OCHRONOSIS- A CASE REPORT

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PRESENTATION OF THE CASE

A 40-year-old patient presented to us with chief complaints of low back pain & large joint polyarthralgia for the last 8 years. Pain was more on bending forward, prolonged standing/sitting. There was also morning stiffness in the back. Pain was mild to moderate in intensity. There was no radiation of pain. Pain used to get relieved with rest & with NSAIDs. Patient also complained of black discolouration of urine when he urinated in the open. There was no other complaint.

On examination he had bluish discolouration of pinna of right ear (Figure 1). On ocular examination, bluish black discolouration was noted between the corneal margin and the inner canthus. Right knee was held in 15 degree of flexion. There was also retrocalcaneal swelling & tenderness. Lumbar lordosis was obliterated and tenderness was present at dorsolumbar spine with paravertebral spasm. SLR was 80 degrees on both sides. No neuro-deficit was present. X-ray Dorsolumbar spine showed calcification of intervertebral disc with degeneration present at multiple levels (Figure 2). X-ray knee showed reduced joint space which was uniform in both knees. There was diffuse osteoporosis but osteophytes were relatively absent. There was extensive subchondral sclerosis and intraarticular loose bodies were also present.

Echo heart was normal and there was no evidence of calcification of valves.

The haematological & biochemical markers were all normal. Routine urine study was undertaken, homogentisic acid (HGA) levels were increased. On exposure to light, the urine turned black (Figure 3).

CLINICAL DIAGNOSIS

Ochronosis

DIFFERENTIAL DIAGNOSIS

- Ankylosing spondylitis
- Argyria
- Arsenic poisoning
- CPPD
- Chondrocalcinosis
- Minocycline toxicity
- Pigmented villonodular synovitis
- Melasma
- Rheumatoid arthritis

PATHOLOGICAL DISCUSSION

Ochronosis is the bluish black discoloration of certain tissues, such as the ear cartilage and the ocular tissue. It is of two types. Exogenous ochronosis occur from exposure to various substances such as phenol, trinitrophenol, resorcinol, mercury, picric acid, benzene, hydroquinone, and antimalarials. Endogenous ochronosis or Alkaptonuria is a rare autosomal recessive disease with a prevalence of 1 case per 1 million population. The highest prevalence of 1 case per 19,000 inhabitants is seen in Slovakia and Dominican Republic.

Ochronosis a autosomal recessive disorder was first discovered by Rudolf Virchow, involves a defect in the metabolism of phenylalanine and tyrosine to fumarate and acetoacetate. On microscopic examination, there is yellowish (ochre-like) discolouration of the tissues. Ochronosis name is derived from this ochre-like discolouration. However, macroscopically the affected tissues appear bluish grey because of Tyndall effect.[1] There is loss of function mutation mapped to chromosome 3q2, which results in the deficiency of the enzyme homogentisate 1,2-dioxygenase leading to abnormal accumulation of homogentisic acid in the body and increased urinary excretion which on standing turns black due to oxidation.[2,3] This can begin in infancy also however other systemic complications are often evident by the third to fourth decade of life, due to selective ochronotic pigment deposition in connective tissues including hyaline cartilage, tendons, ligaments and muscles typically affecting the malar areas, temples, lower cheeks, and neck. Papular and nodular lesions may also be seen.[4] Pigment deposition is also seen in hyaline cartilage in the respiratory tract, endocardium of the heart, heart valves (base and anuli of the aortic and mitral valves), and walls of arteries. Darkening and hardening of ear cartilage is a prominent feature of ochronosis as is seen in our case. Stiffening of the ribs with decreased lung function has also been reported. The intervertebral cartilage is also more prone to herniation.[5]

Back pain is often described as a dull, aching pain with stiffness. Sudden, severe sharp pain can also be the initial symptom if the nucleus pulposus ruptures. Features of advanced diseases include thoracic kyphosis, loss of lumbar lordosis, decreased spinal mobility and loss of overall height can occur.[6] Cervical spine and pubic symphysis joints can also be affected. Sacroiliac and apophyseal joints are not affected. Schober's test signifying loss of lumbar mobility is

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often positive. Significant morning stiffness is not a component of the presentation. Chest expansion and respiratory function can become impaired. Osteoporosis and osteopenia is commonly found in this population, as well as fractures. Our patient also had back pain which was gradual in onset.

Ochronotic arthropathy is a manifestation of long-standing alkaptonuria due to chronic inflammation and micro ruptures. Peripheral arthritis often follows the back pain, and primarily include large joints, often sparing the small joints. Initial arthritic symptoms include pain, stiffness, and limited range of motion, favouring a flexed position.[7] Symptoms may be acute, with joint effusions and synovitis or insidious, with pain on weight bearing. Physical examination findings includes joint tenderness, crepitus, synovitis and loss of range of motion.

Farooq A. Rathore et al (2016)[8] also presented two cases of alkaptonuria resulting in ochronotic arthropathy with advanced secondary generalized osteoarthritis, intervertebral disk calcifications, skin and scleral pigmentation. In these case reports, both patients had symptoms for >10 years before being diagnosed.

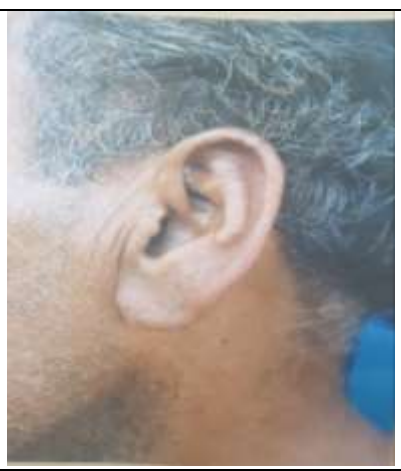

Figure 1. Brownish Black Pigmentation of The Ear Pinna

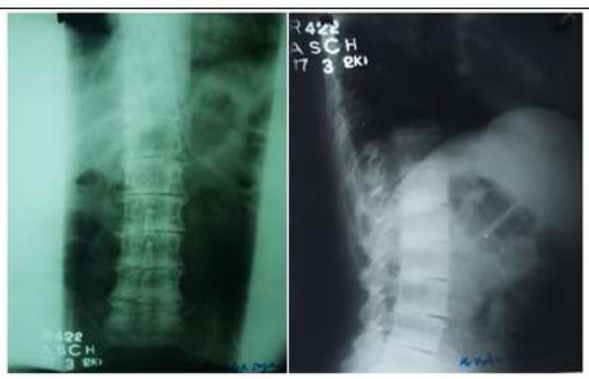

Figure 2. X-Ray Dorsolumbar Spine Showing Calcification of Intervertebral Discs

Figure 3. Urine Sample Before and After Exposure to Sunlight

DISCUSSION OF MANAGEMENT

Treatment done was predominantly preventive. Patient was advised to avoid use of topical phenols and to restrict consumption of milk, meat, fish, beans, cheese, nut etc., which have high amounts of tyrosine. Patient was also advised to take tablet Vitamin C daily.

Arthroscopy was advised to see the extent of joint damage. He was advised to stop using hydroquinone-containing compounds like bleach.

FINAL DIAGNOSIS

Ochronosis.

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