

CASE STUDIES

Alkaptonuria, a rare cause for low back pain

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Abstract

Alkaptonuria is a very rare disease that can cause premature degenerative arthritis of the spine and peripheral large joints. I present here a case of middle-aged woman who developed low back pain secondary to alkaptonuria.

Case report

A 45-year-old female presented with a history of low back pain since 10 years. She had difficulty in bending, restriction in back movements, and scoliosis. She was also experiencing early morning stiffness, which improved with activity, and pain on both the knee joints without swelling. The patient had a family history of similar complaints and the affected family members also demonstrated back pain. She had also reported the occurrence of urine turning black after some time (Fig 1). Physical examination demonstrated the blackish discoloration of ears as well as the pigmentation of sclera (Fig 2). Examination of the spine showed kyphoscoliosis with normal spinal mobility. Anteroposterior X-ray of the spine showed intervertebral disc calcification with normal sacroiliac joint (Fig 3).

Fig 1: Change in urine color when exposed to air



Discussion

Alkaptonuria, a rare defect with autosomal recessive inheritance, is caused by the abnormal tyrosine catabolism due to the deficiency in homogentisic acid oxidase. The enzyme catalyzes the catabolism of homogentisic acid to molecules that can be used in Krebs cycle. Deposition of homogentisic acid produces gray to blue pigment in the tissues. It can cause pigment deposition in the connective tissue of eye, ear, cartilage, and cardiac valves. Generally, the disease will be asymptomatic, but may appear in the middle age due to early degenerative arthritis involving lower spine, and shoulder and hip joints requiring early joint replacement. The current incidence of the disease is estimated to be 1 in 250,000 -1,000,000 live births.¹

Alkaptonuria should be suspected in patients with the following clinical triad:

1. Degenerative arthritis (premature) i.e. in young patients of <45 yrs.
2. Abnormal pigmentation of connective tissues in eye, ear, and cardiac valves.
3. Urine turning blue-black on standing (due to alkalinization).

Since homogentisic acid is absent in the blood plasma and urine of healthy subjects, both of them can be used for the disease diagnosis. In alkaptonuria patients, the average levels of homogentisic acid noted in the plasma and urine are 6.6 micrograms/ml and 3.12 mmol/mmol of creatinine respectively.¹ Confirmatory tests for diagnosis are paper chromatography and thin layer chromatography.

