Journal Homepage: - www.journalijar.com
INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)
Article DOI:10.21474/IJAR01/2609
DOI URL: http://dx.doi.org/10.21474/IJAR01/2609
ISSN NO. 2320-5407

RESEARCH ARTICLE

ALKAPTONURIC PATIENT PRESENTING WITH BLACK DISC AND MULTIPLE SCHMORL'S NODES.

Dr. Sree Harsha. C. K (M. S. Ortho, FNB spine), Dr. Venkata Ramakrishna Tukkapuram (M. S. Ortho, fellow in spine surgery).

Manuscript Info

Manuscript History

Received: 28 October 2016
Final Accepted: 27 November 2016
Published: December 2016

Abstract

The clinical manifestation of ochronosis is blackish discolouration of collagen due to binding of pigment benzoquinone to collagen which leads to degradation and destruction of cartilage. In spine, it involves the disc, endplates and ligaments leading to early degeneration. We present a case where patient presented with short history of right lower limb radiculopathy and weakness. MRI showed osteolytic lesion at L5 right sub pedicular region and multi level disc degeneration with multiple schmorl's nodes. Differential diagnosis of osteoid osteoma was considered. Alkaptonuria was diagnosed intraoperatively after encountering black disc. Here we describe the clinical and radiological features with review of relevant literature.

Introduction:-

Alkaptonuria is a rare metabolic disease. It is the first disease to have Mendelian recessive type [10]. It is a hereditary inborn error of metabolism due to mutation of HDG gene (homogentesate1,2dioxygenase). The deficiency of enzyme HDG in aromatic amino acid degradation pathway causes increased blood levels of HGA (homogentisic acid) [1]. HGA is excreted in urine, which on standing turns black. Ochronosis is deposition of pigmented benzoquinone in tissues. Benzoquinone is a polymeric oxidation product of HGA [9]. This irreversible binding to collagen causes early degeneration of connective tissue which becomes less resilient to stress and weight bearing. Therefore, early degeneration of spine is seen in alkaptonuric patients but, disc prolapse with foraminal stenosis is not a common presentation [3-8].

We report an alkaptonuric patient with multiple level disc degeneration, multiple schmorl's nodes, disc prolapse with right foraminal stenosis at L5-S1. However, the diagnosis was made retrospectively, after finding a black colored disc intraoperatively.

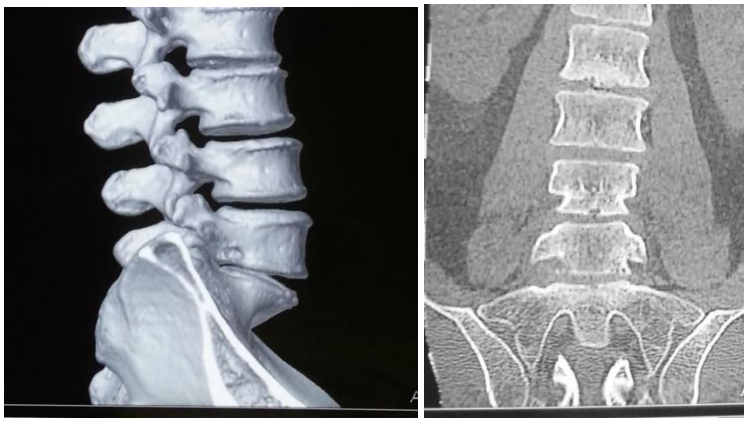
Case Report:-

A 41-year-old male patient, farmer by occupation presented with low back pain and stiffness since one month. Pain started radiating to right lower limb and patient was having difficulty in standing and walking since 3 days. Clinical examination revealed positive straight leg raising test on right side, right EHL/EDL were weak (grade 3/5). There was hypoesthesia over L5 and S1 dermatomes on the right side. X ray showed multiple level degenerative disc disease. MRI showed multiple level disc degeneration with multiple schmorl's nodes. A soft tissue mass was noted on the right infra pedicular region which was extending into the foramen causing severe stenosis. The well subscribed mass aroused suspicion of osteoid osteoma. CT scan was done which revealed a well-defined lytic lesion but there was no nidus.

