Case Report

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Ochronosis of the Lumbar Spine: A Case Report and Review of Literature

Orthopaedics Section

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ABSTRACT

Alkaptonuria is an uncommon genetic condition inherited in an autosomal recessive pattern, characterised by a disruption in the metabolism of phenylalanine and tyrosine, leading to insufficient levels of the enzyme homogentisate and further causes a buildup of Homogentisic Acid (HA). This further leads to blackish discolouration of connective tissues where the acid accumulates. Initially, manifesting as alkaptonuria, it progressively develops into ochronotic arthropathy, which primarily affects the large joints and vertebral discs. Spinal involvement can lead to kyphosis, reduction in height, decreased lumbar flexibility, limited range of motion, and joint effusions. We present a case of a female in her late sixties with complaints of pain in her lower back radiating to her bilateral thighs. Past history revealed dark urine stains on her clothes; she underwent bilateral total hip arthroplasties. On further investigation, she was diagnosed with lumbar disc herniation secondary to ochronosis of the spine. The patient was taken up for L4-L5 decompression. Postoperatively, the patient had improvement of symptoms with no new complaints. We have also included a concise literature review of spinal ochronosis, emphasising the diagnostic hurdles and therapeutic methods associated with this rare condition.

Keywords: Alkaptonuria, Connective tissue diseases, Low back pain, Spinal diseases

CASE REPORT

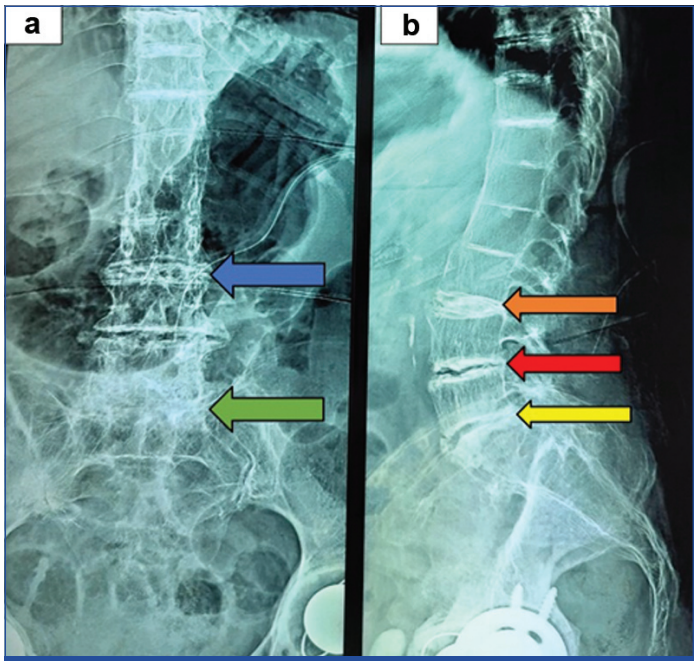
A female in her late sixties, a homemaker by occupation, presented to our department with complaints of pain in her lower back radiating to her bilateral thighs. She also complained of difficulty in walking. She was apparently normal six months prior, when she developed pain which was gradual in onset and progressive in nature, associated with tingling and numbness over her left lower limb. She had no history of claudication. Initially, she underwent conservative management for the same; however, she came to our department due to persistence of symptoms. Past history revealed dark urine stains on her clothes as a child. She also underwent bilateral total hip arthroplasties around 8-10 years prior due to severe pain. There was no family history of similar illnesses.

Physical examination revealed blackish spots over her sclera and discolouration of her nails [Table/Fig-1a-c].

[Table/Fig-1]: Clinical images showing ochronotic pigmentation in a patient with alkaptonuria: (a) Blackish discolouration of the sclera (blue arrow); (b) Hyperpigmentation of the fingernails (both hands); (c) Hyperpigmentation and blackish discolouration of the toenails (blue arrow).

