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ALKAPTONURIA PRESENTED WITH LUMBAR CANAL STENOSIS A CASE REPORT

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Abstract : Alkaptonuria is a rare autosomal recessive disorder with a low incidence of 0.001. Ochronosis is the pigmentation of connective tissues in alkaptonuric patients and clinical manifestation usually appear after the age of 30. Deposition of pigment causes degenerative changes in cartilage, intervertebral disc and other connective tissues by an unknown mechanism. Although intervertebral disc degeneration is frequently seen in alkaptonuria and ochronosis, only a few patients treated surgically for lumbar disc disease have been reported

Keyword : Alkaptonuria, discectomy, lumbar canal stenosis, benedict test

INTRODUCTION

Alkaptonuria is a rare autosomal recessive disorder resulting from congenital deficiency of the enzyme homogentisic acid (HGA) oxidase. Due to lack of HGA oxidase enzymatic activity, HGA cannot be degraded and it accumulates in cartilage and connective tissues for years, then it forms blue-black pigmentation. Deposition of the pigment causes degenerative changes in cartilage, intervertebral disc and other tissues. Although intervertebral disc degeneration is common in alkaptonuria, our review of the literature introduced only 13 patients, including ours, were treated surgically for lumbar disc herniation. Diagnosis of alkaptonuria after black colored lumbar discectomy material is a rare condition.

CASE REPORT

A 40 years old male was admitted with complaints of low back pain for 6months and symptoms of neurogenic claudication. Pain was progressively increased in severity. On clinical examination patient had tenderness L3-L4 level, straight leg raising test was negative and there was no neurological deficit. Xray Lumbosacral spine shows diffuse sclerotic changes on both end plates of lumbar spine and calcification of disc spaces. MRI study of lumbar spine shows L3-L4 canal stenosis with diffuse disc dehydration on all disc levels. Due to diffuse dehydration on all levels in MRI, patient was evaluated for alkaptonuria. There were no clinical signs suggestive of alkaptonuria, but urine turned dark on standing in air for 6 hrs and Benedict's test was positive. Patient had undergone L3-L4 discectomy under GA. Intraoperatively supraspinous and interspinous ligament was found to be black in colour. Adequate decompression of neural structures was achieved. During L3-L4 discectomy it was realized that the extruded disc material was found degenerated and black in color. Post operatively patient was relieved of neurogenic claudication. On 6 months follow up, patient had been relieved of symptoms of claudication with mild low back pain.