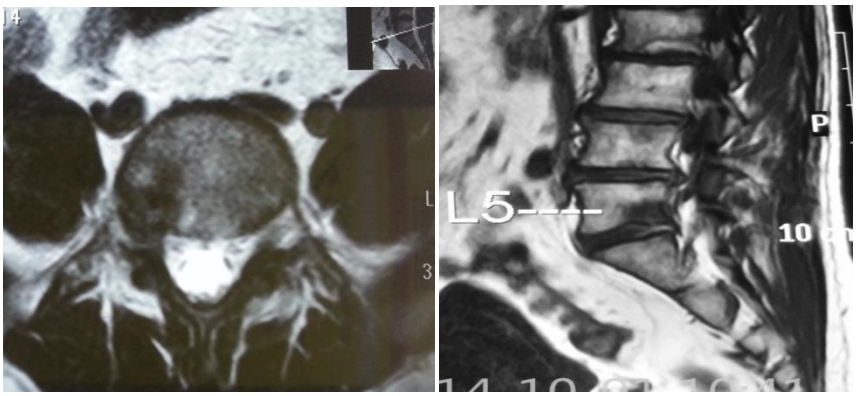
Corresponding Author:-Dr. Venkata Ramakrishna Tukkapuram.

Patient was scheduled for exploration and removal of the mass. As the mass was under the pedicle, unilateral facetectomy was planned to better expose the lesion. On exposure and probing of the lesion, black coloured disc material was encountered. Intraoperatively, the black discolored disc was found herniated into the body of the vertebra in the subpedicular region. The disc was also causing severe foraminal compression of the exiting L5 nerve root. Discectomy was done and unilateral stabilization and inter transverse fusion was done. Sample was sent for histopathological examination which confirmed the diagnosis. Postoperatively, on further questioning the patient gave history of urine turning black during his younger days. For past many years he had not noticed it. The clinical picture and the intraoperative findings pointed towards Alkaptonuria.

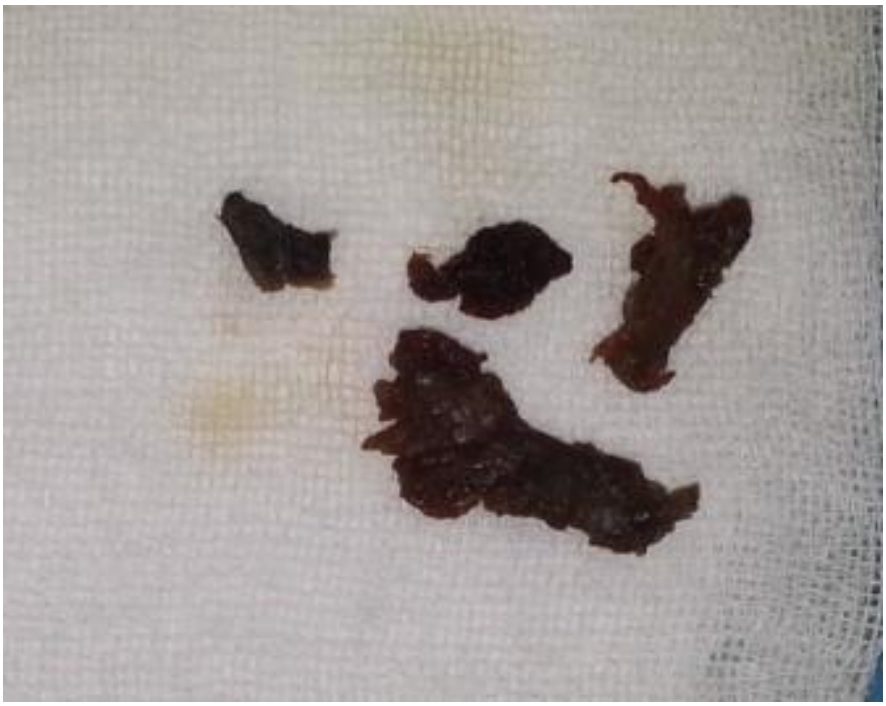
Patient had complete relief of pain in the immediate post-operative period. Neurological deficits also improved and by 6 weeks patient had attained normal neurological status. The postoperative recovery was good with immediate pain relief.



