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An unusual presentation of Darier's disease in unilateral and localized nevoid/ blaschkoid pattern

Harendra Meena M.D Scholar, Department of Dermatology, Venereology & Leprology, R.N.T. Medical College, Udaipur, Rajasthan 313 001

Deepak Sharma M.D Scholar, Department of Dermatology, Venereology & Leprology, R.N.T. Medical College, Udaipur, Rajasthan 313 001

A.K Khare* M.D., Department of Dermatology, Venereology & Leprology, R.N.T. Medical College, Udaipur, Rajasthan 313 001. *Corresponding Author

Neha Agrawal M.D Scholar, Department of Dermatology, Venereology & Leprology, R.N.T. Medical College, Udaipur, Rajasthan 313 001

ABSTRACT A case of unilateral and localized nevoid/ blaschkoid pattern of Darier's disease is reported because of its rarity.

KEY WORDS : ATP2A2, SERCA2

INTRODUCTION

Darier's disease (DD) is an uncommon, inherited disorder of acantholysis characterized by keratotic papules in seborrheic distribution. The disease can rarely present in localized form, as a mosaic form following the Blaschko's line. Darier's disease in a localized pattern was first described in 1906 by Kreibich[1].We document a 39-year-old woman with unilateral nevoid pattern of Darier's disease over left side of forehead.

CASE REPORT

A 30-year-old woman presented with 12 year history of asymptomatic, gradually progressive multiple papules over left side of forehead. There was history of aggravation on sun exposure. Family history was non contributory. No oral, nail or hair abnormalities were detected. General physical examination was normal. Routine hematological and biochemical investigations were with in normal limit. Histopathological examination of the skin biopsy revealed hyperkeratosis and rete ridges elongation. Marked dyskeratosis appreciated by presence of corps ronds and grains in superficial layers of epidermis along with acantholysis. Suprabasal clefting noted with a lymphohistiocytic inflammatory infiltrate in the dermis. Findings were in consistent with Darier's disease. She responded well to topical tretinoin cream 0.05% and sunscreens, resulting in significant regression of skin lesions in 2 months.

DISCUSSION

Nevoid Darier's disease has been thought to be a mosaic form of Darier's disease. It is due to somatic mutation of ATP2A2 gene, which is located on chromosome 12q24.1 locus but not found in all those reported.ATP2A2 geneencodes for sarcoplasmic reticulum Calcium pumpingATPase type 2 (SERCA2). Defect of these channels cause acantholysis and dyskeratosis in epidermal cells because these channel have important role in keratinocyte differentiation and adhesion[2].Over 100 different mutations have been also reported in DD. Müller and colleagues suggested that focal lesions are due to the accumulation of secondary mutations which cause alteration in p53/p21 check points ofcell cycle in stressed SERCA2deficient epidermis[3].

Localized Darier's disease is referred by various terms like segmental, naevoid, zosteriform, linear, or unilateral DD. Segmental Darier's disease usually has late onset comparedto generalized form with a mean age of 27 years [4] . However congenital or infantile segmental Darier's disease has also been reported sparsely. The most common site of involvement of segmental Darier's disease is trunk. Segmental Darier's disease can be of 2 types. Type 1 which is common than type 2, follows Blaschko's lines unilaterally on a background of otherwise normal skin as a result of genetic mosaicism due to post zygotic somatic mutation early in embryogenesis.If above mentioned somatic mutation would happen early during embryogenesis, it can present in gonadal cells. Such patients may have small but real chance of having a child with generalized acantholytic dyskeratosis.

Type 2 represents focal areas of linear streaks with increased severity on a background of generalized Darier's disease.Type II mosacism occurs in setting of heterozygous germline mutation with a second hit in form of somatic loss of heterozygosity of wild type allele in segmental area, therefore increased severity in a linear array.

Late onset, absence of family history, localized distribution of skin lesions, and characteristic histopathological changes confirmed the diagnosis in our case.

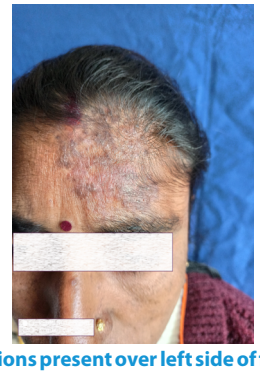

Figure 1: lesions present over left side of forehead

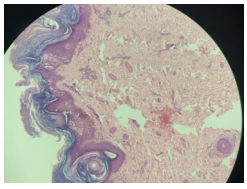

Figure 2: HPE showed hyperkeratosis, acantholysis, dyskeratotic cells

***Corresponding Author** Dr. Harpreet Singh Dhillon

MD (Psychiatry), HOD, Dept of Psychiatry Level III Hospital, Goma, DRC harpreet5467@yahoo.com

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