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DARIER'S DISEASE OF MODERATE SEVERITY WITH MUCOSAL AND NAIL INVOLVEMENT

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ABSTRACT

Darier's disease, also known as keratosis follicularis, dyskeratosis follicularis or Darier-White disease, is a rare, inherited, acantholytic disorder caused by mutation in ATP2A2 gene, which manifests with hyperkeratotic lesions of variable severity. It typically occurs in young age and presents with itching and greasy appearance. The lesions are persistent and disease follows a chronic course with protean prognosis. Extracutaneous involvement may occur in mucosa, nail, eye and salivary gland. We report a case of Darier's disease of teenage onset, and moderate severity with palmoplantar, mucosal and nail involvement.

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INTRODUCTION

Darier's disease, also known as keratosis follicularis, dyskeratosis follicularis or Darier-White disease, is a rare, autosomal dominant genodermatosis, due to mutation in ATP2A2 gene leading to defective calcium pump (1). The disease is characterized by loss of adhesion between epidermal cells and abnormal keratinization leading to acantholysis. It is a chronic disease, which is characterized by aggregated keratotic papules, predominantly in seborrhoeic distribution on face and trunk. It usually has a teenage or adult onset. The disease has a worldwide distribution, with prevalence from 1 in 30,000 to 1 in 100,000 in Europe and affects both sexes equally (2). Treatment involves avoidance of aggravating factors and use of topical keratolytics and oral retinoids. We hereby report a classical case of Darier's disease owing to its rare occurence.

CASE REPORT

A 25 year old, male presented with the chief complaint of generalised, small, brown coloured lesions since 15 years. The lesions were insidious in onset and started over the scalp with few, small, skin coloured lesions which gradually progressed all over the body, in the past 10 years. There was mild itching and malodour of skin. The lesions got aggravated in summer and relieved in winter. There was no similar family history. On cutaneous examination, there was presence of, discrete to confluent, rough, greasy, small, brown, hyperkeratotic papules, all over the body, predominantly over the scalp, face, ears, neck, trunk, axillae and groin (Figure 1a-c).

Fig 1a: Greasy, Hyperkeratotic Papules Over Face And Neck

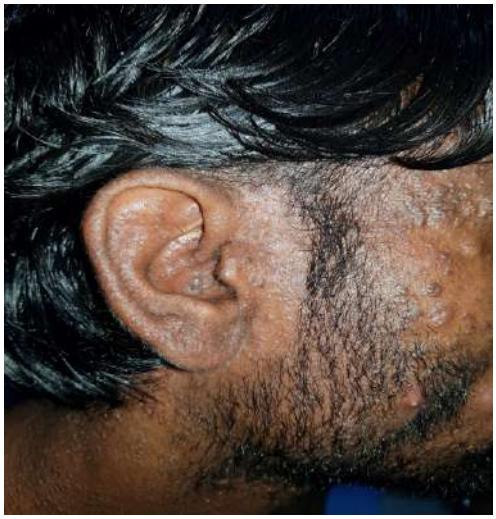

Fig 1b: Coalescing, Keratotic Papules Over Face, Ears And Neck

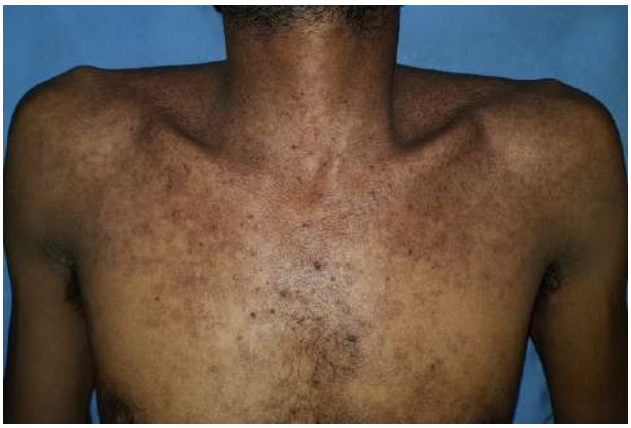

Fig 1c: "Dirty Warty Appearance" Of Papules Over Neck, Chest And Shoulders

Multiple, skin colored, flat topped papules with rough surface were present on the dorsum of hands and feet, forearms and legs. Multiple pits and focal keratosis of palms, were seen (Figure 2).