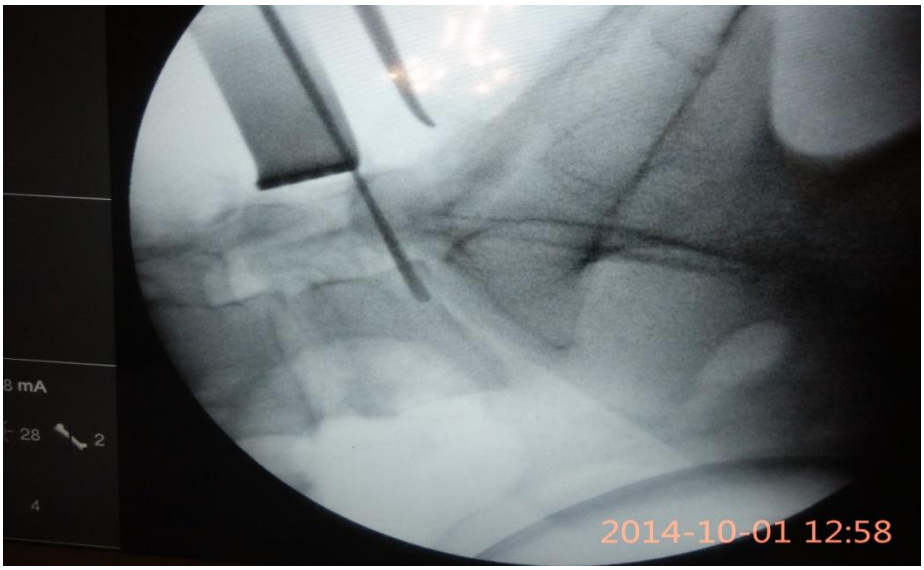
CT scan shows osteolytic lesion at L5 rt sub pedicular region.



MRI shows soft tissue mass extending into rtforamen(causing stenosis) and L5 body



Black coloured disc and endplate retrieved.



C-arm image shows bony deficit at rt L5subpedicular region.

Discussion:-

Alkaptonuria is a rare and a first autosomal recessive condition to be described. The incidence is as low as 0.001% [15]. Allele was mapped to 3q21-q23 which encodes homogentisic acid oxidase [16]. This disease was first described by Scribonius in 1584. First symptom related to spine is back stiffness and back and lower back pain [17]. First symptom was back pain in 60% of cases and sciatica in 17% cases in a study[17]. The degenerative changes are due to the accumulation of benzoquinone in cartilage of joints and intervertebral discs. Spinal manifestation in Alkaptonuria manifests as early degeneration of spine with disc calcification, blackish discoloration of ligaments, intervertebral disc and endplates. Vacuum phenomenon has been reported in few case reports [2,18,19]. Vacuum phenomenon is a radiolucent area found in disc apace which is seen in intervertebral osteochondrosis along with

large disc herniation and schmorl's nodes [2]. The whole spine may be involved, the most common being the lumbar region[17]. A case was reported with T1 hyperintense disc herniation suggesting calcification of disc, which was confirmed on CT scan [20].

Clinically, the spine becomes less flexible, lumbar lordosis is flattened and thoracic kyphosis becomes more prominent which mimics ankylosing spondylitis, which can be differentiated with bamboo sign on x ray, positive HLAB27 and sacroiliac joint involvement [11,17]. It is a multisystem disorder [13], it has a long-term impact on myocardium, renal parenchyma, cartilage, sclera, skin and spine. By diagnosing it early, the long term damage to the multisystem can be prevented by medical management[21] by decreasing phenylalanine and tyrosine intake and having vitamin c rich diet and supplements and occasionally nitisinone.

As the degeneration of spine is quite common in alkaptonuric patients, the disc herniation is seldom encountered [3-8]. The radicular symptoms due to disc herniation can be treated by surgical decompression [10] of the nerve root and stabilization if needed. The disc herniation in Alkaptonuria, requiring surgery are rarely reported. Here we have presented one such case in which the diagnosis was made retrospectively. In our case report, osteolytic lesion in right L5 subpedicular region was seen. As there was a soft tissue signal inside the lytic lesion, osteoid osteoma was suspected. CT scan was taken to look for nidus if any, to rule out osteoid osteoma. L5-S1 decompression, stabilization and intertransvesre fusion was performed. The patient was diagnosed of alkaptonuria retrospectively. The patient gave a positive history of blackish discolouration of urine and on examination, his sclera shows black patchs. The sclera discolouration is refered to as osler's sign [14]. Patient symptoms improved postoperatively. Patient symptoms subsided post operatively was discharged on 3rd post operative day. Patient was followed up till 6 months. Patient was able to perform daily routine and had returned to his occupation .

Conclusion:-

The patient presenting with chronic back pain, stiffness and early degeneration of spine with multilevel schmorl's and calcified disc, consider Alkaptonuria as a differential diagnosis.

