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Darier Disease: Case Report

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ABSTRACT

Background: A change in a gene called Ca^{2+}-ATPase type 2, which controls the movement of calcium in the endoplasmic reticulum, causes Darier disease. Darier disease is a rare skin condition that affects how cells stick together. Also, this illness gets worse with heat and exposure to the sun, sweating, and rubbing, and it is characterized by many bumpy, greasy spots with an oily skin distribution. Sometimes, it can also harm the mucosa (the lining inside our body) and cause our nails to grow strangely. There is a special type of Darier disease called unilateral segmental Darier disease. It causes red, scaly bumps to appear on one side of the body and they are not related to any other illnesses.

Objective: To show an example of a person with Darier's disease. Also, to talks about the main characteristics and the treatments that are available. **Subject and methods:** A case report. **Results:** 51 years old male patient presented with multiple red to brown hyperkeratotic papules coalesce into plaque and led to thickened skin affecting the face, trunk, axilla and dorsa of both upper and lower extremities. Histopathology, there was hyperkeratosis, acanthosis, and papillomatosis. Supra basal acantholytic blister containing acantholytic cells, some of them showed Evidence of dyskeratosis. Corps ronds are present in the uppermost layer, especially the granular layer. Grains are present within the horney layer. No signs of malignancy or granuloma. **Conclusion:** Because there are no confirmed ways to cure DD, it is hard to treat. There are many different ways to treat this problem, like using special creams or medications, having surgery, or using lasers. But these treatments don't work very well and only help a little bit**.** It is important to ensure a multidisciplinary approach in the management of patients with Darier's disease because systemic retinoids have shown to be more effective in achieving some reduction of symptoms in 90% of patients. Isotretinoin was used in our patient with a good outcome because it reduces hyperkeratosis, smoothens papules, and reduces odour.

Keywords: Male, Face, Papules, Darier disease.

INTRODUCTION

The rare genetic skin condition called Darier disease (DD), also known as keratosis follicularis, which passes down in families and can be inherited from just one parent. It can appear differently in different people. Acantholysis happens when there is a change in a gene called ATP2A2 in skin cells. This change makes the skin cells not stick together as well. It affects both boys and girls equally and is found in about 1 in every 30,000 to 1 in every 100,000 people in the general population. It often starts to appear when a person is in their 20s or 30s. The most common symptoms of DD are itchy, widespread bumps and raised areas on the body, especially in areas with more oil and moisture [1].

Darier's disease can appear as lighter patches of skin with a pattern around the hair follicles in people with darker skin. Also, a few DD patients who have the segmental subtype have specific areas of damage. In histology, DD often shows splitting in the layers above the base, abnormal skin cells, round bodies, and small particles along with loss of skin cell connections. Infections like herpes simplex virus can make treating DD harder and cause serious harm. Damaging one's self-esteem and relationships, DD can greatly impact a patient's overall happiness and well-being. Moreover, studies have indicated that developmental disabilities (DD) and neuropsychiatric issues might exist together [2].

Ethical approval: written consent to publish was obtained from the patient, before they participated in the study. The study was conducted in accordance with the Declaration of Helsinki, and the protocol was approved by the Ethics Committee of Al-Badari hospital, Egypt.

CASE REPORT

51 years old male patient presented with multiple pruritic papules on the face, trunk, axilla, and both upper and lower extremities of 20 years duration (Figure1). On examination we found multiple red to brown hyperkeratotic papules coalesce into plaque and led to thickened skin affecting the face, trunk, axilla, dorsa of the hand and both upper and lower extremities (Figures 2-5). Histopathology showed hyperkeratosis, acanthosis, and papillomatosis. Supra basal acantholytic blister containing acantholytic cells, some of them showed Evidence of dyskeratosis. Corps ronds are present in the uppermost layer, especially the granular layer. Grains are present within the horney layer. No signs of malignancy or granuloma (figure 6). Treatment options were:

General treatment that included light clothes and sunscreen. Daily skin care antibiotics, antifungal agents and keratolytic emollients.

Topical treatment: topical retinoids and 5-fluorouracil, corticosteroids.

Systemic treatment: oral retinoids (isotretinoin and acitretin). Cyclosporine in severe cases that do not respond to retinoids.

Surgical treatment: Excision and grafting, Dermabrasion and Laser removal.

