OCHRONOTIC ARTHROPATHY

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SUMMARY

The authors report the case of a patient with ochronotic arthropathy. This rare, inherited disease of autossomal recessive trait is the clinical manifestation of alkaptonuria. The patient presented clinically with chronic low back pain, disability, pain and weakness/stiffness in his shoulders and knees associated with dark urine. The main purpose of the orthopaedic approach in this disease is to control the pain and improve the function of the affected joints by performing arthroplasties when necessary.

Keywords: Ochronosis; alkaptonuria.

INTRODUCTION

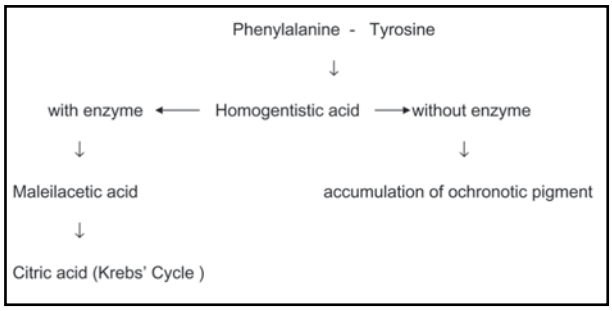
Alkaptonuria is a rare autossomal recessive disease, resulting from a deficiency of the enzyme homogentistic acid oxidase, produced by liver and kidneys, which is involved in the metabolism of two amino acids: phenylalanine and tyrosine[1]. In the absence of this enzyme, an accumulation of ochronotic pigment occurs (Algorithm 1).

Alkaptonuria may be asymptomatic or cause ochronosis, which is characterized by the accumulation of a pigment-like polymer of the homogentistic acid – the alkaptona – on organic tissues[2]. Approximately half the number of alkaptonuria patients develops ochronosis, with a male prevalence of 2:1, usually after the fourth decade of life[3].

The incidence of alkaptonuria in general population is approximately 1:1000.000 births, with no ethnical prevalence[4].

The most important clinical manifestations of ochronosis are: arthropathy, eye and skin pigmentation, dark urine and cardiovascular affection. Diagnostic is clinically provided, corroborated by homogentistic acid presence in urine (approximately 5g/24hs).

In this report, we present the case of a patient with ochronotic arthropathy.



Algorithm 1

CASE REPORT

A 55 year-old male, white patient, born and living in São Paulo, arrived at the Infirmary presenting with a chronic pain picture at the lumbar spine, shoulders, with the left shoulder being more affected, and knees. He told that pain was progressively getting worse, and was associated to weakness and joint functional restraint, not related to trauma or physical effort. He mentioned that his urine has been dark since he was born. At the time, he didn't know the causes for his complaints.

At physical examination, dark spots were seen on eye sclera (Figure 1), on external pinna and on the hands (Figure 2). Spine presented with lumbar lordosis rectification, pain upon lumbar spinous apophysis, and trunk flexion-extension motion restraint. A limitation of the active and passive range of motion was observed on the shoulders: right shoulder with elevation of 160º, abduction of 70º, outer rotation of 20º and inner rotation at lumbar-L1 level. Left shoulder demonstrated an elevation of 150º, abduction of 80º, outer rotation of 10º and inner rotation at lumbar-L1 level.

A discrete atrophy of the quadriccipital muscle was observed in the knees, with no joint swelling, no angle deviation, with a slight active and passive restraint of the range of motion: right knee in 0º of extension and 130º of flexion; left knee in 0º of extension and 120º of flexion.

Diagnostic investigation was initiated with ordinary X-ray images of the thoracic-lumbar spine, knees and shoulders, which showed: 1) in spine: reduction of intervertebral spaces, bone sclerosis, slight osteophytosis, and intervertebral discs calcification especially at lumbar spine (Figure 3); 2) in the shoulders: we verified a reduction of the glenohumeral joint space with arthrosis, without osteophytes (Figure 4); 3) in the knees: we noted a bilateral three-compartmental arthrosis, with no angle deformity, with a slight osteophytosis (Figure 5).

Laboratory tests showed: hemogram with hemoglobin levels of 14.2 g/dl ; hematocrit levels of 41.5 ml eritrac./dl ; leukocyte levels of 8470/mm^3; platelets 340,000/mm^3 and VHS of 21 mm^3/h. Renal function and Type-I urine at normal levels (except for the dark color). Investigation for homogentistic acid in the urine was positive.

Skin biopsy showed the presence of a blacked pigment, identified as alkaptona, compatible with ochronosis diagnostic.

Study conducted at the Orthopaedics and Traumatology Service of the Hospital do Servidor Público Estadual de São Paulo (SOT-HSPE-SP)

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The patient was submitted to total arthroscopy surgery on left shoulder (Figure 6).

