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Case Report
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# A Sporadic Case of Darier's Disease

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***Abstract***

*Darier's disease, an autosomal-dominant genodermatosis, which is due to loss of function mutations in ATP2A2 gene encoding, sarco/endoplasmic reticulum calcium adenosine triphosphate isoform 2 (SERCA 2) that impair intracellular Ca2+ signaling.*

*This article reports a case of sporadic Darier's disease which involved the seborrheic regions (scalp, face, neck, axilla, anterior and posterior trunk and groin) and nails.*

*A remarkable improvement was noted on oral isotretinoin, after 6 months on topical retinoid (tretinoin) and sunscreen with minimal progress. She had no family history of Darier's disease or a similar rash.*

**Keywords:** Cardiopulmonary Resuscitation (CPR), Knowledge, Attitude, Practice, Awareness

## Case Report

A 27year old female presented in February of 2021 with a 19year history of recurrent keratotic pruritic papular rashes. Which were malodourous located on her face, neck, chest and back, axilla, groin, soles of her feet and scalp as crusted plaques with nail abnormalities. She also complained of odour and pruritus of skin lesion which was aggravated by heat, sunlight and sweating mostly noticed when in a crowded place. She has no family history of Darier's disease nor a similar skin lesion.

- Physical examination - revealed multiple skin-coloured and hyperpigmented hyperkeratotic papules, coalescing into plaques on the anterior and posterior trunk and crusted plaques on her scalp.

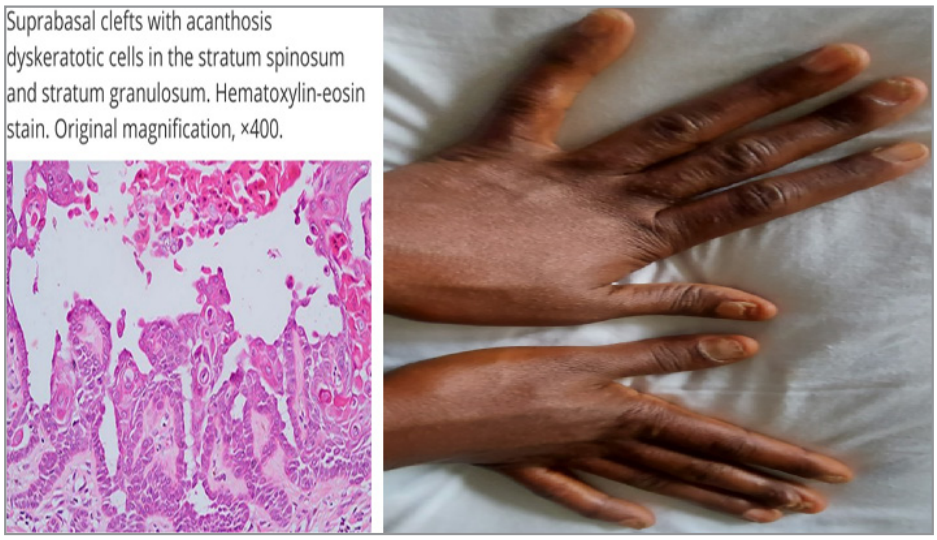
Skin biopsy of sections - of skin tissues revealed epidermis exhibiting hyperkeratosis, acanthosis and ancantholysis. Beneath the stratum granulosum, were some epithelial cells with pyknotic nuclei and brightly eosinophilic cytoplasm (dyskeratotic cells). Also, suprabasal cefts lined by single layer of basal cells seen in some areas and fibrocollagenization of the dermis.

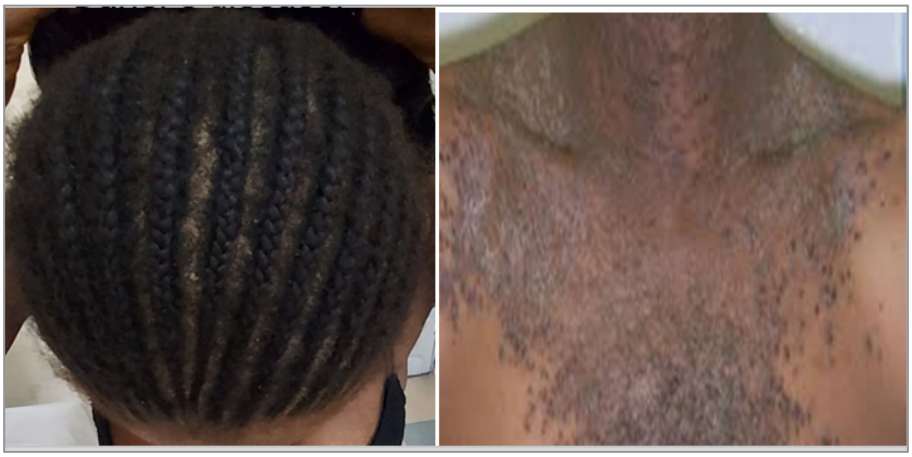
- Clinical and histologic findings - consistent with Darier's disease.
- Management - Retinoid cream (tretinoin), sunscreen with a sun protective factor of 30 or more, oral doxycycline 100mg bd were prescribed and patient advised on wearing cotton clothes, stay out of sun as much as possible and to avoid crowded places.

