

## Case report:

### Alkaptonuric arthropathy: a rare entity

Hemant Kumar†, Sagar Tyagi†, Parveen Hans†, Prof .Virinder Mohan\*

†- Residents, \*- Professor Emeritus

Department of Radiodiagnosis, Rohilkhand Medical college,

Bareilly, (U.P.) India

Corresponding author: Hemant Kumar

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#### Abstract

Alkaptonuria is a metabolic disorder of tyrosine and phenylalanine metabolism in which there is absence of homogentisic acid oxidase whose deficiency leads to excessive accumulation of homogentisic acid in urine resulting in black color of urine on standing.<sup>[1]</sup> The disease is usually diagnosed in adults but a few cases in childhood have also been noted because of black discoloration of their diapers. Homogentisic acid accumulates in cells and body fluids and its oxidized polymers binds to the collagen which leads to progressive deposition of grey to bluish black pigment results in degenerative changes in the cartilage, intervertebral discs and the connective tissues leading to arthritis.

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#### Introduction

Alkaptonuria is an inherited condition which causes urine to turn black when exposed to air for long hours. Ochronosis, build up of dark pigment in connective tissues such as skin and cartilage, is also a characteristic feature of the disorder. This blue-black pigmentation is usually seen after 30years of age. People with alkaptonuria develop arthritis, particularly in the spine and the large joints and starts to begin in early adulthood. Other features of this condition may include heart problems, kidney stones, and prostate stones.<sup>[2]</sup> This condition is very rare condition, affecting 1 in 250000 to 1000000 people worldwide.

#### Case report

A 35 years old male was seen in the medical OPD with history of back pain and stiffness of the back for more than 1 year. The backache was insidious in onset and was progressing day by day with history of morning stiffness which unables the patient to get up from the bed. There was no history of cough, fever. Clinical examination revealed a

young man in sound health, with no significant abnormal findings. Local examination revealed straightening of the dorso-lumbar spine. The spinal movements were restricted. No local tenderness was present. With clinical diagnosis of Low back pain, the patient was sent to Radiology department for X-ray of the lumbo-sacral spine, which revealed osteoporosis with normal vertebral height, reduced disc spaces and calcification of discs, anterior osteophytes and evidence of air (vacuum phenomenon) in multiple discs (Fig 1A & Fig 1B). The radiographic findings were typical of Ochronotic spine. X ray of cervical and dorsal spine were normal. Both knee joint X-ray did not show any calcification of the menisci. However X ray of both pinna (Fig 2) and nasal cartilage reveals calcification (Fig 3), further confirming the diagnosis of Alkaptonuria. Urine sample of the patient turned black when left outside for long hours. Other routine laboratory investigations were reported normal.

## Discussion

Alkaptonuria, a metabolic disorder characterized by homogentisic aciduria, arthritis and ochronosis is a very rare disorder and is one of the first condition in the group of inborn errors of metabolism to have Mendelian recessive inheritance. Since 1958, when the first case was reported, approximately 600 cases have been reported.<sup>[3]</sup> Its reported incidence is 1:250000 to 1:1000000 persons.<sup>[4]</sup> Few such cases have also been reported from India.<sup>[1][5][6]</sup> Ochronotic arthropathy, is a manifestation of long standing Alkaptonuria which occurs because of the accumulation of pigment deposits in the joints of axial and peripheral skeleton.<sup>[2]</sup> Clinically most often these cases are diagnosed as low back pain or early degenerative arthritis. The spinal movements may be painful or restricted. However neurological symptoms are present. Few of the patients may present with same condition to Dermatology Department for stiffness of Pinna and other Cartilage while occasionally the patient may volunteer the history of discolouration of his undergarments.

The X ray findings are almost confirmatory findings of the disease and consists of marked

osteopaenia of the vertebrae, marked reduction in the intervertebral disc height with calcification of the discs, early osteophytes formation and presence of air (vacuum phenomenon) in multiple discs, calcification of the pinna and nasal cartilage are also important findings. Osteoarthritis can be treated symptomatically as for other osteoarthritis, surgical intervention is seldom required. Treatment with ascorbic acid (VitaminC) and dietary restrictions of the food which contains phenylalanine and tyrosine have proved to be successful in alleviating the symptoms.

## Conclusion

Alkaptonuria, ochronosis, and arthritis, represents a clinical entity which has its genesis in a rare metabolic anomaly. Extensive calcification of the intervertebral discs with ankylosis of the spine are typical radiographic findings. Similar calcifications also occur in tendon sheaths, bursal sacs, and in synovial membranes.

A case of this disease is recorded, in which the diagnosis of alkaptonuria was made from the radiographs and confirmed by urine examination, which turned black after it was left in open for long hours.

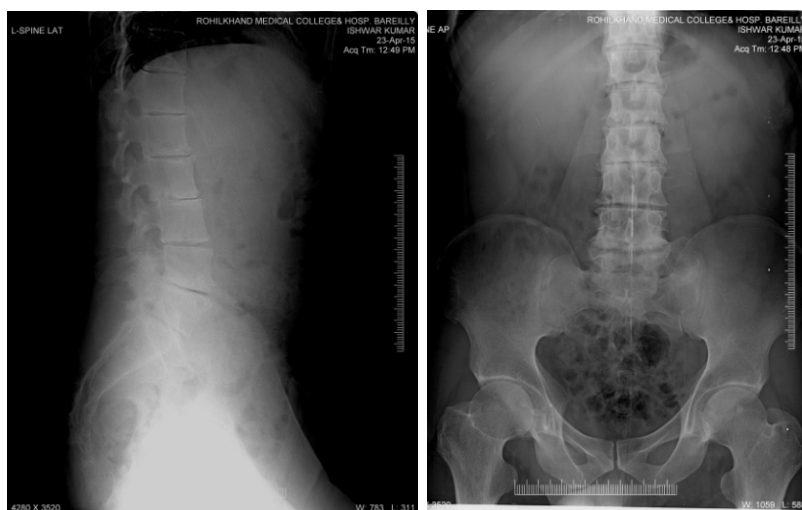


Fig1a & 1b shows osteoporosis with normal vertebral height, reduced disc spaces and calcification of discs, anterior osteophytes and evidence of air (vacuum phenomenon) in multiple discs



Fig 2: X-Ray of both Pinna cartilage reveals calcification



Fig 3: X-Ray of nasal cartilage reveals calcification

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