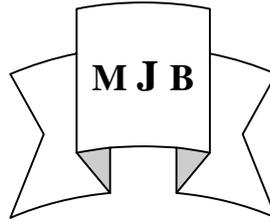


## Ochronotic Spondylitis

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### Case Report



A 47-year-old man presented in August, 2004 with a three years duration, of insidious Low back pain and stiffness.

His back stiffness was more severe and pronounced at waking-up in the morning, the pain and stiffness eased with activity as the day is going on.

The pain was dull in nature, radiated to his buttocks, and poorly localized.

He had no recent illnesses. His past medical history included a lithotomy from Urinary bladder, 5 years ago.

Also he noticed progressive darkening of the skin over his face for the last several years.

On the examination, the patient had dark discolorations of the face mainly over cheeks, nose and triangular dark pigmentation of both sclera.

The normal cervical and lumbar lordosis were obliterated, with restriction of movement of the entire

spine in all planes, while chest expansion was preserved, No local spinal or paraspinal tenderness, no evidence of enthesitis elsewhere in the body. No neurological deficit could be detected.

Routine blood investigation (complete blood picture) was within normal range. General urine examination revealed proteinuria, pyuria. On keeping the urine in test tube, the color progressively darkened, few hours later.

Plain chest x-ray showed no abnormal findings, Echo examination revealed no cardiac or valvular lesions.

Conventional plain x-ray of spine revealed, severe osteoarthritic changes, with severe narrowing of disks, with normal sacro iliac joints. Plain x-ray and ultrasound study revealed presence of stag horn stone in left kidney.

Alkaptonuria "Ochronosis" is a rare autosomal recessive disorder , result from a complete deficiency of the enzyme homogentisic acid oxidase. This defect cause accumulation of homogentisic acid , a normal intermediate in the metabolism of phenylalanine and tyrosine .

Alkalization and oxidation of this acid , which usually is excreted in the urine , cause the urine to turn black .

The homogentisic retained in the body is deposited as a pigment polymer in the cartilage and to the lesser degree , in the skin and sclerae , the darkening of tissue by this pigment is designated Ochronosis.

Patient with ochronosis , clinically presented with progressive degenerative arthropathy , with chondrocalcinosis , initially affecting the spine , later the peripheral joints the Knee , shoulder and hip .the small peripheral joint usually are spared.

The first sign of " ochronotic spondylitis " may be an acute similar to disc prolapsed , later on , it clinically resembles ankylosing spondylitis , with progressive rigidity and fusion . But in contrast to Ankylosing spondylitis , the

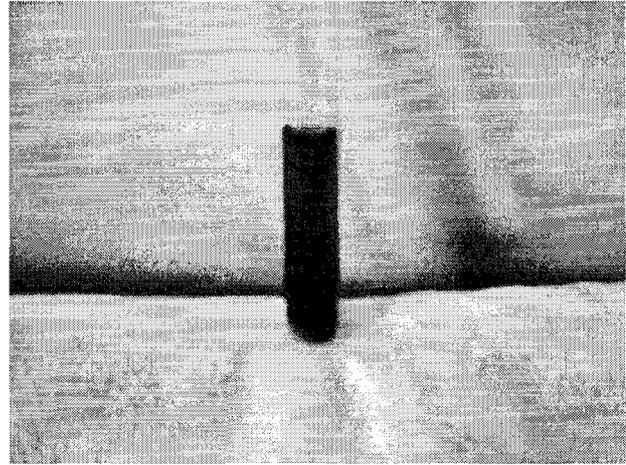
sacroiliac and apophyseal joints are not affected . Extra articular features include bluish discoloration and calcification of the ear pinnae , triangular pigmentation of the sclera and pigmentation over the nose , axillae , groin . Renal calculi and renal impairment may progress to renal failure "Ochronotic nephrosis " . Prostatic calculi are common in men . Cardiac murmur may develop from valvular pigment deposit.

#### **References:**

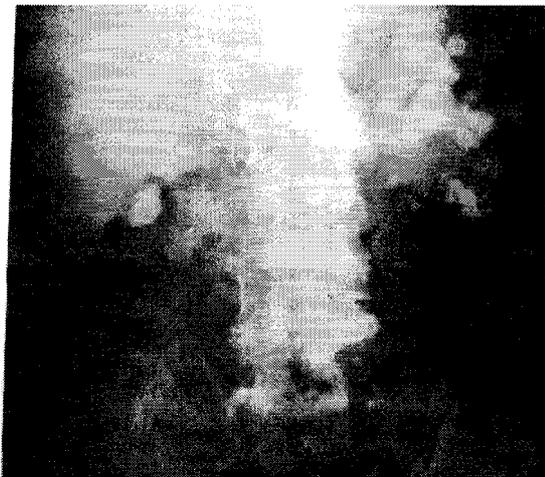
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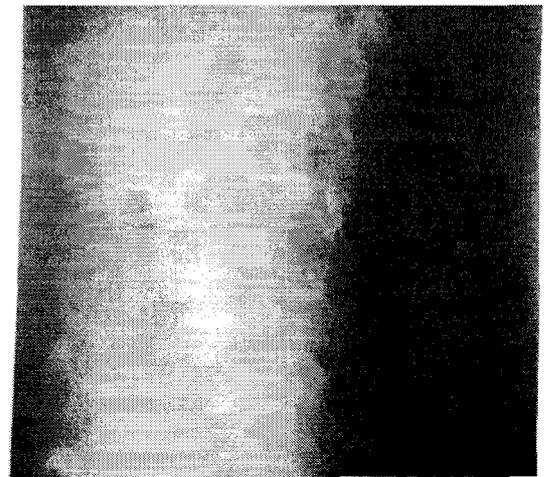
**Figure 1** shows the dark - bluish discoloration over the nose and ear pinnae of the patient



**Figure 2** shows the urine of the patient which turn black several hours later.



**A**



**B**

**Figure 3 -A&B** plain x- ray of lumbo sacral spine A-P & lateral which show marked degenerative changes and chondrocalcinosis of inter vertebral disks with sparing of sacroiliac joints . stag horn stone is seen in the left kidney with another stone is seen in the right kidney