

Alkaptonuria Presenting as Spondyloarthropathy in an Elderly

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Abstract

A case of alkaptonuria presenting in an elderly is reported. Patient presented with cutaneous and scleral pigmentation, low backache and knee pain along with passage of dark urine since birth. Diagnosis was suspected on clinical grounds, supported by X-ray and confirmed by detection of homogentisic acid in the urine.

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Introduction

Alkaptonuria is a rare inborn error of metabolism of amino acid tyrosine. It is caused by deficiency of the enzyme homogentisic acid oxidase leading to accumulation of homogentisic acid, part of which is excreted in urine. Since homogentisic acid has a high affinity to alkalies, it was named as alkapton and this condition as alkaptonuria. The rest of it gets polymerized and is deposited as a pigment in the connective tissue of the body, mainly of the joints, skin, cartilages, outer eye structures, cardiovascular system. The pigment looks ochre (yellow) microscopically and hence called ochronosis. Hereby, we are presenting a case of alkaptonuria in an 80 years old male patient.

Case Report

A 80 years old male patient presented with low backache for 25 years. It was dull aching and continuous without radiation to buttocks or lower limbs. There were no aggravating or relieving factors. There was progressive forward bending of the trunk for last ten years. For 10 years, he had moderately severe pain both knee joints. It used to aggravate on movement and accompanied with difficulty in straightening of the joints.

There was history of change of colour of urine to black on standing since childhood. There was progressive bluish discoloration of the face for fifty years. Initially ear lobules became bluish followed by

periorbital area in next 5-6 years. He also noticed similar discoloration over palm, sclerae and planter aspect of feet. There was no history of photosensitivity, recurrent oro-genital ulceration or small joints involvement, blood in stools, urethral discharge or red painful eyes.

Patient was a chronic bidi smoker (40 pack years), nonalcoholic and vegetarian. He was nondiabetic, with no history of tuberculosis or other chronic illnesses. His younger brother also has similar problem of discoloration of urine since childhood. None of their children or other siblings were suffering from similar illness.

Examination

Bluish discoloration was noted over ear pinna, nose, periorbital area, medial aspect of palm and planter aspect of feet. Greyish discoloration was present in sclerae (Fig 1 & 2). Dorsal kypho-scoliosis was noted with convexity towards right side. Fixed flexion deformity of both knee joints of around 30° was also noted. Schober test revealed 3 cm increase in distance on flexion. Periarticular muscle wasting was present around both knee joints. Rest of general and systemic examination was unremarkable.

Investigations

Hemoglobin – 14 gm%, total leukocyte count – 8420 /cu mm, differential leucocyte count- N₆₀ L₄₀ %, fasting blood sugar – 79 mg%, serum creatinine - 0.5 mg%, blood urea - 25mg%, urine culture –sterile. X-ray dorso-lumbar spine showed narrowing of multiple disc space with intervertebral disc calcification (Fig 3) suggestive of ochronotic spondyloarthropathy. X-ray both knee joints showed reduced joint space,

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Figure 1



Figure 2

Fig. 1 & 2 shows discolouration of pinna and sclerae.

subchondral sclerosis and, osteophytes. Urine was positive for homogentisic acid.

Discussion

Alkaptonuric patients are usually asymptomatic in young age. In fourth decade, pigmentation of the sclera or the cartilage of the ear may start to appear.¹ Pigmentation may be seen in the teeth, buccal mucosa, nails or skin, giving these areas the dusky colouration.² The wide spread pigmentation in the alkaptonuric patients is called ochronosis. Arthritis is the only disabling effect of this condition and occurs in almost all patients as age advances.³ The earliest symptoms are usually in the hips, spine and knees, the large weight bearing joints. Arthritis has the clinical characteristics of rheumatoid arthritis. However, the radiological picture is that of severe osteoarthritis.⁴ In contrast to osteoarthritis, the large joints at the hip and shoulder are most commonly affected, whereas the sacroiliac joints may be spared. The degenerative changes in the hip and spine are characteristic with narrowing of joint spaces and fusion of the vertebral bodies. Ochronotic arthropathy in the hip and knees may be so severe so as to require total joint arthroplasty. There is a high incidence of heart disease due to mitral and aortic valvulitis. Secondary calcification of the aortic valve may be so severe as to necessitate urgent aortic valve replacement.⁵

In our case the typical features of alkaptonuria were present in the form of skin and scleral



Fig. 3: Radiograph of lumbar spine showing narrowing of disc spaces and calcification of intervertebral discs.

discolouration. Joint pain and stiffness was the dominant presentation. Joints involved were mainly knee and spine as documented by O'Brian (1961).⁴ There was no evidence of cardiac illness either in history or on physical examination. Thus considering discoloration of skin, sclerae, with arthropathy and X-ray findings along with presence of homogentisic acid in the urine, the diagnosis of alkaptonuria was entertained. The atypical feature was its presentation at such a late age, which underlines the poor health awareness in Indian public.

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