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A RARE CASE OF ALKAPTONURIC ARTHRITIS

Orthopedics

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ABSTRACT

Ochronosis is an inherited genetic disorder of phenylalanine and tyrosine metabolism due to deficiency of enzyme homogentisic acid oxidase.It is an inborn error of metabolism with autosomal recessive inheritance.Greyish to bluish black pigment in the connective tissues such as cartilage,skin and intervertebral disc occurs due to accumulation of homogentisic acid leading to degenerative arthritis. This condition is rare, affecting 1 in 250000 to 1 million people worldwide.Diagnosis can be made in neonates when blackish stain is noticed in an diaper. Alkaptonuria is treated symptomatically, with Vitamin C and dietary restrictions of food containing phenylalanine and tyrosine have proved to be successful in reducing the symptoms.

KEYWORDS

Ochronosis, Alkaptonuric Arthritis, Alkaptonuria

INTRODUCTION

Ochronosis is also called as Alkaptonuria (AKU) arises from the Arabic word alkapton for "alkali" and Greek word "to suck up oxygen greedily in alkali" based on the observation that the urine becomes black on standing when it becomes alkaline[1]. This disorder is of historical interest in that it was one of the original "inborn errors of metabolism" described. In 1900's, Archibald Garod, a pediatrician, recognized that the syndrome, which has been called AKU, showed a pattern of familial inheritance[2]. It was proposed that AKU and certain other disorders were due to genetic defects, each of which resulted in the lack of activity of a particular metabolic enzyme[3]. Forty five years later, it was confirmed that the biochemical defect in AKU is a lack of homogentisate oxidase in the tyrosine catabolic pathway .Mutations in the HGD gene makes the body unable to properly breakdown tyrosine and phenyl alanine aminoacids leading to accumulation of homogentisic acid[8].This disorder occurs in about 1 in 2,50,000 births[4]. Due to its rarity in the prevalence rate, we report this case of Alkaptonuric arthritis which was diagnosed in Dr.psims and rf.

CASE REPORT

A 40year old lady presented with chief complaints of pain in the low back region for 3 years and pain in both knees since 2 years.

Pain is insidious in onset,progressive in nature,non radiating associated with tingling sensation of the limbs occasionally.

No history of fever,loss of weight or appetite.

Family history-similar history noted in younger brother

General examination of the patient revealed pigmentation over the both sclera (fig 1a),concha(fig 1b) and nose

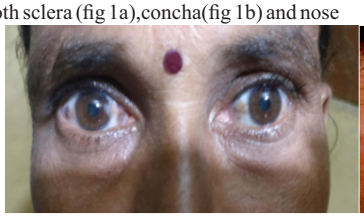

Fig 1a-sclera pigmentation

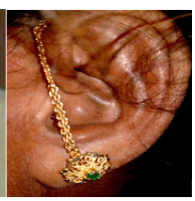

fig 1b- pigmentation in concha

Local examination of spine reveals no kyphotic or scoliotic deformity. tenderness present at the lower lumbar spine region.jointline tenderness present over both the knee joints.

Radiographic findings: a routine lumbar spine radiograph revealed calcification of the intervertebral disc(wafery calcification) with bridging osteophytes and osteoarthritis of both knee joints (fig 2).

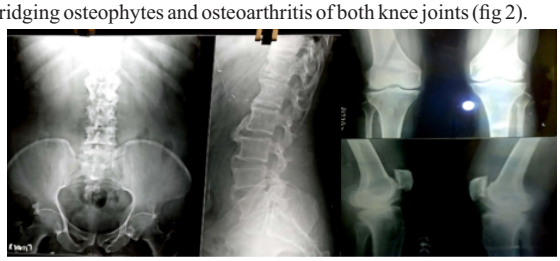

Fig 2-calcification of intervertebral disc with osteoarthritis of knee

Laboratory investigations-

Hb-9.5 g%,ESR-20 mm/hr,serum creatinine-0.9mg% The routine laboratory investigations were normal. In view of clinical examination and radiographic findings alkaptonuric arthritis was suspected and to confirm the diagnosis 24 hour urine sample exposed to air was turned to cola colored.(fig 3)

fig-3-24 hr urine sample

Treatment-

Patient was treated symptomatically with mild analgesics, physiotherapy and vitamin C 500mg daily. Foods high in tyrosine and phenyl alanine such as eggs,dairy products and red meat were adviced to be avoided. Currently the patient is asymptomatic and is under follow up every six months in the outpatient clinic of the hospital.

Conclusion:

AKU a metabolic disorder that is not life threatening.Disability and pain forms the major complaint of these patients. General examination, local examination and radiological examination followed by biochemical tests help in confirming the diagnosis.Administration of Vitamin C will reduce excretion of homogentisic acid but has no effect on the progress of the disease.

REFERENCES

1. Garrod AE. The incidence of alkaptonuria: a study in chemical individuality. Lancet 1902; 11:1616-20.
2. Jaffe HL. Gaucher's disease and certain other inflammatory metabolic disorders. Chapter 18 in metabolic, Degenerative and inflammatory diseases of bones and joints. Philadelphia, Lea and Febiger, 1972; 552-60.
3. Di-Franco M, Caori G, Bonucci E. A morphological study of bone and articular cartilage in ochronosis. Virchows Arch 2000; 436 (1): 74-81.
4. Osler W. Ochronosis: the pigmentation of cartilages, sclerotics and skin in alkaptonuria. Lancet 1904; 1: 10.
5. Yules JH. Ochronotic arthritis: report of a case. Bull N Engl Med Center 1957; 16: 168.
6. O'Brien WM, Banfield WG, Sokoloff L. Studies on the pathogenesis of ochronotic arthropathy. Arthritis Rheum 1961; 4: 137.
7. WolffJA,BarshopB,NyhanWL.Effectsofascorbicacidin alkaptonuria: alterations in benzoquinone acetic acid and an ontogenic effect in infancy. Pediatr Res 1989; 26: 140-4.
8. Hogben L, Worrall RI, Zieve. The genetic basis of alkaptonuria. Proc R Soc Edinb (Biol) 1932; 52: 264.