Follow-up - Minimal improvement was noted after 6 months of treatment. As such, patient was started on oral isotretinoin at a dose of 20mg daily, following adequate counselling on the side effects of oral isotretinoin, the need for contraception and consent obtained.

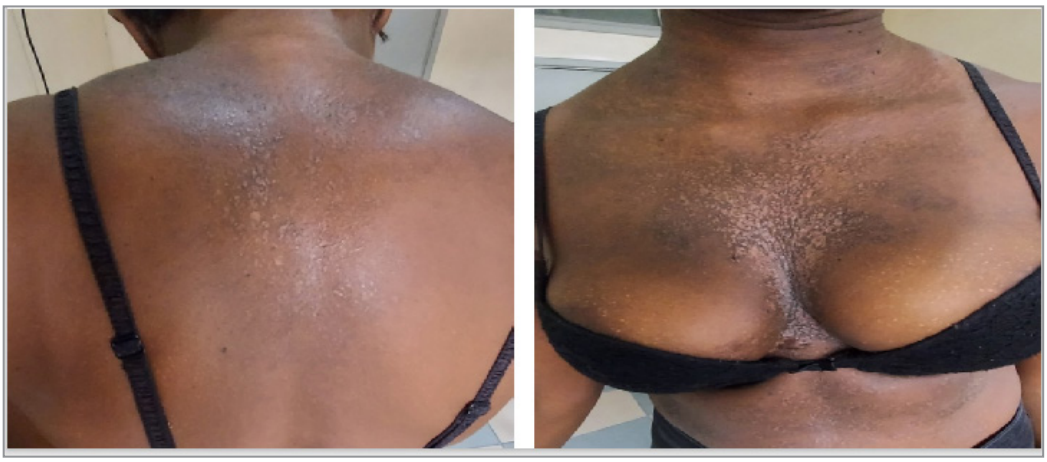
- By the next clinic visit, a month after the commencement of oral isotretinoin significant improvement was noticed as evidenced by improvement of all skin lesions.

**Images**



Scalp, neck and anterior truncal lesions of Darier's disease.



Resolving skin lesions in a Darier's patient after 2months on oral isotretinoin

## Discussion

Darie's disease is an autosomal dominant disease with a high penetrance and variable expressivity, first described by prince Marrow in 1886, later independently by Darier and White in 1889 [1, 2]. Darier named the disease psorospermose folliculaire vegetante. Although Darier's disease is an inheritable disease, approximately 47% of cases are reported to be sporadic with no family history [3]. A negative family history of Darier's disease could be related to the fact that a mild form was of the disease was not been recognized among the family members [4].

However, in this index case, a thorough family history from patient did not reveal a similar skin lesion in any member of her family suggesting a new mutation or an evidence of incomplete penetrance. The mutated gene ATP2A2 involved in the pathogenesis of this condition, encodes for the SERCA2 protein. The SERCA2 is involved in regulating cytosolic Ca2+ concentration which in turn regulates the assembly of desmosomes.

The isoform SERCA2 is widely expressed in the epidermis. Mutation in ATP2A2, thus results in deregulation of intercellular keratinocyte attachment through impaired localization of desmoplakins to the desmosome, resulting in acantholysis [5]. Skin lesion in the index case were hyperkeratotic papules on the face, neck, chest, back. Axilla and pubs, crusted plaques on the scalp and nails abnormalities. This is similar to the presentation report by Hema, et al [6].

Nail abnormalities on examination were thinning and fragility of the mails, longitudinal fissures on the distal aspect of the nails and V-shaped splitting. There was no mucosal lesion, also no multi organ involvement in the case report. Several studies have demonstrated extracutaneous manifestation of the bones, urogenital, corneal, neuropsychiatry, heart, pulmonary in Darier's disease [7-9].

In this case there was no extracutaneous manifestation, however, a low self-esteem was noted in the patient prior to treatment.

## Conclusion

Darier's disease is a rare genodermatosis which can present atypically and diagnosis can be confirmed following a skin biopsy. The use of oral isotretinoin and counselling in this study was effective.

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