EJSS
European Journal of Surgical Sciences

Eur J Surg Sci 2014;5(1):39-42

CASE REPORT

# Undefined Findings of Ochronotic Polyarthropathy-Cervical Pseudoankylosis, Spinal Cord Atrophy and Anterior Mediastinal Mass: Case Report

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## ABSTRACT

*Homogentisic acid oxidase (HGO) enzyme deficiency from birth results in alkaptonuria, a rarely seen metabolic disease involving several systems in the body. The enzyme deficiency results in an accumulation of homogentisic acid seen particularly in connective tissue, which is defined as ochronosis. Depending on the area of accumulation, there may be involvement of joints, skin, eyes, and the cardiovascular, genitourinary, respiratory, endocrine, and central nervous systems. The initial symptoms of ochronosis are phenotypic (pigmentation of the sclera, ears, nails, and generally on the face), radiologic (primary glenohumeral and acromioclavicular arthrosis, whole spine degeneration, spinal stenosis, other joint arthrosis), and intraoperative (blue-black coloration). We could not find any publication in the English literature about pseudoankylosis. We present a 56-year-old male patient with rare findings that included cervical pseudoankylosis, spinal cord atrophy and anterior mediastinal mass.*

***Key words:*** *Ochronosis, Homogentisic acid oxidase, Pseudoankylosis, Spinal cord atrophy, Anterior mediastinal mass*

Received: November 29, 2012 • Accepted: May 18, 2013

## ÖZET

### Okronotik Poliartropatide Tanımlanmamış Bulgular; Servikal Psödoankiloz, Spinal Kord Atrofisi ve Ön Mediastende Kitle: Olgu Sunumu

*Doğuştan homojensitik asit oksidaz enzim eksikliği az rastlanan ve vücutta birçok sistemi etkileyen bir hastalık olan alkaptanüriye neden olur. Enzim eksikliği bağ dokusunda homojensitik asit birikimiyle seyreden ve okronozis olarak adlandırılan rahatsızlığa sebep olur. Birikimin yerine göre eklemler, deri, gözler, kardiyovasküler, genitoüriner, respiratuar, endokrin ve santral sinir sistemi etkilenebilir. Okronozis başlıca belirtilerini fenotipik (sklera, kulaklar, tırnaklar ve yüzde pigmentasyon), radyolojik (glenohumeral ve akromiyoklaviküler artroz, spinal dejenerasyon, spinal darlık veya diğer eklemlerde artroz), eklemlerde mavi, siyah renklenme şeklinde gösterir. İngilizce literatürde psödoankiloz ile ilgili herhangi bir yayına rastlamadık. Bu yazıda servikal psödoankiloz, kord atrofisi ön mediastende kitle nadir bulgularıyla seyreden 56 yaşında erkek hastayı sunuyoruz.*

***Anahtar kelimeler:*** *Okronozis, Homojensitik asit oksidaz, Psödoankiloz, Spinal kord atrofisi, Ön mediastinal kitle*

Geliş Tarihi: 29 Kasım 2012 • Kabul Ediliş Tarihi: 18 Mayıs 2013

## INTRODUCTION

Homogentisic acid oxidase (HGO) enzyme deficiency from birth results in alkaptonuria, a rarely seen metabolic disease involving several systems in the body. The enzyme deficiency results in an accumulation of homogentisic acid seen particularly in connective tissue, which is defined as ochronosis. The development of ankylosis in the vertebral column is a frequently seen finding. To our best knowledge, there is no case in the English literature about pseudoankylosis. We present a patient with an unusual presentation of ochronosis.

## CASE REPORT

A 56-year-old male presented to our polyclinic with pain in the right knee and hip and limited movement of the neck. There was nothing remarkable in the family history.

In the physical examination, blue-black pigmentation was observed in the ears, nail beds and sclera. There was a palpable, hard, painless mass lesion below the sternum in the anterior chest wall. Severely limited movement of the cervical vertebrae was observed.

