CASE REPORT

TWO CASE REPORTS ON DARIER DISEASE: RARE DISORDER OF KERATINIZATION

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INTRODUCTION: Darier disease, also known as keratosis follicularis or dyskeratosis follicularis, is a rare congenital acantholytic disorder which is transmitted as an autosomal dominant inheritance with variable expressivity but complete penetrance. Darier disease was initially described by Prince Marrow in 1886 and simultaneously by Darier and White in 1889, independently. The prevalence of this disorder in the population is 1:100,000, most often affecting males.¹⁻³ Its manifestations appear in childhood and adolescence. It is characterized by persistent eruption of hyperkeratotic papules primarily over seborrheic areas, nail changes like pathognomonic triangular nicking at the free edges and histologically suprabasal acantholysis with dyskeratosis. Associated anomalies have been described in the literature, including mental retardation and psychosis.⁴

KEYWORDS: Keratinization, depression, seborrheic area.

FIRST CASE REPORT: A 22 year old unmarried female presented with dirty, warty, papular lesion all over the body since the age of one year. Initially lesion was present over the forehead and gradually progressed to involve the whole body over a period of two to three years. Lesions increases during summer and also on sun exposure. There is no family history.

CUTANEOUS EXAMINATION: Lesions were hyperpigmented, warty, papular, symmetrically present mainly on head, neck, nasolabial fold in a seborrheic distribution and on flexural aspect. Foul smelling discharge from lesion of scalp is also present.

NAIL EXAMINATION:

RIGHT HAND: Middle and little finger show "v" shaped nicking; index and ring finger show hyperpigmented dystrophic nails.

LEFT HAND: Thumb show longitudinal splitting of nail plate.

PALMS AND SOLES: Show hyperkeratosis with numerous pits.

ORAL CAVITY: Examination shows multiple small discrete papule over hard palate in cobble stone pattern. Other investigations do not reveal any abnormality.

FIGURE SHOWING: Clinical picture of the patient showing hyper pigmented, warty, papular lesions on 1(a) Forehead, 1(b) Dorsum of hands & legs.

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Fig. 1(a)

Fig. 1(b)

SECOND CASE REPORT: 30 year female presented with dirty papular lesions over seborrheic and flexural areas since puberty. The condition is gradually progressive with aggravation during summer and sun exposure.

CUTANEOUS EXAMINATION: shows papular lesions with hyperkeratotic plaques over seborrheic and flexural areas.

Nail changes and mucosal lesion are absent.

HISTOPATHOLOGICAL EXAMINATION: Histopathological examination in both cases shows hyperkeratosis and suprabasal acantholysis, abnormal keratinisation in the form of corps ronds in stratum Malpighi and grains in horny layer are also seen.

FIGURE SHOWS:

2a & 2b Hyperkeratosis & Corps ronds respectively in Case 1.

3a & 3b Showing hyperkeratosis & suprabasal acantholysis in case 2.

Fig. 2a & 2b Hyperkeratosis & Corps ronds respectively in Case 1

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Fig. 3a & 3b Showing hyperkeratosis & suprabasal acantholysis in case 2

DISCUSSION: Darier disease was initially described by Prince Marrow in 1886 and simultaneously by Darier and White in 1889, independently.

The prevalence of this disorder in the population is 1:100,000, most often affecting males.

Clinically characterized by firm, rough skin coloured, yellow-brown or brown papule distributed over seborrhoeic areas of the trunk, face, particularly the scalp margins, temples, ears and over lower legs and arms. Coalescent papules form irregular warty fissured plaques or papillomatous masses, which may become vegetating and malodorous especially in the flexures. On the scalp, a characteristic spiny feel present on palpation. On the dorsa of the hands and feet, discrete papules are present. Nail changes include red or white longitudinal bands with pathognomonic notch at the free margin of the nail. Palms and soles show minute pits or palmoplantar hyperkeratosis. Lesions of the mucous membranes are uncommon, but white umbilicated or cobblestone papules on the palate may be seen. The rash is typically exacerbated by heat and light, especially sunburn or by UVB exposure. Spontaneous remissions do occur, but the disease usually runs a chronic relapsing course, without affecting general health.

HISTOPATHOLOGY: The characteristic change

- Peculiar form of dyskeratosis of resulting in formation of corps ronds and grains.
- Suprabasal acantholysis leading to formation of suprabasal cleft or lacunae.
- Irregular upward proliferation into the lacunae of papillae lined with a single layer of basal cells.

CAUSES: Mutations in the ATP2A2 gene cause functional disruptions in all domains of SERCA2 (the calcium pump), including reduction of ATP affinity, loss of calcium affinity, effects on phosphorylation of ATP and Pi, blocking of dephosphorylation, and uncoupling of calcium transport from ATP hydrolysis.

These defects cause alterations of the cytosolic calcium level. Ultimately, these changes influence adhesion between keratinocytes and cellular differentiation in the epidermis.

TREATMENT: Emollients, avoidance of sun exposure, topical retinoids, antiseptics or steroid/antibiotic combination (for infected plaques) are useful. For more severe disease, oral

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retinoids (alone or with vit.E) are usually effective. Dermabrasion, photodynamic therapy, laser are also useful.

CONCLUSION: The implications of the Darier disease are more associated with cosmetic and aesthetic than functional implications, since this is a benign dermatosis. However, depending on the severity of the disease and affected area, the patient has more complaints and the emotional status may be damaged by aesthetic reasons. So along with medical treatment psychological and genetic counselling is mandatory. Therefore, these patients should be treated by a multidisciplinary team.

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