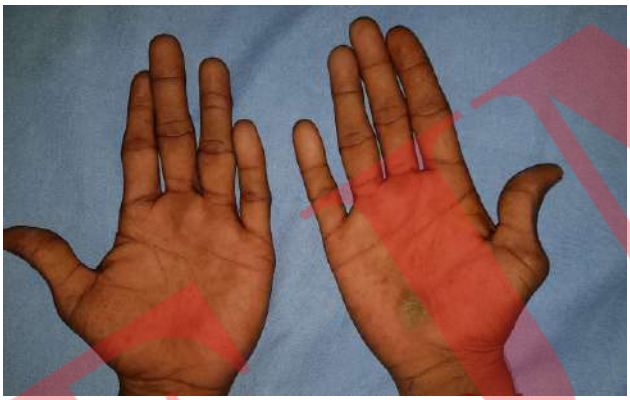

Fig 2: Multiple Palmar Pits With Punctate Keratosis

Patient, was affected with grade II (moderate) severity, with <30% body surface area (BSA) involvement. Examination of oral mucosa revealed white, umbilicated and cobblestone papules on the palate (Figure 3).

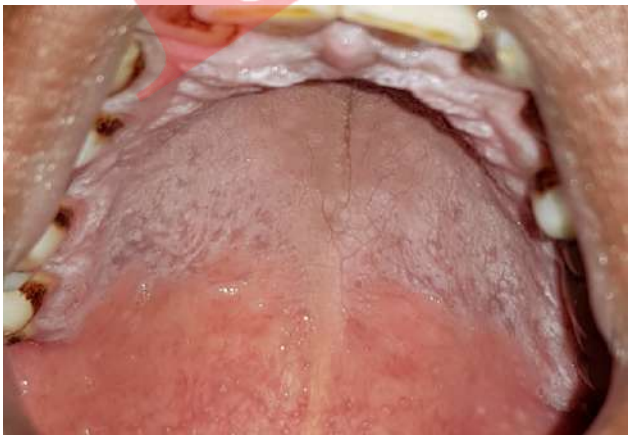

Fig 3: Cobblestone Appearance Of The Palate

Brittle nails with multiple, red and white longitudinal bands terminating in V-shaped nicks were present in the fingernails (Figure 4a and 4b). Neurological examination revealed no abnormality.