Examination of direct radiographs showed findings of right coxarthrosis, gonarthrosis, and spondylosis. On the lateral thoracic radiograph, a lesion 8 cm in length with mass was determined below the sternum (Figure 1). In the vertebral column, besides generalized findings of arthrosis, a high degree severe narrowing between discs was observed, and osteophytes were seen extending anterior-posterior.

On the spinal magnetic resonance imaging (MRI), facet joint degeneration was observed in the cervical area, as well as spinal stenosis and spinal cord atrophy. Pseudoankylosis was seen between C7 and T1 vertebrae (Figures 2, 3). The neurological examination was determined as normal.

Total hip replacement for the right coxarthrosis was applied first to relieve the patient's complaints. During surgery, pigmentation was seen on the femoral head and neck, and the diagnosis was confirmed by pathological examination (Figure 4). Six months later, the patient underwent total knee replacement for the right gonarthrosis. Black discoloration in the form of brushstrokes was seen in the joint cartilage, and black degeneration was noticed in the meniscus and tendons (Figure 5). No problems were encountered in the postoperative one-year follow-up (e.g. loosening, infection, dislocation, etc.) (Figure 6).

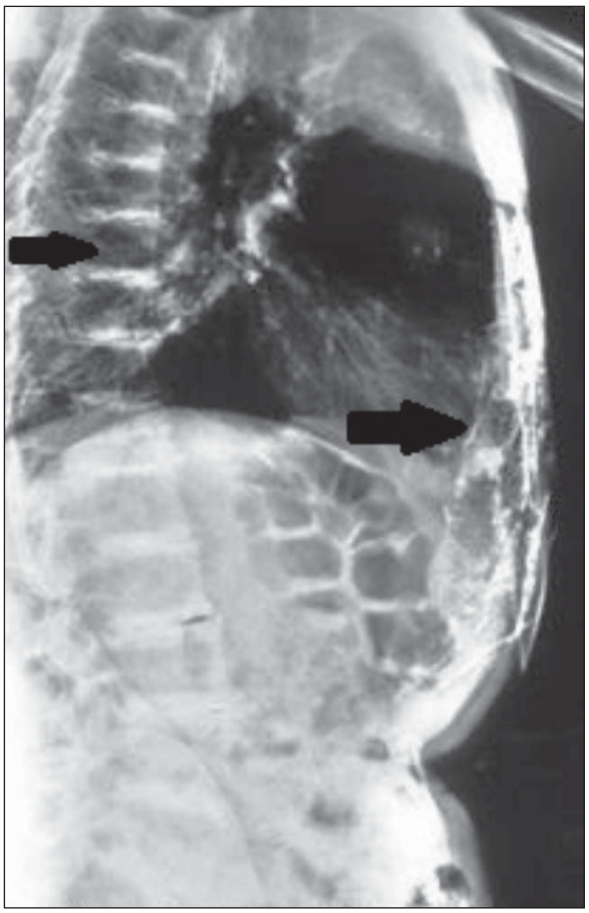

**Figure 1. Vertebral spondylosis and anterior mediastinal mass.**

## DISCUSSION

Disease associated with the lack of HGO enzyme, which plays a role in the metabolism of phenylalanine and tyrosine, is an autosomal recessive disease seen in approximately 1/100.000-250.000[1,2]. The disease was first described by Virshow in 1866. In 1915, Sonderbergh named this condition 'osteitis deformans alkaptonuria', defining the relationship between ochronosis and spondylosis.

On the lateral thoracic radiograph, a mass was observed starting in the anterior mediastinum of the infrasternal area and extending along the linea alba. The mass was evaluated as painless and hard. It was compared with only one similar case in the literature[3].