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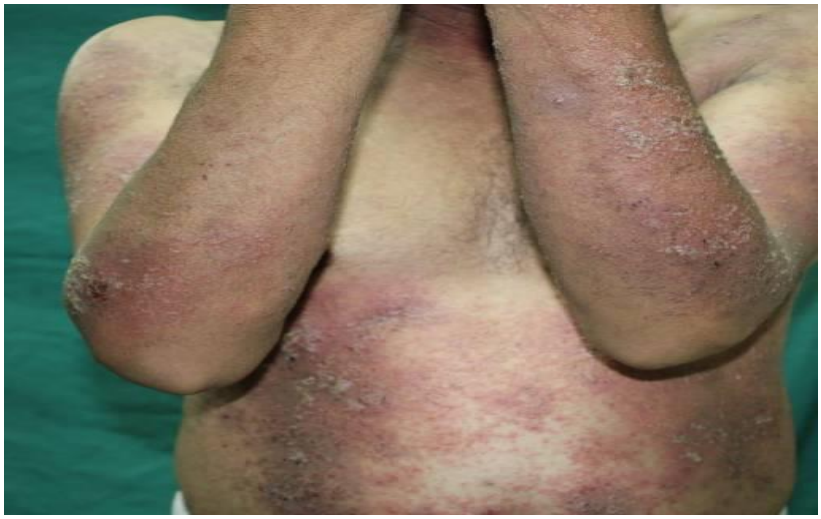

Figure (1): Upper limb and trunk affection.

