A case of ochronosis: MRI of the lumbar spine

Abstract We present the MRI features of the lumbar spine in a patient with ochronosis.

Key words Alkaptonuria · Ochronosis · Spine · Magnetic resonance imaging

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 (ochronosis). Degenerative arthritis and spondylolisthesis occur in the later stages. We report the MRI findings in the lumbar spine of a patient of ochronosis with multiple prolapsed lumbar discs.

Case report

A 35-year-old man presented with low backache for 7 months, radiating to the anterior aspect of the right thigh. Although there was pain in the right knee there was no history of swelling or restriction of movement. There was no history of dark urine or staining of the clothes. 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.

Plain films of the lumbar spine showed decalcification of the vertebral bodies, a decrease in the disc spaces and sclerosis of the endplates (Fig. 1). The spinal canal was not narrow and there were generalised degenerative changes. MRI showed multiple prolapsed discs (Fig. 2), which gave low signal on T1- and T2-weighted images.

At surgery all the lumbar discs were prolapsed. They were soft, spongy and greyish-black, leading to initial consideration of metanotic deposits or haemorrhagic degeneration. A frozen section was reported as showing only collagen. L2–5 laminectomy and L2–3, 3-4, 4–5 and L5–S1 discectomies were done. Postoperatively the patient had relief of pain.

Histological examination showed markedly degenerate, fibrillated fibrocortilage with brownish pigmentation which did not stain for iron. This was reported as consistent with ochronosis, which apart from iron is the only pigment which stains the stroma. Urine test for ochronotic pigment was positive.

At follow-up 2 years later, the patient had no leg pain and very minimal back pain.

Discussion

In alkaptonuria homogentisic acid, a product of tyrosine and phenylalanine metabolism, cannot be further metabolised, due to deficiency of the enzyme homogentisic acid oxidase. It is excreted in the urine which, on standing, oxides and turns dark. It is also excreted in sweat which may also be pigmented, leading to staining of the clothes in areas such as the axilla and groin. High levels of homogentisic acid lead to deposition in the tissues where it slowly oxides, forming a pigment grey to bluish-black macroscopically, but ochre microscopically, and is hence termed ochronosis. Pigmentation increases with age, and cartilage, fibrous tissues, fibrocortilage,
tendons and ligaments may be densely pigmented even to the point of being coal-black [1, 2].

Ochronotic pigment accelerates degeneration of cartilage [2]. Long standing alkaptonuria may lead to “ochronotic arthritis” which resembles osteoarthritis radiologically [3]. Changes suggesting alkaptonuria are degeneration and narrowing of the intervertebral discs with calcification of the remaining disc material [3]. There may be decalcification of the vertebral bodies [4]. The lumbar spine is affected first, followed by the dorsal and cervical regions. In contrast to degenerative osteoarthritis, in which lumbosacral changes are predominant, the sacroiliac joints are not affected [4, 5].

There is a variable degree of fusion of the vertebral bodies. In contrast to rheumatoid spondylitis, there is minimal osteophyte formation or calcification of the intervertebral ligaments [1]. In our patient plain films showed osteoporosis and generalised narrowing of the discs with sclerosis of the end plates, but little calcification.

Fig. 1a, b Plain anteroposterior and lateral views of the lumbar spine showing multiple disc degeneration with osteophytes and decalcification of the vertebrae. (Typically, disc calcification would also be expected)

Fig. 2a, b T1- and T2-weighted sagittal MRI of lumbar spine. Low signal is seen in all the discs. Multiple prolapses are demonstrated in b

MRI findings were first reported by Ryan et al. [6]; Hamdi et al. [7] have since reported two further cases [7]. The striking feature is the multiple levels of disc prolapse with the uniformly prominent low signal on T2-weighted images in all the discs, suggesting generalised degeneration. Generalised osteophytic changes, as seen on the plain films are common in elderly patients but not in the young.

Although alkaptonuria is rare, similar plain film and MRI findings may lead to suspicion of the correct diagnosis, especially in patients who may not exhibit other features of the disease.

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