Darier's Disease: A rare genodermatosis

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Abstract
Darier’s disease or darier white disease or keratosis follicularis is a rare inherited autosomal dominant genodermatosis which are clinically characterized by multiple hyperpigmented, firm, greasy, warty lesion usually in seborrheic distribution. They also show palmar pits, and mucosal involvement. Histologically they present with suprabasal splitting of epidermis with presence of acantholytic and dyskeratotic cells. we report a case of a female of 30 years old who shows clinical and histological features of darier’s disease.

Keywords: Genodermatosis, Darier’s disease, Keratotic papules, Dyskeratosis, Hyperpigmented papules.

Case Report
30 year old female patient presented to dermatology OPD with complaints of dark raised lesion over face, neck, upper back and lower limb. The lesion was also present on the abdomen (Fig. 1). The lesion became itchy and infected during summer. Illness started during her teens and with multiple hyperpigmented warty lesions over the face, neck, upper back and lower limb. On examination she has multiple hyperpigmented warty papules and macules over face, neck (Fig. 2) upper back and lower limb. Nails showed diffuse light and dark longitudinal bands. She had diffuse scalp hair loss. Blood count, sugar, urea, creatinine were all normal.

Skin biopsy was taken from lesion on right side of abdomen and sent for histopathological examination showed stratified squamous epithelium with hyperkeratosis, focal vertical parakeratosis (Fig. 3), spongiosis (Fig. 5) with suprabasal splitting of epidermis, focal acantholysis, areas of dyskeratosis forming corps, ronds and grains (Fig. 4) with underlying dermis shows perivascular lymphocytic inflammatory infiltrate.

Fig. 1: Multiple hyperkeratotic papules and plaques over abdomen

Fig. 2: Presence of warty plaques over the upper back area

Fig. 3: Photomicrograph showing hyperkeratosis, parakeratosis (H&E stain x100 magnification)
The differential diagnosis includes acanthosis nigricans, confluent reticulate papillomatosis, seborrheic dermatitis, prurigo pigmentosa acne vulgaris and reticulate erythematous mucinosis syndrome. Features which differentiate these conditions from Darier are as follows. Hyperpigmented lesions are present in acanthosis nigricans. Flat lesions and distribution over upper trunk is a distinguishing feature of confluent reticulate papillomatosis. Palpation findings such as harsh papules differentiates it from prurigo pigmentosa and reticulate erythematous mucinosis syndrome which appear similar on inspection. Benign familial pemphigus, pemphigus vulgaris, Warty Dyskeratoma, Acantholytic Dyskeratosis are the histological differentials for Darier. In familial Benign Pemphigus the supra basal separation appears as a bulla, large area of epidermis is affected by Acantholysis. Corps Ronds and Grains are not prominent. Warty Dyskeratoma shows a cup shaped invagination connecting with Epidermis and Corps & Ronds are seen only in the upper portion. Acantholytic Dyskeratosis shows suprabasal cleft with overlying Acantholysis and Dyskeratotic cells. Pemphigus Vulgaris shows a suprabasilar bullae with a single row of keratinocytes and there is acantholysis in the follicular infundibula. Immunofluorescence can differentiate among different acantholytic disorders. Genetic measures such as hygienic practices, wearing loose and free clothes preferably cotton, avoiding excess heat, sunlight and use of sunscreens. Urea and lactic acid containing moisturisers and topical retinoids can decrease scaling and hyperkeratosis. Oral retinoids aids in reducing the keratinisation, smoothing of papules and also helps to reduce odor. Antibiotics and antivirals may be required to suppress secondary bacterial and viral infections. Other modalities of treatment includes dermabrasion, electrosurgery, laser ablations of recalcitrant plaques. Photodynamic therapy and surgical excision of thickened plaques has also been reported.

Irrespective of patients presentation and treatment all patients should undergo genetic counseling and should be clearly explained regarding the risks of genetic transmission. In case of oral lesions a biopsy is mandatory for final diagnosis and the patient might need a dermatological examination based on the result. Patients should be counseled regarding the complications of this disorder and the required care. Psychological evaluation and counseling may be required in severe forms, hence efficient management of the disease requires a multidisciplinary approach.

References


How to cite this article: V Divya, V Sudha, Darier’s Disease: A rare genodermatosis. *J Diagn Pathol Oncol* 2019;4(2):153-155.