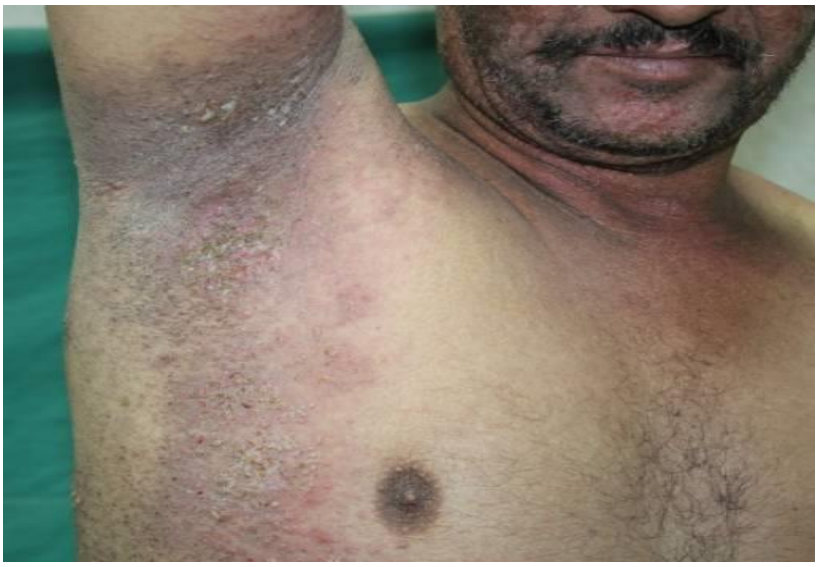

Figure (2): Axilla affection

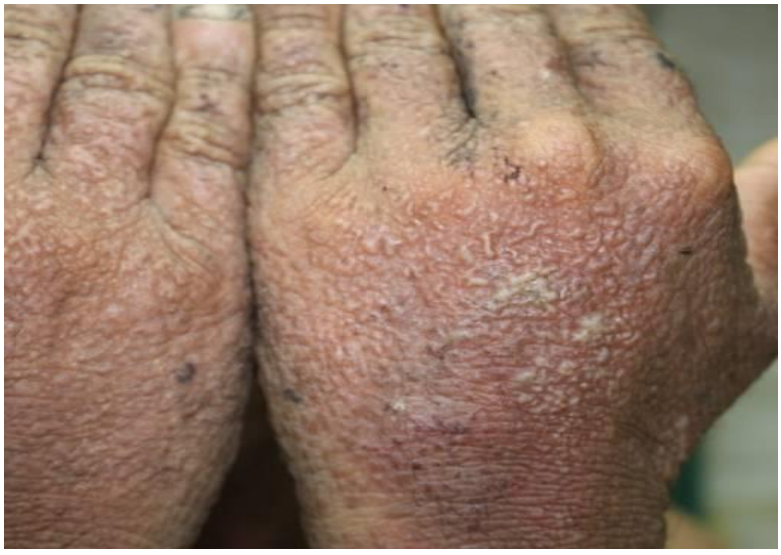

Figure (3): Hands affection

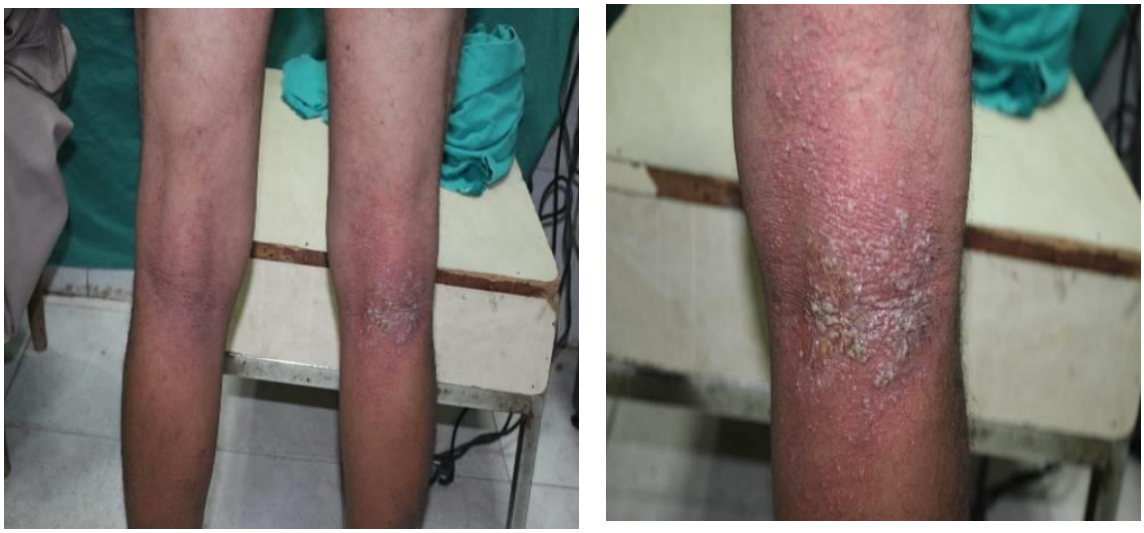

Figure (4): Lower limb affection.