She had severe tenderness over L4, L5 and S1 regions and over her paraspinal muscles in the corresponding regions. The straight leg raising test was positive over the left side and she had no sensory or motor deficits.

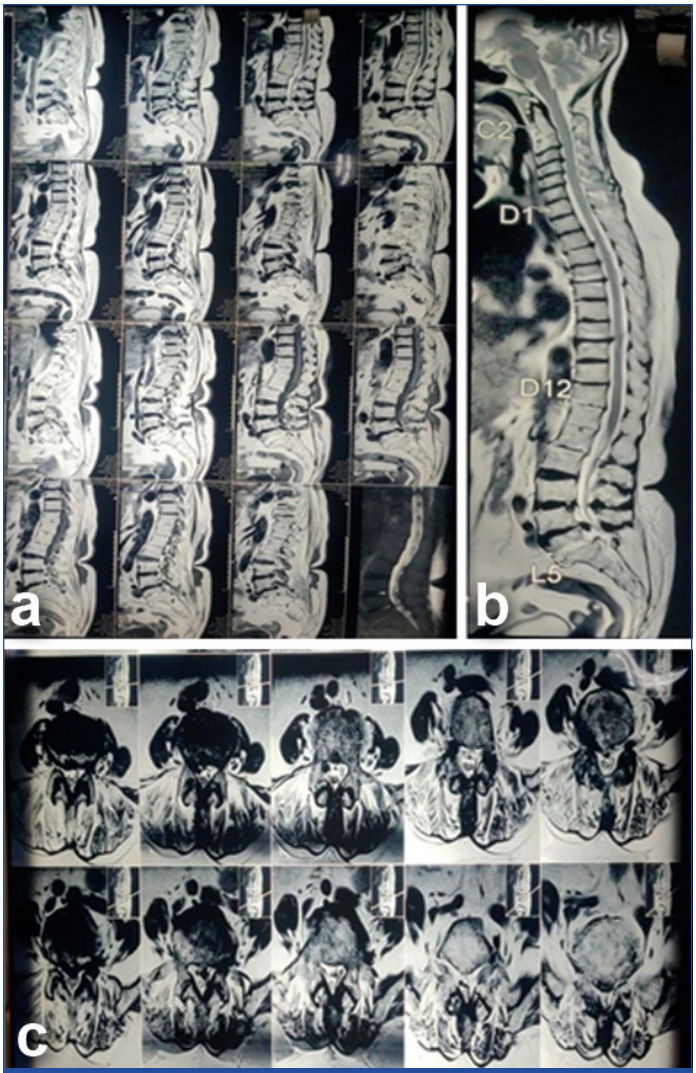
X-Ray of the lumbar spine revealed advanced arthritis with significant reduction in disc spaces with bony ankylosis at D11-D12, D12-L1, L1-L2, L2-L3 with loss of lumbar lordosis and sclerosis at the end plates of D11-D12, L4-L5 levels and vacuum phenomenon [Table/Fig-2a,b].



[Table/Fig-2]: Pre-operative radiographs of the lumbosacral spine: a) Anteroposterior (AP) view: Blue arrow showing advanced intervertebral disc space narrowing and calcification, suggestive of disc degeneration. The green arrow indicates reduced disc height and vacuum phenomenon at the L5-S1 level; b) Lateral view showing multiple levels of intervertebral disc space narrowing with bony ankylosis and vacuum phenomenon. The orange arrow indicates the presence of gas within the disc (vacuum sign), the red arrow indicates reactive sclerosis at adjacent vertebral endplates and the yellow arrow indicates significant disc space collapse.

Magnetic Resonance Imaging (MRI) showed L4-L5, L5-S1 disc protrusion with lumbar spinal cord compression. There was also hypertrophy of ligamentum flavum at L3-L4, L4-L5, and L5-S1 levels, leading to severe spinal canal stenosis [Table/Fig-3a-c].