Fig 2: Pigmentation of the ear and sclera

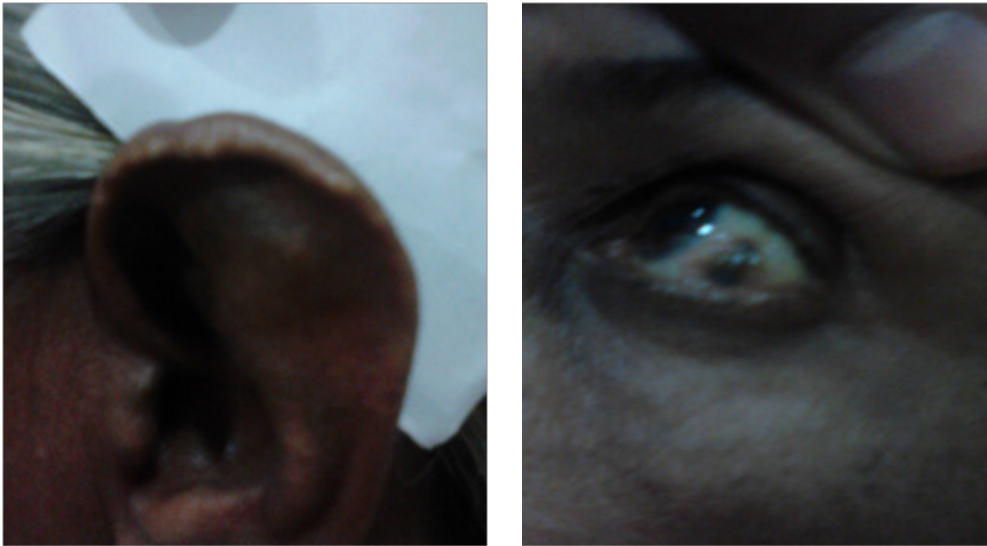
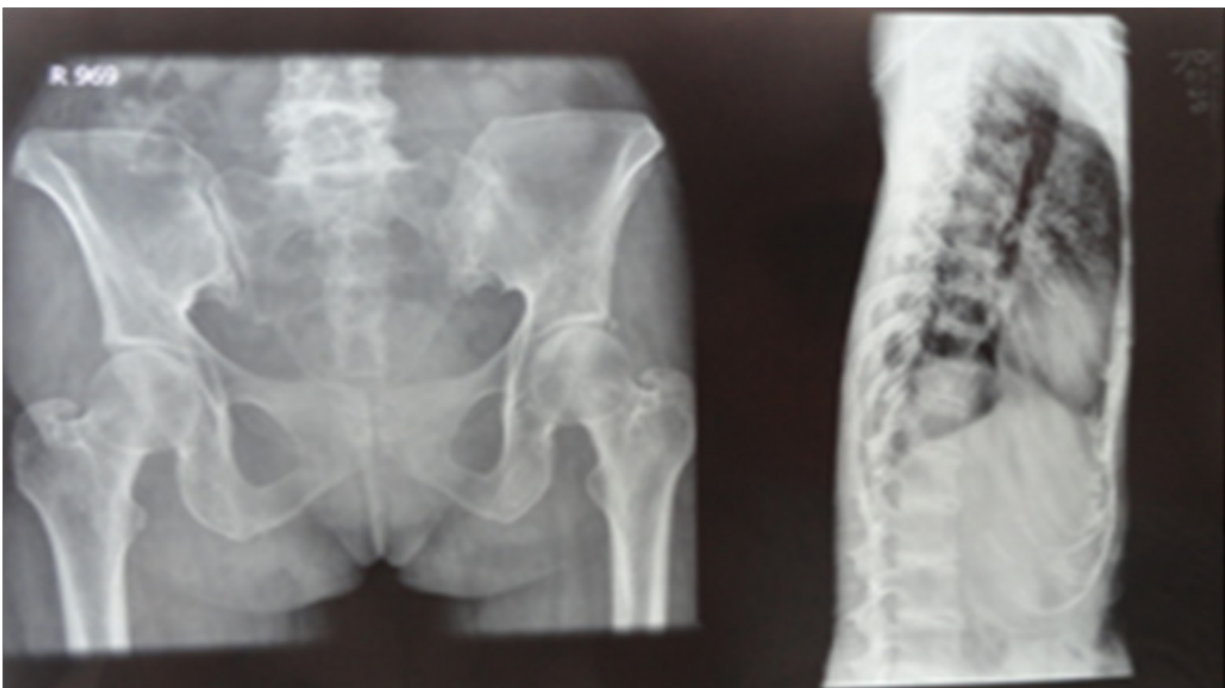


Fig 3: X-ray showing pelvis with normal sacroiliac joint and lateral thoracolumbar spine with intervertebral disc calcification



Characteristic radiological findings include vertebral disc calcification, chondrocalcinosis, and osteoarthritis of multiple joints (especially spine) at young age.

Since most of the young patients with alkaptonuria present with low back pain, it can mimic ankylosing spondylitis. Generally, the disease could be suspected in young patients with low back pain and X-ray showing intervertebral disc calcification with normal sacroiliac joint. They can also

have premature osteoarthritis of knee or other major joints. A detailed history and examination may reveal black staining of the undergarment, change in the urine color on longstanding, pigmentation of sclera, and grayish hue discoloration of the ears. First-degree relatives may have symptomatic scleral pigmentation and ear discoloration before they develop any joint manifestation (as many are unaware of the urine color change or staining of clothes).

No treatment modality has been unequivocally proven to reduce the complications of alkaptonuria. Commonly advocated treatment strategies include large doses of ascorbic acid (vitamin C) and dietary restriction of tyrosine and phenylalanine. Although, dietary restriction may be effective in children, it has not shown any benefits in adults.¹

Competing interests

The author declares that he has no competing interests.

Citation

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