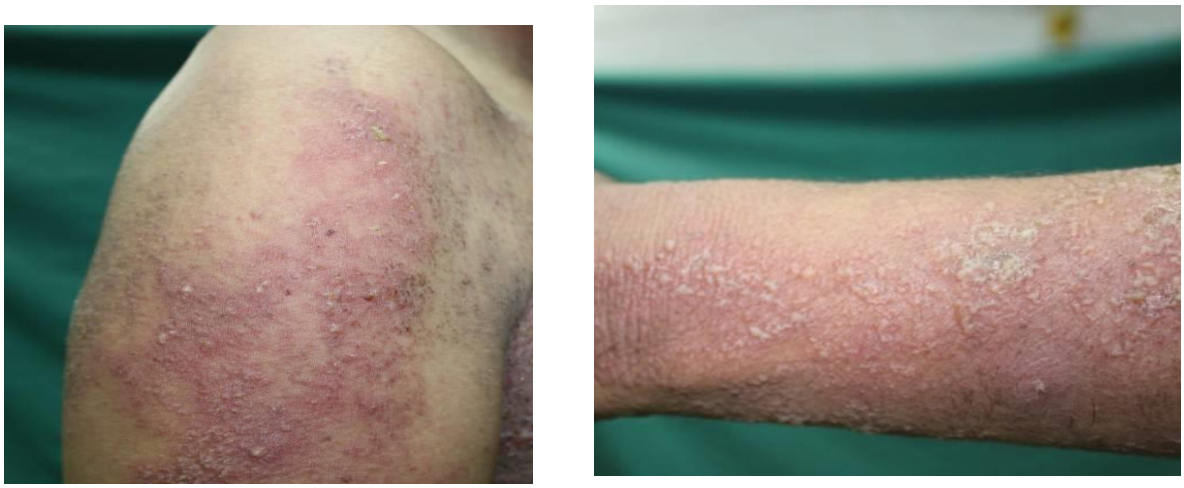

Figure (5): Upper limb affection

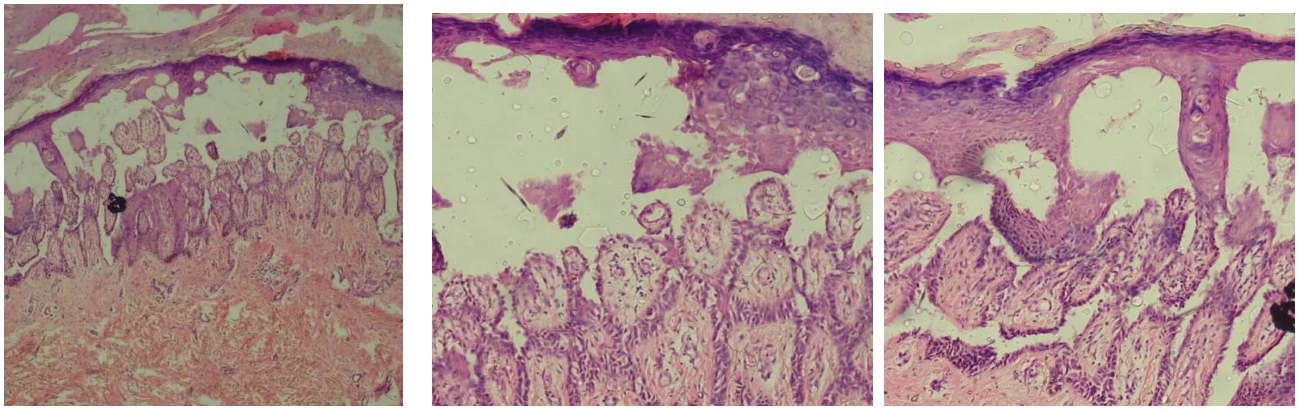

Figure (6): Hyperkeratosis, acanthosis, and papillomatosis. Supra basal acantholytic blister containing acantholytic cells, some of them showed evident dyskeratosis. Corps ronds were present in the uppermost layer, especially the granular layer. Grains were present within the horney layer. No signs of malignancy or granuloma.

DISCUSSION

Between 1 in every 30,000 to 100,000 people will be affected by Darier disease. It is a skin condition that passes down through genes and does not favor any specific gender [(4)].

Besides the bumpy marks on the top of their hands and feet and holes on their palms and under their nails, you might also watch rough bumps in oily or hidden areas on their skin. The current situation only slightly showed these conclusions [(4)].

Acantholysis and dyskeratosis are features of Darier's disease. They are shown by "corps ronds" and "grains" when you look at the tissue under a microscope. Corps ronds are basophilic masses that are found in the granular layer of the skin, and they have a clear halo around them. Electron microscopy can show the loss of desmosomes, the failure of keratin intermediate filaments to attach, and the aggregation of keratin intermediate filaments near the cell nucleus. The results from examining the tissue in our patient's body were consistent with what we expected [(5)].

Possible diagnoses for the condition could include skin conditions like dandruff, pimples, dark patches on the skin, bumpy skin with a net-like pattern, and itchy discolored bumps. The condition must be differentiated from Grover's disease, pemphigus vulgaris, and Hailey-Hailey disease by examining tissue samples. Skin biopsy immunofluorescence can help identify different skin disorders that cause acantholysis [(5)].

Because there are no confirmed ways to cure DD, it is hard to treat. There are many different ways to treat this problem, like using special creams or medications, having surgery, or using lasers. But these treatments don't work very well and only help a little bit [(6)].

It is important to ensure a multidisciplinary approach in the management of patients with Darier's disease because systemic retinoids have shown to be more effective in achieving some reduction of symptoms in 90% of patients. Isotretinoin was used in our patient with a good outcome because it reduces hyperkeratosis, smoothens papules, and reduces odour [(7)].

The main problem with our case is that we did not do enough tests to confirm and link the genetic, molecular, and functional similarities between DD and AKVH, even though the clinical and histologic evidence suggest it.

The person should get advice from a genetic counselor about their inherited disease and the chances of passing it on to their children. This is important to do, even if the disease isn't very severe or there are different treatment options available. To confirm the final diagnosis of this finding, it is necessary to have a biopsy. Patients need to be told about the difficulties that can arise from this condition and the important care they need to receive. A group of people with different specialties should take care of treating these patients.

DECLARATIONS

- **Consent for Publication:** I confirm that all authors accept the manuscript for submission
- **Availability of data & material:** Available
- **Competing interests:** none
- **Funding:** No fund
- **Conflicts of Interest:** The authors declared no conflicts of interest regarding the publication of this paper.

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