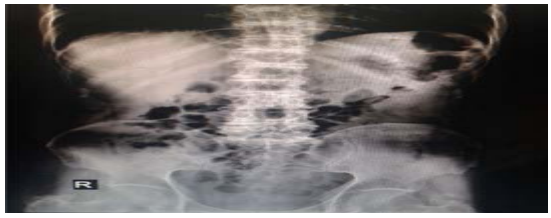

Fig-1

Fig-2

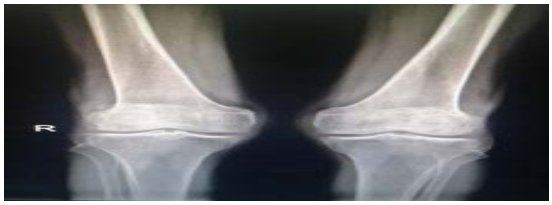

Fig-3

Fig-4

Fig-5

Fig-6

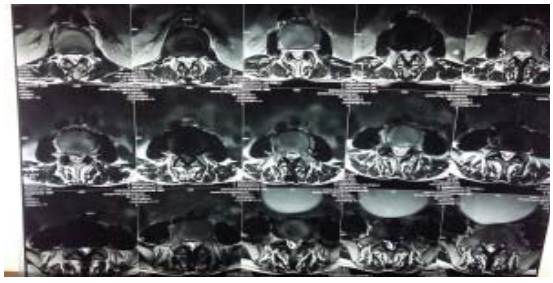

Fig-7

Fig-8 Benedict Test

Fig-9 Urine Turns Dark

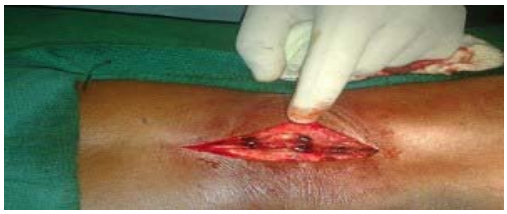

Fig-10 Supra spinous ligament Black in colour

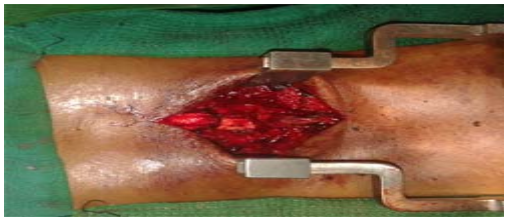

Fig-11 Discectomy done

Fig-12 Disc Material black in colour

DISCUSSION

Alkaptonuria was first described in 1584 in a child whose urine was black. The alkaptonuria gene 3q21-q23 has been found to be responsible for homogentisic acid oxidase, an enzyme in metabolic pathway of aromatic aminoacids. A black pigment produced by oxidation and polymerization of homogentisic acid deposits in all connective tissues. This pigment has a high affinity with fibrillary collagens surrounded by abundant mucopolysaccharide substances, such as that of hyaline cartilage of the major peripheral joints and intervertebral discs The clinical manifestation of ochronosis consist of blue pigmentation of external ear and tympanic membrane, black discolouration of cerumen, blue, blackor brown staining of sclera, blue to black tinting of skin in axillary and genital region. Accumulation of this molecule in the cartilage of joints and intervertebral discs causes degradation of cartilage. These changes may lead to intervertebral disc herniations, most prominently involves the lumbar region. Stiffness of lumbar spine, gradual loss of lordosis and exaggeration of thoracic kyphosis are the initial presenting signs. Vacuum disc phenomenon presumably represents areas of severe degeneration

within the disc. More rarely, it may begin with acute backache resulting from rupture of the annulus fibrosus. The first symptom was sciatica in approximately 17% of patients with alkaptonuria in one report and was back pain in 60% of patients in another. In alkaptonuric spondylosis, degenerative changes may be seen along the whole of the spine; however the most common involvement is in the lumbar spine. The most common radiological changes of alkaptonuric spondylosis include squaring of vertebral bodies, intervertebral disc calcification, bridging syndesmophytes, and apophyseal joint involvement. When changes are advanced, the radiologic appearance is like bamboo spine. The disease usually progresses from simple alkaptonuria to alkaptonuric ochronosis and finally to alkaptonuric arthropathy. Ocular pigmentation is especially prominent and appears in approximately 70% of the patients. It has been reported that arthropathy develops in approximately 30% of cases of alkaptonuria. Although intervertebral disc degeneration is frequently seen in alkaptonuria and ochronosis, only a few patients treated surgically for lumbar disc disease have been reported. Peripheral joint arthritis develops in the later stage and generally large joints are involved. Involvement of small joints is rare. Renal functions may be deteriorated in patients with alkaptonuria due to accumulation of pigment in prostate. Calcification in prostate gland may cause an obstruction and renal dysfunction due to obstruction may occur. Cardiovascular involvement may also occur in alkaptonuric patients. Systolic murmurs were found 15- 20% of the patients. Aortic and mitral valve involvement is usually seen in alkaptonuria. Also atherosclerotic plaques may develop in these patients, in whom myocardial infarction is the most common cause of death

CONCLUSION:

There is no effective treatment for alkaptonuria. In medical treatment, phenylalanine and tyrosine deficient diet as well as high dose of ascorbic acid is suggested. Non-steroid anti-inflammatory drugs may be required for arthropathy. As a conventional treatment, physical therapy and rehabilitative interventions can cause notable symptomatic relief. In addition, the use of antioxidants such as Nacetyl cysteine and vitamin E has been showed to reduce HGA accumulation in vitro. Consequently, black disc material is a suspicious value for alkaptonuria in patients who undergo lumbar discectomy. These patients are candidates of other systemic involvements of alkaptonuria. After lumbar discectomy, where black disc material has been found, diagnosis of alkaptonuria is essential. These patients must be placed on long term follow-up. Therefore, the spine surgeons should keep in mind the metabolic disorders such as alkaptonuria in the differential diagnosis of degenerative disc disease Because only a few alkaptonuric patients treated surgically for lumbar disc disease have been reported, little is known about outcome. The review of the literature showed that all alkaptonuric patients, including ours, were significantly improved after lumbar discectomy. So, lumbar diskectomy has been helpful in this patient group.

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