The patient was taken up for L4-L5 decompression. Under general anaesthesia and with the patient in the prone position, parts were painted and draped. A midline incision was made from L3-L5 levels. Skin and subcutaneous tissue were dissected.



[Table/Fig-3]: Magnetic Resonance Imaging (MRI) of the whole spine: a,b) Sagittal cuts; c) Axial cuts. The images demonstrate reduced height in multiple intervertebral disc spaces, notably at L4-L5 and L5-S1, with disc protrusion causing lumbar spinal cord compression. Additionally, hypertrophy of the ligamentum flavum is observed, resulting in severe spinal canal stenosis.

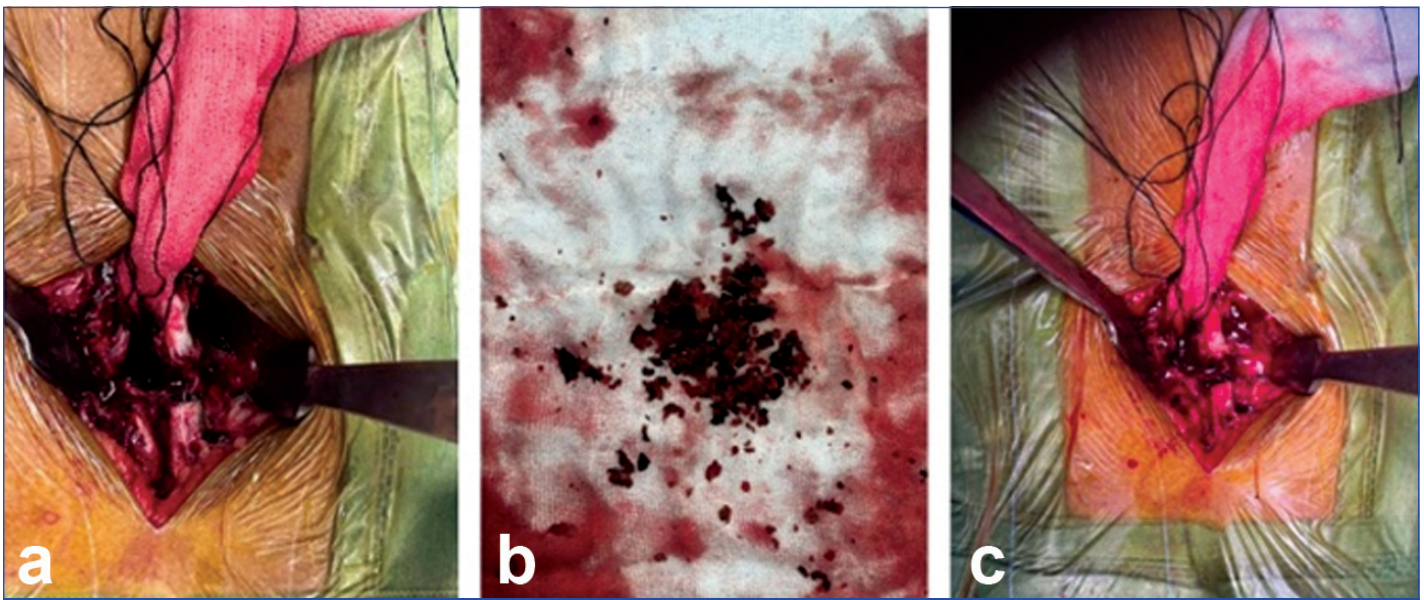
Adequate haemostasis was achieved using electrocautery. L4 hemilaminectomy was performed and ligamentum flavum was found to be hypertrophic along with blackish discolouration. Ligamentum flavum was removed and the disc was found to be firm and blackish [Table/Fig-4a-c], which was removed and sent for histopathological evaluation. L5 laminectomy was performed and similar findings were noted. A thorough wound wash was given and the wound was closed in layers over a Romo Vac drain and anaesthesia was reversed. The operative procedure was 90 minutes with an estimated blood loss of 600 cc. On extubation, the patient was monitored in the Intensive Care Unit (ICU) and was subsequently shifted to the ward on Postoperative Day (POD) 1. On POD 2, she was made to sit bedside and encouraged to do in-bed mobilisation. Postoperative X-ray showed L4-L5 laminectomy defect [Table/Fig-5a,b].