When the intraoperative evaluation is not normal due to blue-black coloration, ochronosis should be the first diagnosis to come to mind. It should not be forgotten that these patients may experience sponta-

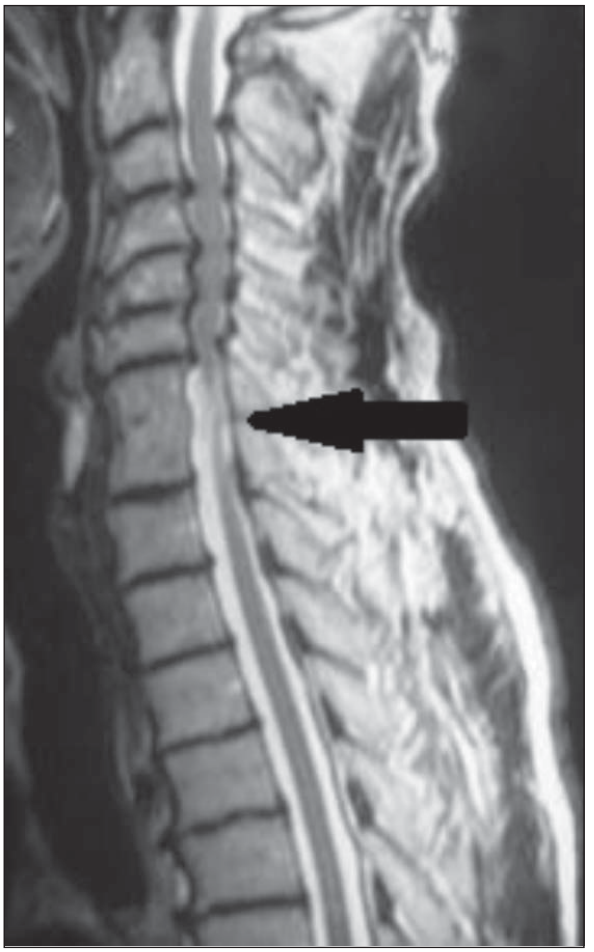

**Figure 2. Cervical MR sagittal section showing pseudoankylosis and cord atrophy between C7-T1.**

neous or minimal energy fractures and ruptures, and excessive stresses should be avoided [4,5].

Varying forms of ochronotic vertebrae involvement have been published in the literature, such as thoracic myelopathy, root canal stenosis, intervertebral disc tears, herniations, calcifications, ankylosis, and spondylolisthesis[6-9]. In our patient, there were concomitant findings of cervical spine degeneration, spinal stenosis, cervical pseudoankylosis, and cord atrophy. To the best of our knowledge, there is no similar case in the literature with which to compare the presented patient.

In conclusion, ochronosis is a disease that may be diagnosed after a thorough physical examination and imaging evaluation. In the presence of eye and skin findings together with advanced-stage degenerative

