



Case Report

Neglected alkaptonuric patient presented with low back pain and radiculopathy: A case report

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ABSTRACT

Background: Alkaptonuria (AKU) is a rare hereditary disorder in which excess homogentisic acid (HGA) deposits in connective tissues (ochronosis). Here, we report the unusual presentation of a lumbar disc herniation occurring in a patient with AKU warranting surgical intervention.

Case Description: A 28-year-old male presented with 1 year of low back pain. The lumbar magnetic resonance imaging showed an extruded disc at the L4-L5 level accompanied extensive disc space narrowing and osteophyte formation. At surgery, the interspinous ligaments, facet joints, and disc herniation were black. In addition, the postoperative re-examination revealed a black discoloration of the nasal and ear cartilage. Finally, the diagnosis of AKU was confirmed when the urine specimen was positive for HGA.

Conclusion: Rarely, younger patients with AKU who develop excess black deposits of HGA in connective tissues (i.e., ochronosis) may present with lumbar disc herniations and spondylosis.

Keywords: Alkaptonuria, Disc herniation, Low back pain, Radiculopathy

INTRODUCTION

Alkaptonuria (AKU) is a rare autosomal recessive inheritance disorder attributed to dysfunction of the third enzyme in tyrosine degradation (i.e., homogentisic acid [HGA] dioxygenase).^[8] It involves chromosome 3q21-q23 that encodes homogentisate 1,2-dioxygenase.^[4,5] Oxidation and polymerization of HGA contribute to the black discoloration of urine, connective tissues, cartilaginous tissues, the sclera, intervertebral disk, and large joints.^[4] Further, AKU deposition can promote earlier and more extensive thoracolumbar degenerative changes. Here, we report a 28-year-old male with AKU that was undiagnosed until he underwent surgery for lumbar disc herniation where multiple structures (i.e., interspinous ligament, facet joint, and disc) were “black.”

CASE REPORT

A 28-year-old-male presented with 1 year of low back pain and a left foot drop (3/5 level). The lumbar magnetic resonance (MR) imaging revealed a disk extrusion at the L4-L5 level,

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Table 1: Demographic characteristics of previously reported cases and the present case.

Author	Age/sex	Symptoms	Physical examination	Laboratory examination	Diagnostic imaging		
					X-ray	CT	MRI
Choudhury et al., 2000 ^[1]	35/M	Low back pain and right leg pain	No pigmentation	Ochronotic pigment (+) in the urine	Decalcification of vertebral bodies, disc space narrowing, and endplate sclerosis	Unavailable	Multiple prolapsed discs
Farzannia et al., 2003 ^[3]	30/F	Low back pain and left leg pain	Discoloration of nasal and ear cartilages	Positive HGA in the urine and dark urine after waiting	Unavailable	L4-5 disc protrusion	Unavailable
	28/M	Lumbago and right leg pain	Discoloration of sclera and nose darkening	Positive HGA in the urine	Unavailable	Unavailable	L5-S1 disc protrusion
	36/M	Low back pain and left leg pain	Discoloration of nasal and ear cartilages and fingernails	Positive HGA in the urine	Unavailable	L5-S1 disc protrusion, osteophytes, and vacuum phenomenon	Unavailable
Gürkanlar et al., 2006 ^[6]	45/M	Low back pain and left leg pain	Discoloration of nasal and ear cartilages	Positive HGA in the urine and dark urine after waiting	Disc space narrowing, osteophytes, and calcification	Protrusions, osteophytes, vacuum phenomenon, and facet degenerations	L4-5 disc herniation, degenerative changes, and disc space narrowing
Kahveci et al., 2013 ^[7]	45/M	Low back pain and left leg pain	No pigmentation	Increased HGA excretion in urine	Disc space narrowing, osteophytes	Unavailable	L3-4 disc protrusion
Present case	28/M	Low back pain and left foot drop	Discoloration of nasal and ear cartilages	Positive HGA in the urine	Unavailable	Unavailable	L4-5 disc extrusion, disc space narrowing, and osteophytes

CT: Computed tomography, MRI: Magnetic resonance imaging, HGA: Homogentisic acid

disk space narrowing, and osteophyte formation out of proportion to the patient's young age [Figure 1]. When the patient underwent an L4-L5 laminectomy, the interspinous ligaments, facet joints, and disc were black [Figures 2 and 3]. Further clinical examination revealed a black discoloration of the nasal/ear cartilages. Finally, the urinalysis was diagnostic for AKU.

DISCUSSION

AKU is a hereditary autosomal recessive metabolic disorder that occurs due to the failure to metabolize HGA (i.e., HGA oxidase deficiency). The excess of HGA is excreted in the urine, sweat, and deposits in the tissues; this is responsible for the formation of gray pigmentation termed ochronosis.^[2] Ochronosis usually presents in the ear and sclera at first, especially in young adults, followed by low back pain, leg pain, and finally to accelerated spinal degeneration.^[1,3,6,7] Disk degeneration usually appears in the third to fifth decades of age.^[9]



Figure 1: Lumbar magnetic resonance imaging images in a 28-year-old patient in the sagittal (a) and axial (b) planes show disk protrusions (white arrows), disk extrusion (black arrow in a and asterisk in b), Schmorl's node (white asterisk in a), and Modic degenerative changes at vertebral end plates (black asterisks in a).



Figure 2: Interspinous ligaments and facet joints were black.



Figure 3: The extracted disc material was black and degenerated.

Radiographic findings of AKU

Plain X-ray, computed tomography, and MR scans of the spine (i.e., especially the lumbar spine) typically document osteoporosis, calcification of the interspinous ligament, decreased disk space height, and disk calcification.^[1,7]

Surgery for AKU

The diagnosis of AKU, although rare, is usually confirmed during discectomy as the ligaments, facet joints, and disc are typically black.^[1,6,9] Here, we reviewed demographic features of the previous cases and retrospectively diagnosed with AKU by the observation of a “black” disc removed at surgery [Table 1].

CONCLUSION

Early diagnosis of patients with AKU may reveal a black discoloration involving multiple tissues.^[7] Here, a 28-year-old male undergoing surgery for a lumbar disc herniation was diagnosed with AKU when supraspinous ligaments, facet joints, and the disc material were black.

Declaration of patient consent

Patient’s consent not required as patients identity is not disclosed or compromised.

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Conflicts of interest

There are no conflicts of interest.

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