The patient showed significant improvement and enhancement in symptoms following surgery, with no additional neurological issues. The histopathological examination [Table/Fig-6] revealed uniform interstitial brownish-black pigmentation, chondronecrosis, and melanin-like deposits inside chondrocyte cytoplasm with no indications of dysplasia or neoplasia. Subsequently, the patient was referred to the department of general medicine for further evaluation, metabolic workup, and long-term intensive care and counselling. At the 1-month follow-up, clinical assessments, including neurological examination, pain scoring, and functional status evaluation, indicated substantial improvement. The patient reported no neurogenic symptoms and was able to continue day-to-day activities independently, reflecting a positive surgical outcome.

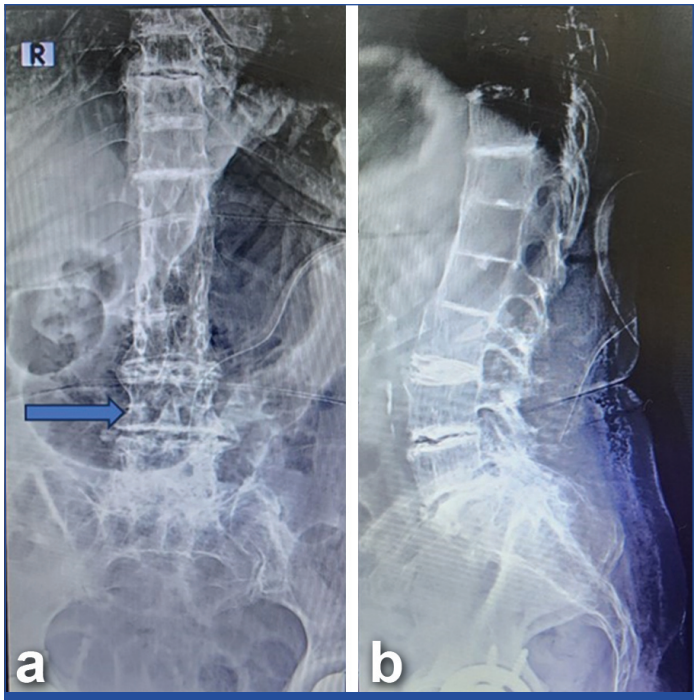
DISCUSSION

Alkaptonuria (AKU) is an uncommon autosomal recessive metabolic condition occurring from a lack of the enzyme homogentisate 1,2-dioxygenase, leading to the buildup of Homogentisic Acid (HGA). HGA accumulates as ochronotic pigment in connective tissues, especially in cartilage, intervertebral discs, and ligaments over time [1]. This results in gradual deterioration, typically manifesting clinically after the third decade of life. The classic triad includes: darkening of urine upon the addition of an alkaline substance, blackish discolouration of connective tissue and progression of arthritis. Spinal ochronosis presents as persistent lower back discomfort, radiculopathy, and occasionally neurogenic claudication, resembling degenerative disc disease or ankylosing spondylitis. Nonetheless, sacroiliac joint fusion, a characteristic feature of ankylosing spondylitis, is conspicuously missing in ochronosis [2]. Radiological indicators may encompass disc calcification, diminished disc height, kyphosis, and the vacuum phenomenon. The intraoperative observations of necrotic, rigid discs and enlarged ligamentum flavum, coupled with histological validation, corroborated the diagnosis.