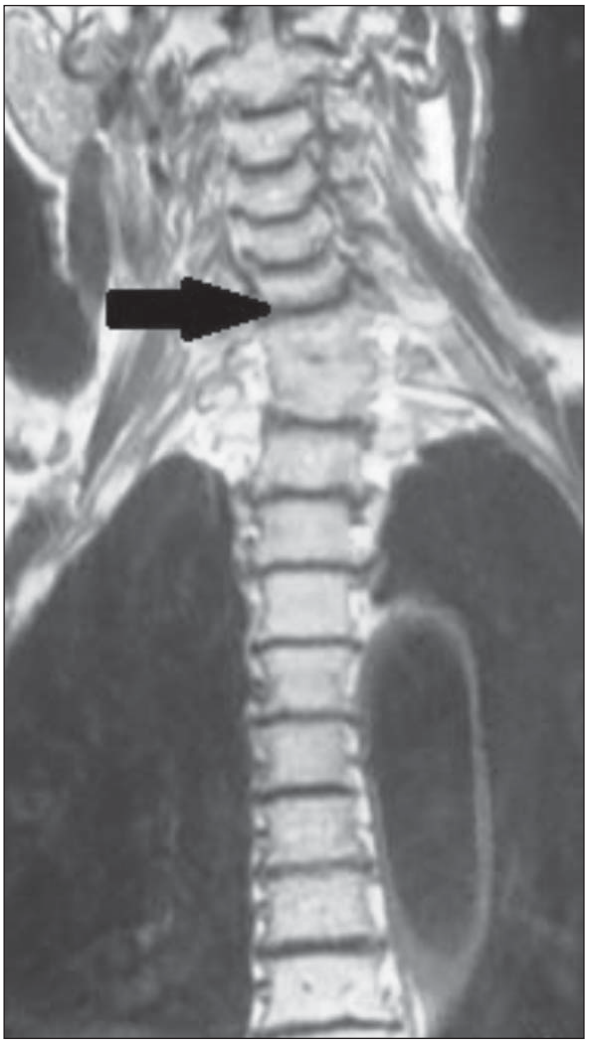

**Figure 3. Cervical MR coronal section showing pseudoankylosis and scoliosis of the cervicothoracic region.**

findings in the spinal column (osteophytes, narrowing between discs, disc calcification) and early onset of degeneration in the shoulder, knee and hip joints, ochronosis should be kept in mind. When the surgical intervention is being planned, cervical vertebrae involvement should be kept in mind, and care should be taken in the selection of the anesthesia technique with respect to difficult intubation and possible complications[10]. Furthermore, patients with spinal cord atrophy should be monitored neurologically. A patient with an anterior mediastinal mass should be informed of the possible need for midline incision surgical intervention.

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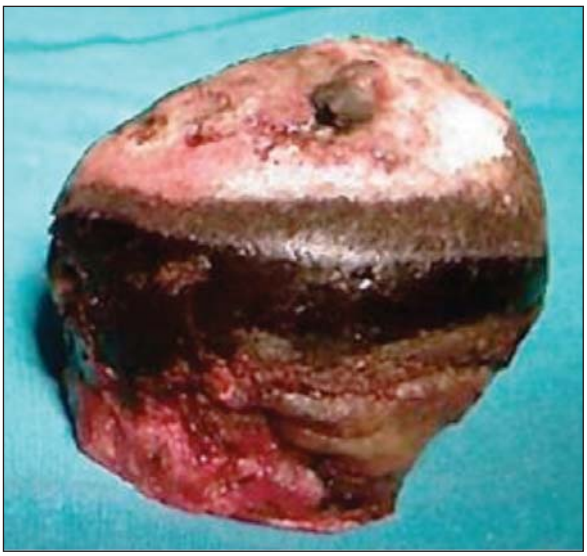

**Figure 4. Femoral head.**

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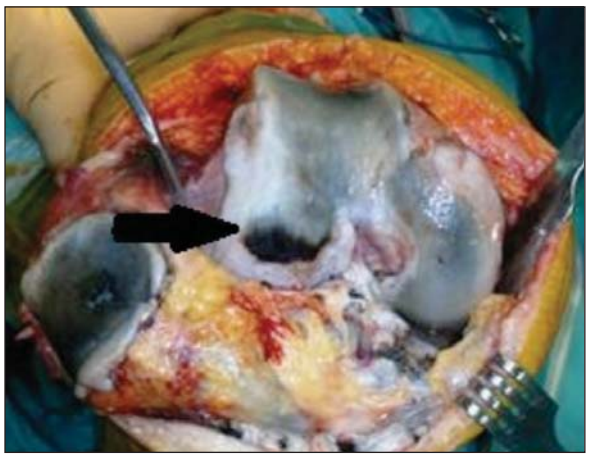

**Figure 5. General view of the knee.**

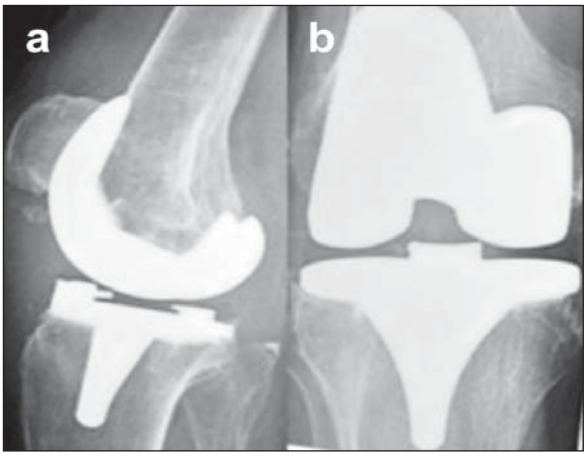


**Figure 6. Postoperative 1-year follow-up of total knee arthroplasty.**

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