods containing phenylalanine and tyrosine are used in the treatment.[4,5] Here we present a case of ochronotic spondylosis with sacroiliac joint involvement.

Case Report

A 51-year-old male presented with mechanical right hip pain. The patient complained of non-inflammatory low back pain for 15 years, however, he did not benefit from the physical therapy and rehabilitation program for degenerative disc disease. As his complaints increased, he was referred to us with a pre-diagnosis of axial spondyloarthropathy.

He had a 10-minute morning stiffness accompanied by degenerative waist, knee, shoulder and hip pain. She had no history of weakness, fever, or weight loss. Blackening was noticed in his diaper by his mother when he was a baby. While both knee joints were operated for osteoarthritis, black pigment accumulation was observed in the joints and cartilages (Figure 1). On examination, bilateral punctate brown pigmentation was observed on the sclera (Figure 2)

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and limited spine movements in the lumbar and cervical regions. The femoral nerve stretching test and the Laseque test was negative. Movements of both hips were limited and painful in all directions.

Laboratory examinations revealed slightly acidic urine (pH=6), normal C-reactive protein levels and erythrocyte sedimentation rate, and a negative Human Leukocyte Antigen B27 test. Sodium hydroxide was mixed in the patient's urine. The color of the urine turned black (Figure 3). Radiographically, anteroposterior and lateral lumbar radiographs showed decreased lumbar lordosis, decreased intervertebral space, and intervertebral disk calcifications (Figure 4). There were rough syndesmophytes, osteophytic degenerative changes on the anterior vertebrae, and marginal subchondral sclerosis on cervical vertebra radiographs (Figure 5). On the sacroiliac joint graphy, severe narrowing, irregularity, and erosions were detected in the bilateral coxofemoral areas, especially on the right, which might suggest grade 3 sacroiliitis (Figure 6).

On magnetic resonance imaging of the sacroiliac joints, hyperintense lesions were observed in Cor STIR series on both the sacral and iliac surfaces, which may be compatible with sacroiliitis (Figure 7). Ochronotic arthropathy and