References:-

1. Garrod AE. The incidence of alkaptonuria: a study in chemical individuality. Lancet 1902;2:1616-20.
2. 2.SV PHATAK, PK KOLWADKAR, TJ HEMNANI, MS PHATAK, JT HEMNANI Pictorial essay: AlkaptonuriaInd J RadiolImag 2006 16:4:573-576
3. Reddy DR, Prasad VS. Alkaptonuria presenting as lumbar disc prolapse. A case report and review of the literature. Spinal Cord 1998;36:523-4.
4. Emel E, Karagöz F, Ayd›n H, Hac›saliho¤lu S, Seyithano¤lu MH. Alkaptonuria with lumbar disc herniation. A report of two cases. Spine (Phila Pa 1976) 2000;25:2141-4.
5. Choudhury R, Rajamani SS, Rajshekhar V. A case of ochronosis: MRI of the lumbar spine. Neuroradiology 2000; 42:905-7.
6. Farzannia A, Ghaffar S, Hadidchi S. Alkaptonuria and lumbar disc herniation. Report of three cases. J Neurosurg 2003; 98(1 Suppl):87-9.
7. Gürkanlar D, Daneyemez M, Solmaz I, Temiz C. Ochronosis and lumbar disc herniation. ActaNeurochir (Wien) 2006;148:891-4.
8. Laskar FH, Sargison KD. Ochronoticarthropathy. A review
9. La Du BN. Alkaptonuria. In: Scriver CR, Beaudet AL, Sly WS, Valle D, editors. The metabolic and molecular basis of inherited disease. 8th ed. New York: McGraw-Hill; 2001. p. 2109-23.
10. Goldsmith L. Disorders of metabolism. In: Fitzpatrick T, Arthur E, Wolff K, Irwin F, Frank A, editors. Dermatology in General Medicine, 3rd ed. New York: McGraw-Hill Book Co; 1987. p. 1642-6.
11. Ramazan KAHVEC‹1, Mehmet Fikret ERGÜNGÖR2, Ahmet GÜNAYDIN2, Atiye TEM‹Z3Alkaptonuric patient presenting with "black" disc:a case report ActaOrthopTraumatolTurc 2013;47(2):134-138 doi:10.3944/AOTT.2013.2767.

12. Sharon E. Albers, MD [1], Stephen J. Brozena, MD [1], L. Frank Glass, MD [1], Neil A. Fenske, MD [1,]Alkaptonuria and ochronosis: Case report and reviewVolume 27, Issue 4, October 1992, Pages 609-614
13. Alkaptonuria and ochronosis: Case report and reviewAuthor links open the overlay panel. Numbers correspond to the affiliation list which can be exposed by using the show more link.Sharon E. Albers, MD , Stephen J. Brozena, MD , L. Frank Glass, MD , Neil A. Fenske, MD
14. Osler's sign revisitedAnnapurna Kumar, Kaliaperumal Karthikeyan,[1] and Malhar T. VyasAuthor information ► Copyright and License information ►Indian Dermatol Online J. 2015 Jul-Aug; 6(4): 308–309.
15. .GürkanlarD ,Daneyemez M , Solmaz I , et al . Ochronosis and lumbardisc herniation .*ActaNeurochir (Wien)* 2006 ; 148 : 891 – 4 .
16. Fernandez-Canon JM ,Granadino B , Beltran-Valero de Bernabe D , et al. The molecular basis of alkaptonuria .*Nat Genet* 1996 ; 14 : 19 – 24 .
17. Feild JR, Higley GB, DeSaussure RL. Ochronosis with lumbar disc: Case report. J Neurosurg 1963;20:348–51.
18. Deeb Z, Frayha RA. Multiple vacuum discs. An early sign of ochronosis: Radiologic findings in two brothers. J Rheumatol 1976;3:82–5.
19. Millea TP, Segal LS, Liss RG, Stauffer S. Spine fracture in ochronosis: Report of a case. ClinOrthop 1992;281:208–11.
20. T1 Hyperintense Disc in Alkaptonuria Alan A. Sag , MD , * Richard Silbergleit , MD , * Rick E. Olson , MD , † Jon Wilson , MD , ‡ and Anant Krishnan , MD * SPINE Volume 37, Number 21, pp E1361–E1363 ©2012, Lippincott Williams & Wilkins
21. GrompkeM . Disorders of tyrosine metabolism . In: Basow DS, ed. *UpToDate*. 2009 Accessed May 28, 2010.