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Case Report

Alkaptonuric Ochronosis - A Case Report

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ABSTRACT

We describe a 60 year old woman who presented with backache and pigmentary changes involving her palms and sclera of 2 years duration. Patient's urine turned black on standing and also after alkalization with Benedict's reagent. Radiographs of her dorso-lumbar spine showed typical intervertebral disc calcification, narrowing of disc space and vertebral osteoporosis suggestive of Alkaptonuria. We diagnosed the patient to have Alkaptonuric Ochronosis.

Key Words: Alkaptonuria, Ochronosis.

INTRODUCTION

Ochronosis is a syndrome caused by the accumulation of homogentisic acid in the connective tissues. It was first described by Rudolf Virchow in 1865. [1] The condition was named after the yellowish discoloration of the tissue seen on microscopic examination. However, macroscopically the affected tissues appear bluish grey because of alight scattering phenomenon known as the Tyndall effect.

CASE REPORT

A 60 year old female presented to the Orthopaedics department of our hospital with backache of 2 years duration. She was referred to the DVL OPD for pigmentary changes in the palms. She gave history of pigmentation of her palms and eyes of 2 years duration. There was history of blackish staining of the undergarments. There was no history of use of any topical or systemic medication that could cause the pigmentation. She was born to non-consanguineous parents. Her past medical history was unremarkable.

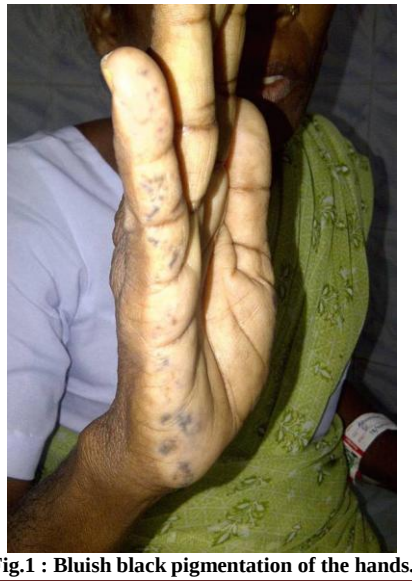

Fig.1 : Bluish black pigmentation of the hands.

Physical examination revealed bluish black pigmentation in a bilaterally symmetrical pattern involving the thenar and hypothenar eminences, ulnar border of the hands and little fingers and radial border of the thumbs (Fig.1). Associated thickening and pitting was seen. Ophthalmic examination revealed bluish black pigmentation of the sclerae. She had diffuse tenderness over the dorso-lumbar spine and paraspinal region. Mobility of the dorso-lumbar spine was partially restricted.

Routine haematological and biochemical profile was within normal limits. Radiographs of the dorso-lumbar spine showed intervertebral disc calcification, narrowing of disc space, diffuse vertebral osteoporosis and osteophytes. On exposure to air her urine turned black. Alkalization of urine with Benedict's reagent also turned it black (Fig.2).

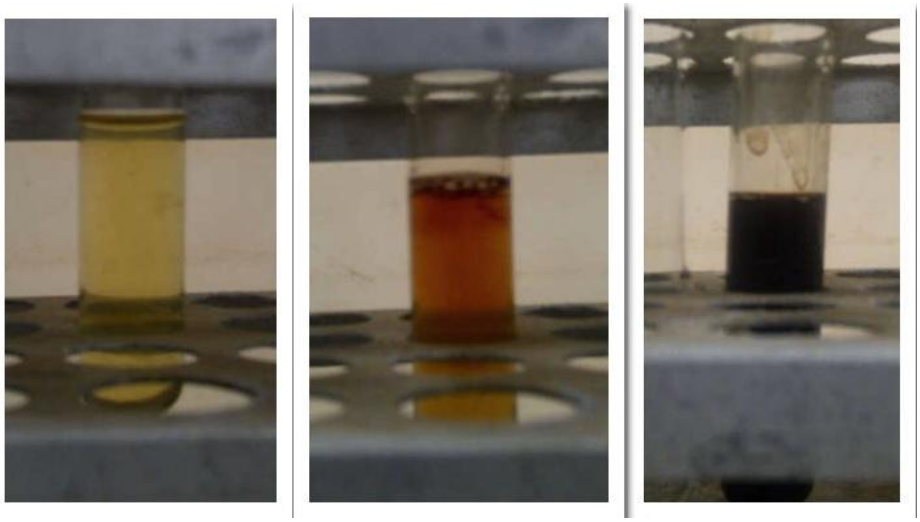

Fig. 2: Urine turning black on alkalization with Benedict's Reagent.

DISCUSSION

Ochronosis is a condition caused by the accumulation of homogentisic acid in connective tissue. It was first described by Rudolf Virchow in 1865. [1] The condition was named after the yellowish (ocher like) discoloration of tissues seen under the microscope. [2] The affected tissues however appear bluish grey macroscopically due to Tyndall effect. Ochronosis is most often associated with Alkaptonuria but can also occur from exogenous administration of phenol complexes like hydroquinone. Albrecht (1902) was the first to suggest Alkaptonuria as the cause for Ochronosis.

Alkaptonuria is a rare genetic disorder of phenylalanine and tyrosine metabolism inherited as an autosomal recessive condition. Its incidence is about 1 in 10,00,000 persons. [3] It is due to deficiency of the enzyme homogentisic acid oxidase leading to accumulation of homogentisic acid in various tissues of the body. Early manifestations like dark urine (at birth), axillary pigmentation (at puberty) and ear lobe pigmentation (20-40 years) may go unnoticed. The cases are most easily recognized in the fourth or fifth decade with a peak incidence in the fifth decade. Our patient became symptomatic in the sixth decade by when she presented with palmar and sclera pigmentation and intervertebral disc calcification.

Treatment is directed towards reducing connective tissue damage by high doses of ascorbic acid along with analgesics and physiotherapy. [4] A low protein diet limiting phenylalanine and tyrosine is recommended. [5] It may be beneficial in children but its benefit in adults has not been demonstrated. [6] Clinical trials are on to study the efficacy of Nitisinone in the treatment of Alkaptonuria. [7]

REFERENCES

1. Findlay GH, et al. Ochronosis. Clinics in Dermatology 1989;7:28-35.
2. O'Brien W.N., La Du B.N., Bunim, J.J. Biochemical, pathologic and clinical aspects of alkaptonuria, ochronosis and ochronotic arthropathy. Am. J. Med.1963; 34: 813-838.
3. Janocha S, Wolz W, Srsen S, Srsnova K, Montagutelli X, Guénet JL, Grimm T, Kress W, Müller CR. The human gene for alkaptonuria (AKU) maps to chromosome 3q.Genomics. 1994 Jan 1;19(1):5-8.
4. Morava E, Kosztolanyi G, Engelke UF, et al; Reversal of clinical symptoms and radiographic abnormalities with protein restriction and ascorbic acid in alkaptonuria. Ann Clin Biochem. 2003 Jan;40(Pt 1):108-11.
5. Mayatepek E, Kallas K, Anninos A, Müller E. Effects of ascorbic acid and low-protein diet in alkaptonuria.Eur J Pediatr. 1998 Oct;157(10):867-8.
6. de Haas V, Carbasius Weber EC, de Klerk JB, et al; The success of dietary protein restriction in alkaptonuria patients is age-dependent. J Inherit Metab Dis. 1998 Dec;21(8):791-8.
7. Suwannarat P, O'Brien K, Perry MB, et al; Use of nitisinone in patients with alkaptonuria. Metabolism. 2005 Jun;54 (6):719-28.

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