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Medicine

CASE REPORT ON DARIER DISEASE- A RARE ENTITY

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ABSTRACT Darier disease is also known as Darier White disease, dyskeratosis follicularis. It is a rare autosomal dominant genetic disease manifested predominantly by symmetric reddish brown keratotic papules over the seborrhoeic areas on head, neck and thorax. This paper reports a case of a 22 years old girl presenting with reddish brown keratotic papules all over her body.

KEYWORDS :

INTRODUCTION

Darier disease was initially described by Prince Marrow in1886 and simultaneously by Darier and White in 1889, independently[1]. Darier's disease is a rare keratinisation disorder. It is an autosomal disorder with high penetrance and variable expressivity. Clinical signs include hyperkeratotic papules main;y affecting seborrheic areas on the head, neck, thorax, hands, palms and foot sole[2].

CASE REPORT

This paper reports a case of a 22 years old female who presented with the chief complaint of reddish brown keratotic papules all over the body for more than 15 years(fig. 1). The papules first appeared on the face and finally spread to all over the body. She also complaint of itching over the papules. She also gave the family history in his father who had the disease in severe form. The biopsy was taken from skin of anterior abdomen. Biopsy specimen consisted of one greyish white to greyish brown skin covered soft tissue piece measuring 1*0.8*0.4 cm. The tissue was further fixed and processed.

MICROSCOPIC FEATURES

Sections examined showed stratified squamous epithelium lined tissue showing epithelial hyperplasia, hyperkeratinisation, keratinous cyst formation. At few places suprabasal clefts and subepidermal cleft formation were seen(fig. 2,3). There was mild inflammatory cell infiltration comprising of lymphocytes.

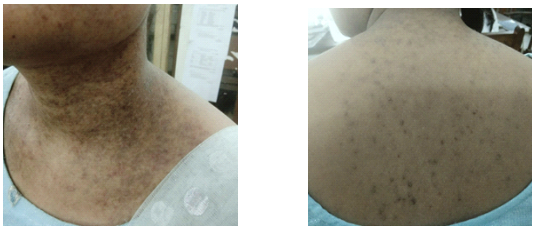

FIG. 1- SHOWING REDDISH BROWN KERATOTIC PAPULES

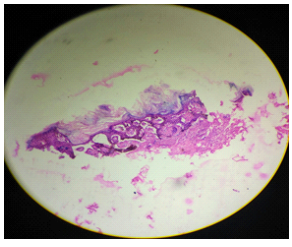

FIG. 2- 10X VIEW OF EPITHELIUM SHOWING HYPERKERATINISATION

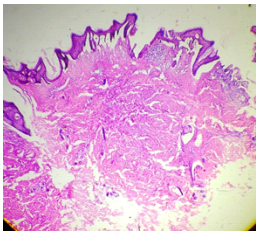

FIG 3- 40X VIEW SHOWING SUBEPIDERMAL CLEFTS ALONG WITH LYMPHOCYTIC INFILTRATE

DISCUSSION

Darier disease is a rare autosomal dominant genetic disease which is characterised by reddish brown keratotic papules particularly over the seborrhoiec areas[2]. May involve oral mucosa in 50% of the cases[3]. Initially, papules are reddish which may coalesce, forming crusts that may be ulcerated .They may give verrucous appearance if present very close together[3,4]. Prevalence of this disorder is 1:100,000[2]. Males and females are equally affected but males appear to be severely affected than females[1,2,5]. It is usually due to mutation in ATPA2 gene present on chromosome 12q23. This gene encodes for SERCA enzyme that is required to transport calcium within the cells[2,6,7]. Mutations in this gene affects calcium homeostasis and result in abnormality in desmosomal stability and adhesion[8]. Histologically, the lesions are characterised by suprabasal clefts in which acantholytic cells called grains are found. The dermal papillae covered by a layer of basal cells form small villi at the base of the lesion. In addition, within the epidermis large individually dyskeratotic cells called corp ronds are found[9,10].

Most of the patients have mild and unnoticed disease. More severe cases may have a chronic relapsing remitting pattern. Sometimes, there is association with serious psychotic disorder, particularly schizophrenia in some patients[11]. Flare ups may be caused by exposure to sunlight, topical corticosteroids, bacterial infection and herpes simplex[2].

CONCLUSION

Thus after looking all the prospects of history, physical examination, family history and microscopic findings, the diagnosis of DARIER DISEASE was made.

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