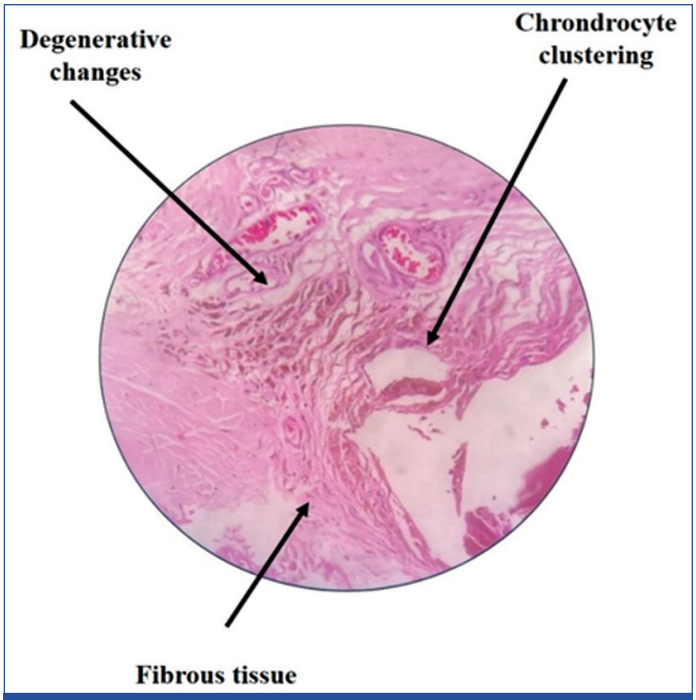
The conclusive diagnosis requires the urine measurement of HGA, often conducted using Gas Chromatography–Mass Spectrometry (GC-MS) [3]. Although not diagnostic, an elevated Erythrocyte



[Table/Fig-4]: Intraoperative Images: a) Exposure of the lumbar spine showing hypertrophy and blackish discolouration of the ligamentum flavum, consistent with ochronotic pigmentation; b) Extracted disc material appearing firm and blackish, indicative of ochronotic degeneration; c) Blackish and thickened tissues seen intraoperatively.



[Table/Fig-5]: Postoperative radiographs on Postoperative Day (POD) 5 of the lumbosacral Spine. (a) Anteroposterior view; and (b) Lateral view demonstrating postoperative changes with evidence of laminectomy defect at the L4–L5 level (arrow in A).



[Table/Fig-6]: Histopathological image of intervertebral disc biopsy showing degenerative changes. The tissue was stained with Haematoxylin and Eosin (H&E) and observed under X100 magnification. The arrow indicates areas of fibrocartilaginous degeneration. A biopsy taken from the lumbar intervertebral disc.

Sedimentation Rate (ESR) is noted in certain cases, sometimes resulting in confusion with seronegative spondyloarthropathy [4].

The treatment mostly addresses symptoms. Vitamin C has been investigated for its antioxidant properties, although it demonstrates inconsistent long-term effectiveness. Nitisinone, initially employed for hereditary tyrosinaemia, has demonstrated encouraging outcomes in decreasing HGA levels and decelerating disease development. Nonetheless, it necessitates vigilant oversight owing to possible deleterious effects associated with tyrosinaemia [5]. Restricting dietary protein and implementing lifestyle adjustments serve as supplementary interventions [6].

To explain this case, we performed a study of 10 relevant case reports with the criteria of inclusion being patients presenting with back pain/ neckpain, diagnosed with ochronosis or AKU with manifestations in the spine and treated with any surgical intervention [Table/Fig-7] [7-15]. The average age of the patients reviewed was 47.3 years (ranging from 31-71 years). Female gender predominance was observed (n=8). The order of presentation from most common to least common was as follows: radiculopathy, low back pain, neck pain, and neurogenic claudication. The most commonly affected region was the lumbar region (n=7), followed by the cervical region (n=3), with L4-L5 being the most common

