Alkaptonuric Arthritis – A Case Report with Review of Literature

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Abstract

A case of alkaptonuria is reported for its rarity with extensive involvement of spine, sacroiliac joints and symphysis pubis.

Key Words

Alkaptonuria, Scoliosis, Calcification.

Introduction

Alkaptonuria is a rare disorder of metabolism and very few cases of this condition have been reported in India (1-4). A case of alkaptonuria is reported for its rarity and its extensive involvement of spine, sacroiliac joints and symphysis pubis.

Case Report

A 55 year old male patient reported with discomfort in the low back. He was little affected by his complaints until few years back when he noticed his back stiffer than normal and occasionally painful. He had never severe pain in the back. The patient was moderately built for his age with slight kyphosis in the thoracic spine with obliteration of lumber lordosis and slight lumber scoliosis. Straight leg raising test was positive at about 60°. There was complete immobility of thoracic and lumber spine. The pinnae of both ears felt stiffer and thicker than normal and there was blackish brown pigmentation of both ears, sclera of both eyes, skin of peri-orbital regions, dorsum of hand. The patient walked with forward bend(Fig. 1.2.3).

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Laboratory investigations revealed routine urine examination to be normal but it was positive for homogentisic acid. Urine turned black when exposed to air and addition of sodium hydroxide accelerated the process. Addition of Benedict's reagent to a urine sample gave rise to greenish brown precipitate and brownish black supernatant. Haemoglobin, ESR, blood urea, serum uric acid, serum calcium, serum phosphorus and serum alkaline phosphatase were within normal limits. HLA-B27 was negative.

X-ray of the lumbosacral spine showed scoliosis towards the right, calcification in the disc spaces with bony ankylosis of the lumbar spine. The pelvis showed irregularity and distinct sclerosis of articular surface of sacroiliac joints and partial fusion. The symphysis pubis was narrow (Fig. 4). The dorsal spine also showed diffuse calcification with secondary osteoarthritic changes. There was diffuse rarefaction of all vertebral bodies (Fig. 5). In the knee joints the joint space was narrowed with marginal osteophytes (Fig. 6). As there is no specific treatment for this condition, the patient was put on ascorbic acid, analgesics and was advised to cut down food rich in tyrosine like cow and buffalo milk and corn.
Discussion

Alkaptonuric ochronosis is an autosomal recessive metabolic disorder that affects approximately one in one million individuals. It is a rare disorder of tyrosine metabolism, in which there is deficiency of homogentisic acid oxidase enzyme which results in increased excretion of homogentisic acid and its polymers in soft tissues. The disorder may remain unnoticed till middle age. The usual age of onset of arthritic symptoms is 40 years. The most common clinical features are homogentisic aciduria, pigmentation of cartilage and other connective tissues, ochronotic arthritis.

Different authors have reported the involvement of different systems in body. Cardiovascular involvement with affection of heart valves demonstrated on echocardiography was reported by Kocyigit et. al. (5) Palmoplantar pigmentation associated with blunt pigmented patches over the sclera of the eyes have been reported by Vijaikumar et al. (4). Stiehl and Klunger, reported synovial effusion in a 55 year old man with numerous microscopic visible bluish black rods of a length of 3 to 5 mm (6). Mulay et al, found jet black discoloration of femoral head and other soft tissues during surgical intervention for fracture neck of femur in a 74 year old male (2). Gulati as well as Shetty et. al. reported alkaptonuric spondylitis associated with scoliosis, bladder stone and calcification of pinnae (1,3).

In the later stages of disease, the spine gets fixed in kyphosis and scoliosis and rarely bony ankylosis may occur which is indistinguishable from ankylosing spondylitis. The distinguishing features are that in ochronosis the primary changes are in discs rather than ligaments and interarticular joints. The anterior longitudinal ligament is conspicuously not affected and also sacroiliac joint does not become fused (7). But in the present case the bony ankylosis of lumber spine in kyphoscoliosis with gross involvement of both sacroiliac joints and their partial ankylosis indicates that sacroiliac joints are not immune to this metabolic disorder. Also in the symphysis pubis, the inter-pubic space is very narrow indicating that ochronosis acts as a predisposing factor for osteitis pubis, perhaps by reducing the vitality of the interpubic cartilage which may also progress to ankylosis in due course of time.

References