DISCUSSION

Alkaptonuria is a rare disease, with a prevalence ratio of 1:1000.000 births. Joint achronosis is one of the forms of clinical manifestation of the disease and occurs in approximately half the number of patients with alkaptonuria after the fourth decade of life. The suspected diagnosis of alkaptonuria should be considered in patients presenting with dark urine, skin changes (darkening of hands and ears, spots on eye sclera), and joint pain (shoulders, knees and hips). Diagnosis is confirmed by findings of homogentistic acid in the urine[2,3]. Among clinical findings, ochronotic arthropathy is characterized by ochronotic pigment deposits (alkaptona) on joint cartilage and on intervertebral disc, with painful symptoms initially at the spine and subsequent affection of peripheral joints, with a higher incidence in knees, shoulders and hips, with elbows, ankles and wrists being rarely affected. Clinical evolution of ochronotic arthropathy is slow and progressive. In X-ray studies of the spine, ochronotic arthropathy shows few signs of osteophytosis and a marked calcification of intervertebral discs, especially at the lumbar spine. In peripheral joints, X-ray images are characterized by a reduction of the joint spaces, subchondral bone sclerosis and a slight or absent osteophytosis[4] .

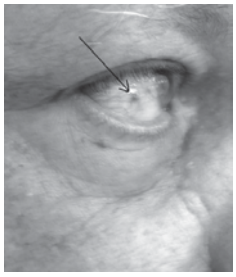

Figure 1 - *Dark spot at sclerotic region of the eye*

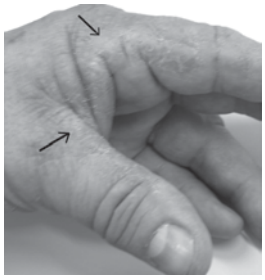

Figure 2 - *Dark pigmentation on the hand*

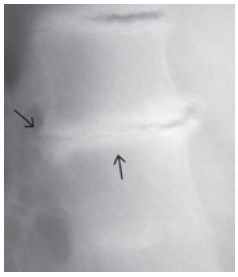

Figure 3 - *X-ray image of the lumbar spine*

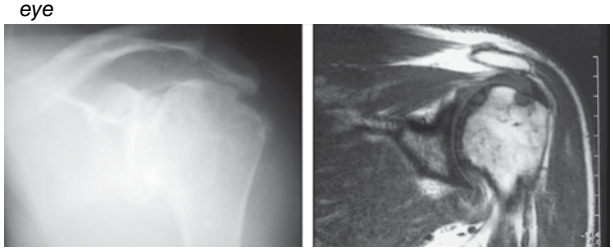

Figure 4 - *X-ray and magnetic resonance image of the left shoulder showing glenohumeral arthrosis*

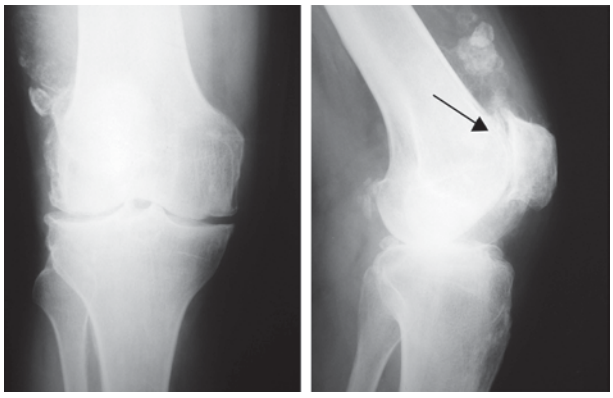

Figure 5 - *X-ray images of the left knee showing arthrosis*

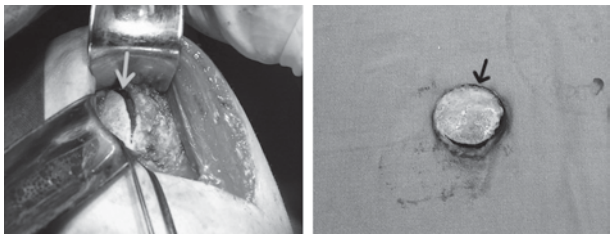

Figure 6 - *Per-operative photographs showing arthrosis and a black spot at humeral head*

Regarding cardiovascular involvement in ochronosis, its highest incidence is at the endocardium, inner layer of the aorta and cardiac valves. Nevertheless, there is no relationship between the degree of pigmentation of such structures and the symptoms presented by the patient[2,3].

The genitourinary ochronosis is characterized by a darkened urine present since birth, many times with no other associated symptom. We may also see an increased incidence of renal lithiasis and prostate pigmentation by alkaptona in male patients after the fifth decade of life[3,4].

The involvement of skin and mucosa occurs in a frequency order, initially on ear lobes, eye sclera, and then, on the hands[3].

Other diseases may be associated to alkaptonuria: gutta, osteoporosis, diabetes mellitus, polycystic kidneys, ankylosing spondylitis, Addison's disease, and pseudogutta.

Alkaptonuria does not change patient's survival rates[2,3,4], although in many cases the joint ochronosis' clinical picture is extremely disabling.

Orthopaedic treatment consists of follow-up in an outpatient basis, symptomatic treatment of joints affected by joint ochronosis, and procedures of replacement arthroplasties, especially in knees, shoulders and hips, whenever necessary.

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