S. no.	Author	Year	Age/Sex	Symptoms	Imaging findings	Biopsy findings	Spine level	Treatment	Outcome
1.	Nelanuthala M et al., [7]	2018	38/F	Tingling, numbness in the bilateral upper limbs	Degenerated C3-C4, C6-C7 discs with prolapse	Disc and ligaments black	C3-C4, C6-C7	ACDF	Symptom improvement
2.	Baradaran Bagheri A et al., [8]	2019	45/F	Persistent back pain, left radiculopathy	Osteophytes, reduced disc space, prolapsed disc	Ochronotic nucleus pulposus	L3-L4	Discectomy	Improved functionality
3.	Jayakumar S et al., [9]	2019	34/F	LBP, right radiculopathy, motor deficit	Disc narrowing, annular bulge, paracentral extrusion	PLL bluish, nucleus pulposus black	L5-S1	Laminectomy and Discectomy	Pain and neurological improvement
4.	Yucetas SC et al., [10]	2019	71/M	LBP, neurogenic claudication	Calcification, narrowed disc space, lateral recess stenosis	Degenerated and black ligamentum flavum	L3-L4, L4-L5	Laminectomy (L3-L4, L4-L5)	Symptom relief
5.	Alisi MS et al., [11]	2020	55/F	Neck pain, radiculopathy, low back pain	Calcification, cervical myelopathy, lumbar degeneration	Stiff black disc	C3-C4, C4-C5	2-level ACDF	Neck pain and paraesthesia improved
6.	Bansal ML et al., [12]	2023	31/F	Chronic LBP, left radiculopathy	Degenerative changes, calcification, disc prolapse	Black disc, chromatin staining in chondrocytes	L4-L5	TLIF	Symptom relief
7.	Ding H et al., [13]	2023	61/F	Chronic back pain, bilateral radiculopathy	Degenerative changes, spinal canal stenosis	Blackened bone-ligament junctions	T10-T11, L1-L2, L2-L3	Decompression + stabilisation	Pain reduction, improved power
8.	Broekx S et al., [14]	2024	46/M	Neck pain	Cervical stenosis, disc herniation	Black nucleus pulposus	C6-C7	ACDF	Symptom relief
9.	Sang P et al., [15]	2025	49/F	Chronic LBP, right radiculopathy	Calcification, vacuum, spondylolisthesis, disc herniation	Ochronotic pigmentation of the disc and tissue	L5-S1	TLIF	Neurological improvement
10	The present case	2024	68/F	Lower back pain, left radiculopathy	Bony ankylosis, calcification, sclerosis, vacuum, disc protrusion	Hypertrophic black ligamentum flavum and disc	L4-L5, L5-S1	L4 Hemilaminectomy, L5 Laminectomy	Significant improvement

[Table/Fig-7]: Summary of reported ochronosis spine cases [7-15].
ACDF: Anterior cervical discectomy and fusion; LBP: Low back pain; PLL: Posterior longitudinal ligament; TLIF: Transforaminal lumbar interbody fusion

level affected, followed by L5-S1. All patients required surgical intervention in the form of decompression, Transforaminal Lumbar Interbody and Fusion (TLIF), and Anterior Cervical Discectomy and Fusion (ACDF). All patients had symptomatic improvement and a good outcome following surgery.

CONCLUSION(S)

The present case underscores that spinal ochronosis, although rare, must be considered in patients with lumbar disc prolapse and intraoperative dark pigmentation. Timely surgical decompression offers symptomatic alleviation, with histopathological analysis corroborating the diagnosis. Recognition during surgery is essential for postoperative management, counselling and lifestyle modifications. A prompt diagnosis can avert long-term neurological impairment and enhance quality of life. Timely identification facilitates the screening for systemic involvement, especially cardiac and renal symptoms, which may accompany ochronosis. An expedient and thorough strategy can prevent enduring neurological deficits, diminish the likelihood of recurring symptoms, and eventually improve the patient's quality of life.

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