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"Ochronosis with Lumbar disc prolapse-A case report"

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ABSTRACT: Alkaptonuria is a rare autosomal recessive disorder with incidence is as low as 1 in 1000000 births, caused by deficiency of homogentisate 1,2-dioxygenase, an enzyme that converts homogentisic acid (HGA) to maleylacetoacetic acid in the tyrosine degradation pathway. The three major features of AKU are the presence of HGA in the urine, ochronosis (bluish-black pigmentation in connective tissue), and arthritis of the spine and larger joints.
Lumbar disc herniation requiring surgery is unusual in alkaptonuria, with only a few reports in middle to elderly age group. The authors report a case of 27-year-old patient who presented clinically with chronic low back pain, bilateral leg pain, heaviness in lower legs and claudication distance of 10 minutes. Radiologically, he had multiple level disc degeneration, L3-4 canal stenosis and L4-5-disc herniation causing severe canal compromise for which he was taken up for discectomy and decompression.Intraoperatively ligamentum flavum and nucleus pulposus were found to be having black discoloration.Histopathology of the disc material reported ochronoid pigmentation, his urine was positive for HGA. Alkaptonuria was diagnosed after discectomy.Radiologically and clinically there was no other joint involvement nor any black tissue discoloration, which can occur in AKU, and as such ochronosis does not have any adversities. The patient, who is the youngest so far reported in literature, was symptomatically improved after surgery.

Key Words: Alkaptonuria, inter vertebral disc prolapse, ochronosis spine

I. INTRODUCTION

Alkaptonuria is a rare autosomal recessive disease, resulting from a deficiency of the enzyme homogentisic acid oxidase, involved in the metabolism of two amino acids: phenylalanine and tyrosine (1,7,12,15) with subsequent excretion of homogentisic acid in urine [2,3] or is accumulated in the connective tissues.

In the absence of this enzyme, an accumulation of ochronotic pigment occurs. Alkaptonuria may be asymptomatic or cause ochronosis, which is characterized by the accumulation of a pigment-like polymer of the homogentisic acid – the alkapton – on organic tissues like skin, the teeth, the nails, and the patient's buccal mucosa, hyaline cartilage of large joints and intervertebral discs (2,4, 9, 10,12). The oxidation and the polymerization of homogentisic acid leads to black coloration of standing urine and all connective tissues where it is deposited [2,3].

The incidence of alkaptonuria in general population is approximately 1:1000.000 births, with no ethnical prevalence (5,7,8,16,17). Approximately half the number of alkaptonuria patients develops ochronosis, with a male prevalence of 2:1, usually after the fourth decade of life (6,18).

With the advance of age, usually in the third and fourth decade, severe degenerative disorders occur in the joints and the spinal column, mainly in the thoracic and the lumbar area [2,11]. It has been reported that arthropathy develops in approximately 30% of cases of alkaptonuria. (13). Although intervertebral disc degeneration and calcification is frequently seen in alkaptonuria and ochronosis, (3) only a few patients treated surgically for prolapsed lumbar disc have been reported (2,3,1219,20). The spinal involvement results in kyphosis, height loss, and decreased lumbar flexion, and decreases the range of motion and causes effusions [15]. The metabolic disorder does not reduce the normal life span of the patients; however, there is a high rate of disability, especially later in life. (14)

II. CASE REPORT

A 27-year-old man presented with 2-year history of low back pain, bilateral leg radiation and tingling numbness in the left lower leg for 1 year and claudication distance of less than 10 minutes, at the outpatient department of authors hospital.History was otherwise unremarkable, neurological examination revealed left leg L4 and L5 dermatome hypoesthesia and hypoalgesia and normal motor power. Straight leg raising test result was negative. There were no abnormalities in results of hematologic and biochemical laboratory

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studies. Plain radiographs of thelumbar spine showed significant narrowing of lumbar disc (Figure 1,2). MRI scan showed prominent degenerative changes and narrowing in all lumbar disc spaces, stenosis of L3-4 and L4-5 with prolapsed disc at L4-5 (Figure 3).