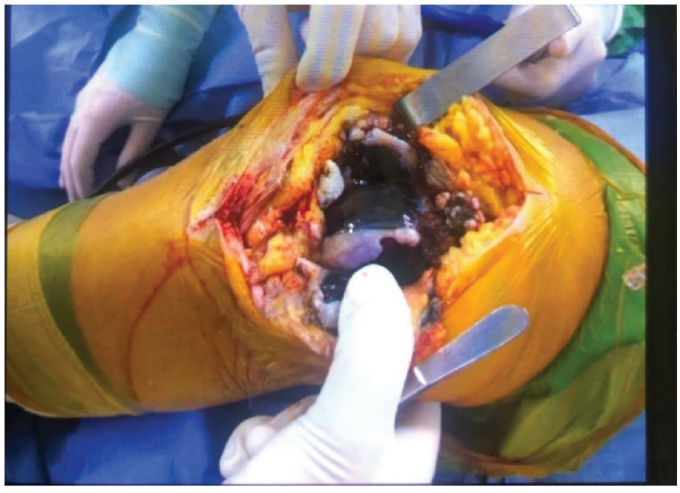

Figure 1. Pigment accumulation in the cartilage

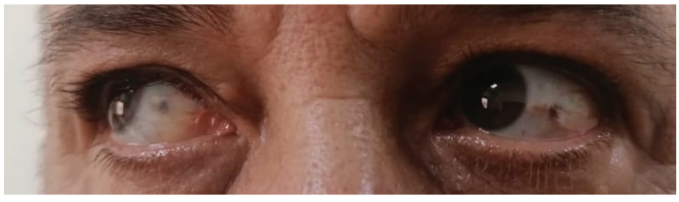

Figure 2. Pigmentation in the scleras

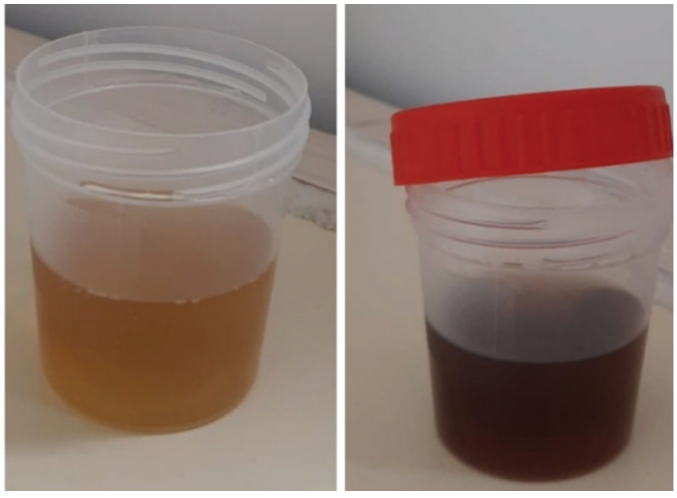

Figure 3. Black urine

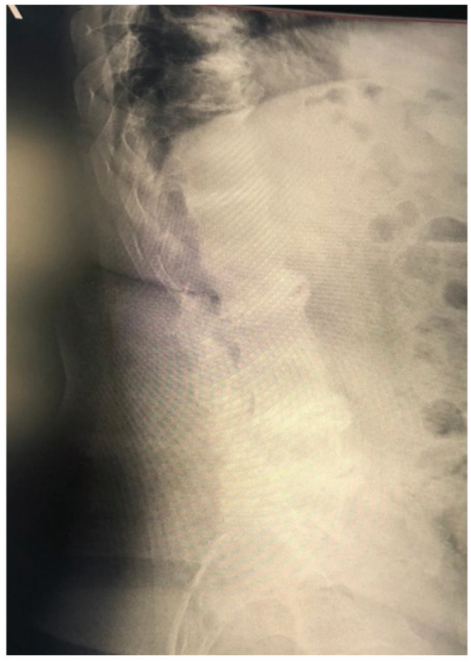

Figure 4. Lumbar vertebrae radiography

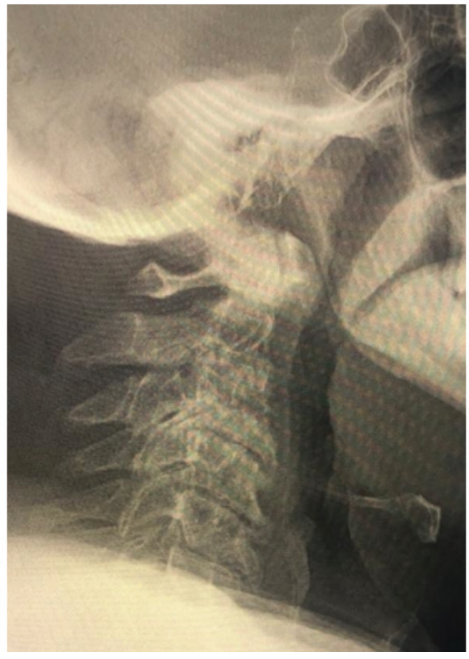

Figure 5. Cervical vertebra radiography

ochronotic axial spondyloarthropathy were diagnosed with secondary osteoarthrosis, ochronotic hyperpigmentation in the sclera, dark urine, and observation of ochronotic pigment accumulation in knee joint surgery.

Discussion

Ochronosis is an autosomal recessive metabolic disease.[6] Because of the deficiency of HGA oxidase enzyme, HGA accumulates in various body tissues. Increased HGA accumulates in the conjunctiva, sclera and in the periferal joints, disc spaces and ear cartilage. A dark brown or dark yellow occurs due to the deposition of HGA in the tissues. [7] Alkaptonuria is a genetic disease that is present at birth. Over time, ochronotic pigments accumulate in these organs. The symptoms appear in the fourth decade.[8] In ochronosis, the thoracolumbar vertebrae are frequently affected, and it may have significant degenerative involvement and may be disabling.[9]

Conclusion

Ochronotic arthropathy is frequently overlooked. Diseases such as hyperparathyroidism, ankylosing spondylitis, amyloidosis, hemochromatosis, degenerative joint diseases are included in the differential diagnosis of ochronosis.[10] This case is an example of ochronosis, one of the easily missed and extremely rare mimics of ankylosing spondylitis.

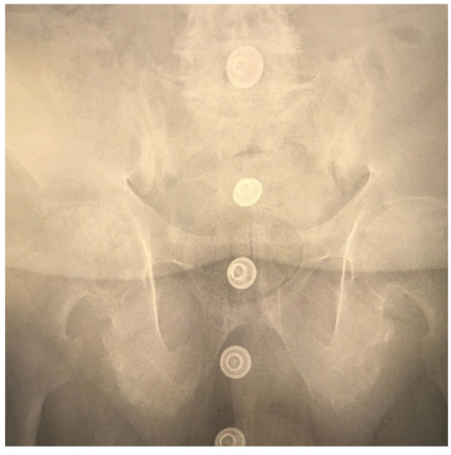

Figure 6. Sacroiliac joint radiography, 361x360 mm (72x72 DPI)

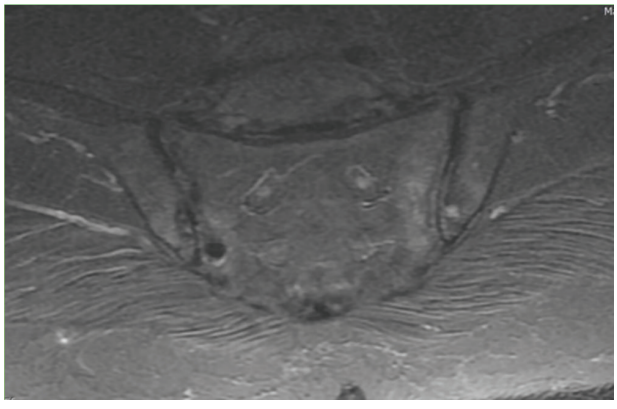

Figure 7. Magnetic resonance imaging of the sacroiliac joints

Ethics

Informed Consent: Written informed consent was obtained from the patient.

Peer-review: Externally and internally peer-reviewed.

Authorship Contributions

Concept: Z.Ö., S.M.T., E.G., Design: Z.Ö., S.M.T., Data Collection or Processing: Z.Ö., S.M.T., D.K., N.E., E.G., Analysis or Interpretation: Z.Ö., S.M.T., D.K., N.E., E.G., Literature Search: Z.Ö., S.M.T., E.G., Writing: Z.Ö., S.M.T.

Conflict of Interest: No conflict of interest was declared by the authors.

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