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A case of ochronosis: upper extremity involvement

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Abstract We present an ochronotic patient with spondylitis and upper extremity involvement. We also evaluated radiologic findings of joints that were involved and MRI features of the lumbar spine.

Keywords Ochronosis · Spine · Magnetic resonance imaging · Upper extremity

Introduction

Alkaptonuria is a rare hereditary autosomal recessive disease in which a defect in the metabolism of homogentisic acid leads to pigmentation of the tissues termed ochronosis. Alkaptonuria occurs in about 1 in 200,000 individuals. Alkaptonuria itself is a symptomless condition. Clinical signs develop when pigment is deposited in cartilage [2]. Degenerative arthritis and spondylitis occur in the later stages [1].

We report not only the radiologic and clinical findings of the lumbar spine but also the involvement of hand and shoulder joints, which is seen rarely in ochronosis.

Case report

A 58-year-old man with alkaptonuria–ochronosis was admitted in March 2001 to our department because of pain in both knees and a low backache for 1 year. He also complained of pain and stiffness in his hands and shoulders. The low backache was radiating to the anterior aspect of the right thigh. There was a history of dark urine.

On physical examination, there was dark pigmentation on his ear cartilage and on his face. Bilateral knees were tender, and minimal effusion was palpated. We did not attempt to aspirate, as synovial effusion was minimal, and there was no sign of infection clinically. There was also pain, tenderness and restriction of movement in the distal interphalangeal joints of his hands. His shoulder movements were also painful. There was wasting of the right thigh and calf and weakness of the knee and ankle. There was sensory impairment over the right L5 dermatome.

Laboratory results showed erythrocyte sedimentation rate was 39 mm/h (westergen), c-reactive protein 3.2 mg/l. (normal value 5 ≤ mg/l), hemogram, white blood cell count, liver enzymes were within normal limits. Latex test and HLA-B27 was negative, BUN 43 mg/dl (normal value 4.67–23.36) urea 95mg/dl (normal value 10–50), and blood creatine 2.7mg/dl (normal value 0.5–1.2), urine protein 191.36 mg (normal value 50–80 mg), calcium 8.8mg/dl, phosphorus 4.6mg/dl. A pelvic sonography revealed right nephrectomy and left atrophic kidney (8 cm).

Due to possible pyelonephritis, a right nephrectomy had been performed 5 years previously. The small left kidney with tubulointerstitial nephritis caused the patient to be on a chronic hemodialysis program. The patient had no history of renal stones.

A histopathological section, stained with hematoxylin-eosin showed ochronotic pigment free in the dermis with its characteristic yellow-brown color.

Radiography of the knees showed joint space narrowing, mild osteophytes of the tibial plato, and soft tissue swelling. Radiography of the spine showed multiple disc degeneration, disc calcification (typical wafer-like calcification), narrowing of the disc space, vacuum phenomena and vertebral body osteoporosis (Fig. 1). An MRI showed multiple disc prolapses, which gave a low signal on T1- and T2-weighted images (Fig. 2).

Radiography of the bilateral small joints of the hands (Distal interphalangeal joints and first carpometacarpal joints) bilateral shoulders showed joint space narrowing, sclerosis and small osteophytes (Fig. 3, Fig. 4).

Discussion

In alkaptonuria, homogentisic acid, a product of tyrosine and phenylalanine metabolism cannot be further metabolized, due to deficiency of the enzyme homogentisic acid oxidase [1]. It is excreted in the urine, rendering it a dark color. High levels of homogentisic acid lead to deposition in the tissues, especially in
connective tissue and cartilage. It forms a pigment, which is termed ochronosis [2,3]. Ochronotic arthropathy is a manifestation of long-standing alkaptonuria, and symptoms and signs appear in the fourth decade of life [1, 2,3]. Our patient has had complaints for ten years. Pigmentation increases with age, and pigment accelerates degeneration of cartilage and may lead to ochronotic arthritis in the peripheral skeleton, which resembles osteoarthritis radiologically [1]. However, osteophytes and subchondral cysts are not prominent at peripheral joints, which is different from osteoarthritis. The lumbar spine is affected first, followed by the dorsal and cervical spine. In our patient, plain film showed osteoporosis, generalized narrowing of the discs, calcification in the intervertebral disc space at the lumbar region, which is consistent with the findings of ochronosis.

In rare cases, the small joints of the hands, shoulders, wrists, and elbows are affected. Alkaptonuria can lead to an arthropathy resembling degenerative joint disease of the glenohumeral joint in one or both shoulders. In this patient, there was involvement of distal and first metacarpophalangeal joints of both hands, which was characterized by degeneration, sclerosis and joint space narrowing on radiography [1, 2, 3]. The shoulders were also affected bilaterally in this patient. Shoulder pain is less frequent and develops later than spondylosis [2]. Rheumatoid arthritis and osteoarthritis should be excluded because of the involvement of small joints and the shoulders. In rheumatoid arthritis, the glenohumeral joint is affected by osteoporosis, symmetric loss of joint space, osseous erosions and tear of rotator cuff, which is different from osteoarthritis and ochronosis.