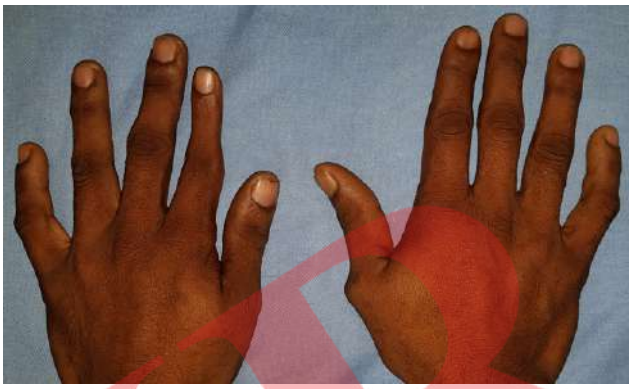

Fig 4a: Flat Papules On Dorsum Of Hands And Distal, Triangular Notching Of Nails

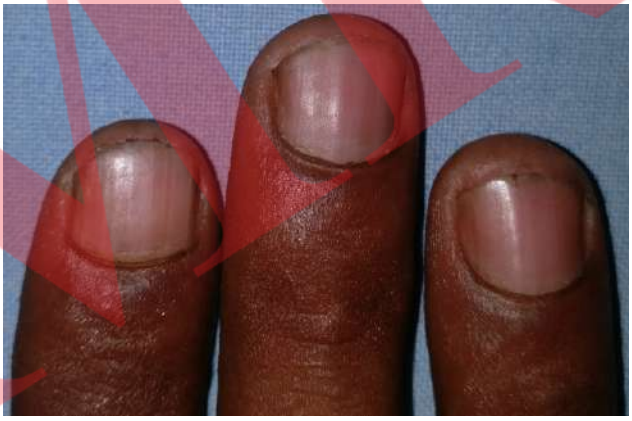

Fig 4b: "Sandwich" Of Red And White Longitudinal Bands Of Fingernails

Skin biopsy showed hyperkeratosis, irregular acanthosis and papillomatosis. The granular layer, showed intracellular basophilic granular material. Focal and mild acantholytic dyskeratosis in suprabasal layer was seen. Superficial dermis had collection of lymphocytes (Figure 5).

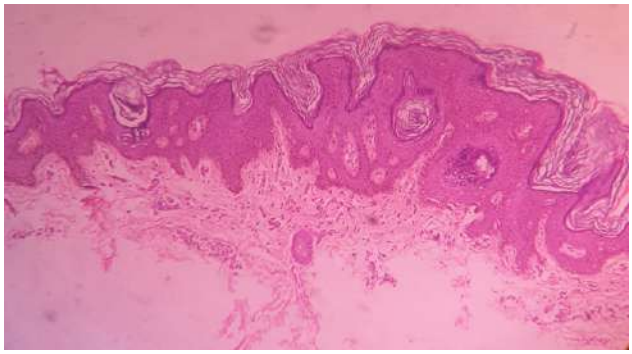

Fig 5: H & E (40x Magnification) Photomicrograph, Showing Hyperkeratosis, Acanthosis, Papillomatosis And Focal Acantholytic Dyskeratosis In Suprabasal Layer

Based on history, examination and histopathology, a diagnosis of Darier's disease was made. Patient was treated with Isotretinoin 30 mg once daily and emollients. The lesions partially resolved in 1 month.

DISCUSSION

Darier's disease was first described by Darier and White in 1889 (1). It typically presents with greasy, warty, skin colored to erythematous brown colored papules, which are symmetrically distributed, predominantly over face and trunk (3). "Sandwich" of longitudinal, alternating, white and red bands, is the most pathognomonic nail sign (4). The rare clinical presentations are zosteriform, linear, cornifying, vesiculobullous, isolated acral hemorrhagic, acrokeratosis verruciformis of Hopf (AKV), comedonal and leukodermic macules. The ATP2A2 gene, encodes sarco/endoplasmic reticulum Ca2+ ATPase type 2 (SERCA2), its defect causes abnormal intracellular Ca^{2+} signaling and decreases calcium store in endoplasmic reticulum, leading to poor connection between keratinocytes (5). A family history is usually absent (6). The commonest clinical differential diagnosis include seborrheic dermatitis and acrokeratosis verruciformis of Hopf (AKV). [1-3,6] Neuropsychiatric disorders, of depression, bipolar disorder, schizophrenia, epilepsy and intellectual difficulties, are more frequent than in the general population. Histopathologically, the characteristic finding is acantholysis and dyskeratosis. It follows a relapsing course, without spontaneous remission and has an unpredictable severity. Avoiding aggravating factors and treatment with topical keratolytics and oral retinoid are helpful (1-3). Our case was unique because the patient had classical presentation of the disease with both mucosal and nail involvement and was classified with grade II (moderate) severity (1). We report this case due to its rarity, especially in Indian subcontinent.

CONCLUSION

Darier disease is a rare genodermatosis, which presents with greasy, erythematous papules over seborrheic areas and trunk. The disease shows an aggravation in summer and the diagnosis is confirmed by histopathological examination. It follows a chronic course and treatment is usually of long duration. Early recognition of this condition is important for its holistic management. The associated psychosocial stress should also be promptly addressed to improve the patients quality of life.

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