Figure 1 **Figure 2**

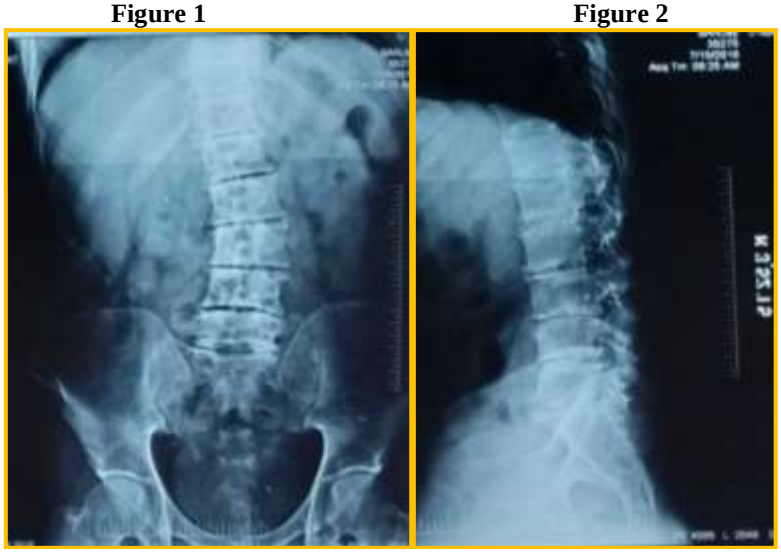
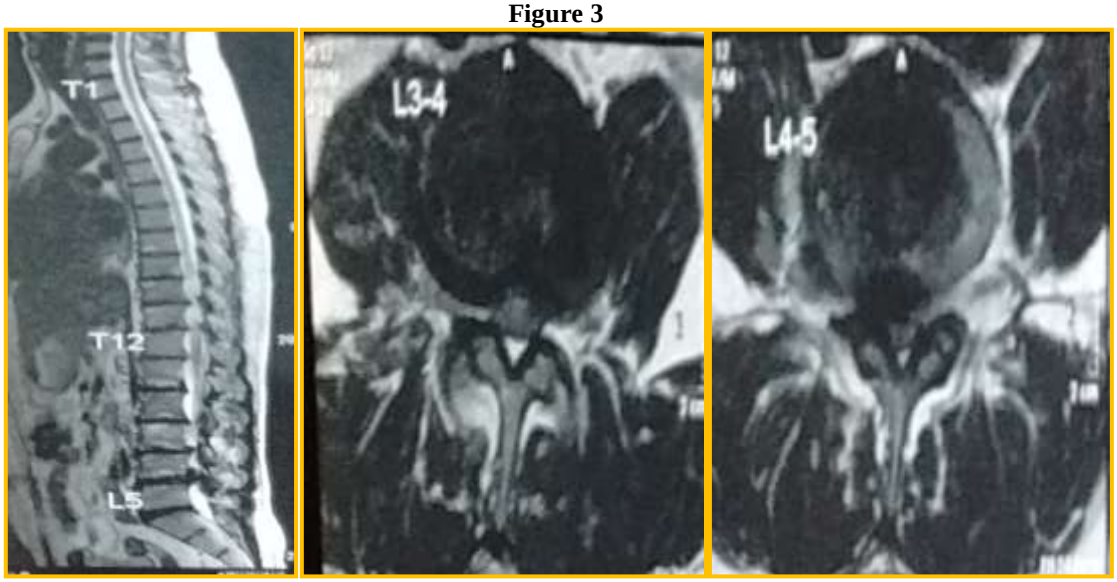


Figure 3



Decompression was achieved by L4 complete laminectomy and L3, L5 hemilaminectomy, lateral recess decompression at both levels along with L4-5 discectomy from right annulotomy. There was blackishdiscoloration of ligamentum flavum and when anulus pulposus was incised, the disc material and the nucleus pulposus removed from the L4-5 disc showed black discoloration (Figure 4). Histopathologic examination confirmed that this material was having ochronotic pigmentation. Retrospectively urine examination was positive for HGA Retrospective examination showed that there was no discoloration of the sclera, cornea, pinna's, skin, or fingernails of the patient. After surgery patient's leg pain and hypoaesthesia were resolved. On follow-up examination after 4 months patient was symptomatically relived.

Figure 4

III. CONCLUSION

Alkaptonuria is a rare metabolic disease which causes early disc degeneration with or without calcification but its presentation with lumbar disc herniation and degenerative lumbar canal stenosis requiring surgical intervention is rarer. It is rarely suspected preoperatively and all cases reported so far are diagnosed retrospectively. The classic features and surgery are no different from routine lumbar surgery for disc prolapsed and stenosis.

IV. DISCUSSION

There is no difference in clinical and surgical management of patients with prolapsed intervertebral disc with ochronosis. We should always have a suspicion of ochronosis in cases of disproportionate disc or dorsolumbar spine degeneration. In such events test for HGA in urine may be performed preoperatively or after detection of black disc. It will be also helpful in prognostication for other large joint involvement in ochronosis. It has no effect on the lifespan of the patient but patient may have disability. Ours is the youngest patient reported so far.

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