RESEARCH PAPER | Medical Science | Volume : 5 | Issue : 6 | June 2015 | ISSN - 2249-555X



Alkaptonuria - A Case Report

KEYWORDS

Dr.Arindam Sur

SCB Medical College,Cuttack

A Patient named, Mr. Bijay Kumar Singh,aged 42 yrs was referred to the Dept of Biochemistry, from the Dept. of Orthopedics OPD presents with **Chief complains** of –

1. low back pain for the last 20 yrs.
2. Restricted movement of lumbar region for the same duration.
3. Darkening of urine on standing also for the same duration.
4. Pain & restricted movement of the knee joint for last 4-5 months

Family History-

- Simillar complaints of 2 brothers.(i.e low back pain,restricted movement of lumbar spine,darkening of urine on standing)
- No history of similar complaints in either of the parents was present
- No history of consanguinous marriage of the parents

Treatment History-

The patient visited the Dept of orthopedics OPD,S.C.B Medical College,Cuttack where he was provisionally diagnosed having ALKAPTONURIA & was given analgesics,antacids,calcium tablets,vitamin C

ON EXAMINATION

Musculo-Skeletal Examination-

- The patient had a stooping posture
- Examination of the spine- Tenderness at the dorsolumbar region and the movements were restricted in all directions

Examination of other joints-

- Painful and restricted range of movements of both knee joints..
- Rest of the joints had satisfactory range of motion
- Local Examination didn't reveal any pigmentation..
- Systemic examination of Cardiological , Respiratory , Central Nervous Systems were within normal limit

RADIOLOGICAL FINDINGS-

- The MRI of the LUMBAR SPINE(with screnning of the whole spine) SHOWS-
- Mild kyphosis with exaggerated lumber lordosis is seen.
- Altered signal intensity with posterior protrusion of L 2-3, L -3,4, L 4-5, L 5-S1 discs are seen.It is more severe in L 4-5 disc.
- Bilateral narrowing of neural foramina with nerve roots entrapment are seen at the said disc levels.
- Thecal sac narrowing with moderate to severe entrapment of CAUDA EQUINA fibres at L 3-4 & L 4-5 discs was observed.
- Screening whole spine shows dessication with posterior protrusion of rest of the discs of cervical region. Mild bulging of few other discs were also seen.
- X ray shows-Diminished disc spaces in all lumbar vertebras.



BIOCHEMICAL TESTS-

1.Benedict's Test- +ve (greenish precipitate)
2.Ferric chloride Test- +ve (forms transient green colour)

HPLC of urine showed presence of Tyrosine.

History,clinical examination,biochemical tests and radiological imaging were suggestive of **alkaptonuria**.

Discussion:

An inborn error of amino acid metabolism.

AUTOSOMAL RECESSIVE inheritance.

Prevalance → 1 in 5,00000

Characterized by a triad of→

1..Homogentisic aciduria
2…Ochronosis
3….Arthritis

History: The disease was first identified in an Egyptian

Volume : 5 | Issue : 6 | June 2015 | ISSN - 2249-555X

mummy in 1500 B.C.It is one of the orphan diseases of the community.Also it is the earliest metabolic disorder to be identified.
Biochemical Defect:The metabolic defect is the deficiency of the enzyme homogentisic oxidase.This results in excretion of homogentisic acid in urine.

Course: The disease is compatible with fairly normal life. The only abnormality is the blackening of urine on standing.The homogentisic acid is oxidized by polyphenol oxidase to benzoquinone acetate.It is then polymerized to black colour alkaptone bodies.By the 3rd or 4th decade of life patient may develop ochronosis.Black pigments are deposited over the connective tissues including the joint cavities to produce arthritis.

Treatment: No specific treatment is required.But patient is advised to have minimum protein intake with phenylalanine less than 500mg/day.