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CUTANEOUS CRUX OF A METABOLIC DEFECT. – CASE REROPT ON OCHRONOSIS.

Pathology

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KEYWORDS:

INTRODUCTION:

Alkaptonuria is an autosomal recessively inherited inborn metabolic disorder in which there is lack of homogentisic oxidase, an enzyme that converts homogentisic acid to methylacetoacetic acid in the tyrosine degradation pathway. As a result, homogentisic acid accumulates in the body and binds to collagen in connective tissues, tendons and cartilage, imparting to these tissues a blue black pigmentation (Ochronosis) most evident in the ears, nose and cheeks.

CASE HISTORY:

A 50 year old female presented with complaints of joint pain and bluish grey pigmentation with thickening of skin of both palmar eminence(Fig.1), web spaces(Fig.2), tips of fingers and toes(Fig.3).

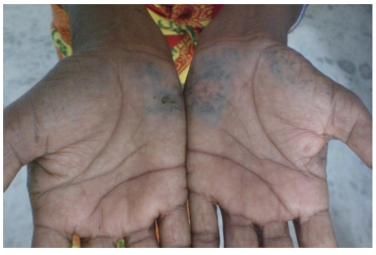

Fig.1 : Skin thickening with bluish grey pigmentation of palmar eminence of both hands.

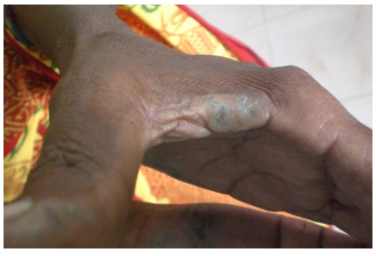

Fig.2: Thickened skin with bluish grey pigmentation of web spaces.

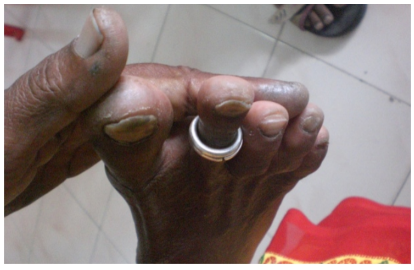

Fig.3: Bluish grey pigmentation of tips of fingers and toes.

HISTOPATHOLOGY:

Skin biopsy from the palmar lesion stained with Haematoxylin and Eosin showed hyperkeratotic epidermis with yellow - brown (ochre) coloured swollen collagen bundles in the mid dermis(Fig.4&5). Hence histopathological diagnosis of Ochronosis was offered.

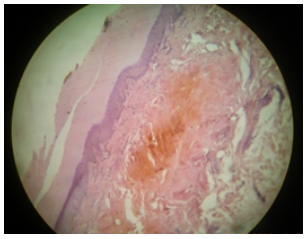

Fig.4: Low power shows hyperkeratotic epidermis with yellow - brown (ochre) coloured swollen collagen bundles in the mid dermis.

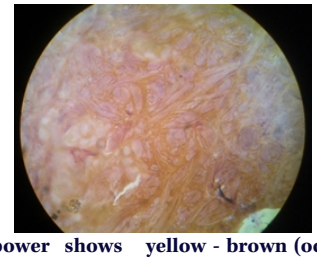

Fig.5: High power shows yellow - brown (ochre) coloured swollen collagen bundles in the mid dermis.

RETROSPECTIVE EVALUTION:

On retrospective evaluation, the patient gave history of discolouration of soil after passing urine since childhood and similar complaints among family members. Urine examination showed black discolouration after standing for 30 minutes(Fig.6) as well as after addition of sodium hydroxide(Fig.7).

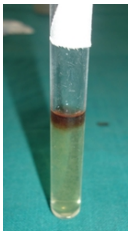

Fig.6: Black discolouration of urine after standing for 30 minutes.

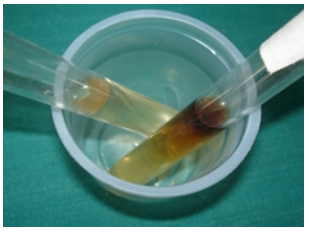

Fig.7: Black discoloured urine after addition of sodium hydroxide in comparison to without adding alkali.

DISCUSSION:

Alkaptonuria (Endogenous -Ochronosis), the first human inborn error of metabolism to be discovered, is an autosomal recessive disorder in which there is lack of homogentisic oxidase, an enzyme that converts homogentisic acid to methylacetoacetic acid in the tyrosine degradation pathway[1]. As a result, homogentisic acid accumulates in the body. A large amount is excreted, imparting a black colour to the urine if allowed to stand and undergo oxidation. The retained homogentisic acid bound irreversibly to collagen fibers as a polymer after oxidation to benzoquinone-acetic acid[2]. Homogentisic acid accumulates in the cartilage of joints, ears, nose; in ligaments and tendons; and in sclera. In this case, the presenting complaint was pigmentation of palms and soles with overlying hyperkeratosis and pitting[3]. In the course of time, homogentisic acid accumulates in the dermis in sufficient amounts to cause patchy brown pigmentation of the skin. The gene for homogentisic acid oxidase maps to chromosome 3q[4].

CONCLUSION:

Based on clinical findings, family history, urine examination and histopathological study, the diagnosis of Ochronosis (Alkaptonuria) was concluded. In view of the limited treatment options available, patient was educated about the possibilities of cardiovascular complications and arthropathy. Since the preventive nature and autosomal recessive inheritance pattern of the disease, patient was counselled against consanguineous marriage among family members.

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