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DERMOSCOPY OF A RARE GENODERMATOSIS: DARIER'S DISEASE

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AUTHOR AND AFFILIATION

Kenza Khachani[1], Meryeme Marhraoui[1] , Mariame Meziane[1] , Laila Benzekri[1], Nadia Ismaili[1]

[1] Department of Dermatology, Mohammed V University in Rabat, Ibn Sina University Hospital, Morocco.

Corresponding author: Kenza Khachani,

ABSTRACT

Darier's disease is a rare autosomal dominant genodermatosis with a chronic, relapsing course that can pose diagnostic challenges during severe flares. We report the case of a 30-year-old woman with a longstanding history of Darier's disease presenting with an extensive, erosive flare involving the neck, trunk and flexural folds. Dermoscopy revealed a pinkish background with characteristic polygonal, star-like brown areas, while onychoscopy showed a pathognomonic V-shaped notch with red-and-white longitudinal bands. Histology confirmed suprabasal acantholytic dyskeratosis with typical round bodies and grains. Notably, dermoscopy also uncovered post-inflammatory hypopigmentation, a feature not typically seen in other inflammatory dermatoses. This case highlights dermoscopy and onychoscopy as fast, accessible, non-invasive tools that sharpen clinical suspicion, guide biopsy site selection and support long-term monitoring of Darier's disease, offering real potential to shorten the diagnostic pathway for this rare but manageable disorder.

KEYWORDS

Darier's disease, genodermatosis, dermoscopy, polygonal brown areas, onychoscopy, V-shaped notch

MAIN ARTICLE

INTRODUCTION:

Darier's disease is a rare autosomal dominant genodermatosis caused by mutations in ATP2A2, which encodes the endoplasmic reticulum calcium ATPase. It follows a chronic, relapsing course, typically presenting with recurrent reddish-brown keratotic papules and plaques affecting seborrheic areas [1]. Although diagnosis usually relies on clinical examination and histology, dermoscopy has emerged as a valuable, non-invasive adjunct that enhances recognition of active lesions and disease follow-up, and may also reveal post-inflammatory changes not typically seen in other inflammatory dermatoses [2]. We report a case of severe Darier's disease flare, highlighting its characteristic dermoscopic and onychoscopic features.

CLINICAL CASE:

A 30-year-old woman with no relevant medical history, followed for Darier's disease since age 4, was admitted for a severe flare. Examination revealed large erosive, oozing keratotic plaques on the neck, trunk, axillary, inguinal and intergluteal folds, papules on the dorsal hands and feet, with nail involvement (Figure 1).

Dermoscopy revealed a pinkish background featuring centrally located brownish/yellowish polygonal, star-like and roundish areas, each surrounded by a thin white halo. Active lesions showed scattered brown crusts, a few white scales and a hyperkeratotic follicular plug without any vascular pattern. Hypopigmented post-inflammatory areas were noted. Onychoscopy showed a V-shaped notch with alternating red and white longitudinal bands (Figure 2).

Histology showed an acanthotic, papillomatous epidermis with suprabasal acantholysis, dyskeratotic cells (round bodies and grains), and villi lined by a single layer of basal cells, consistent with Darier's disease.

DISCUSSION:

Dermoscopy enhances both clinical recognition and follow-up of Darier's disease by identifying distinctive features in active lesions, such as a pinkish background with polygonal or star-like brown areas. It also reveals post-inflammatory changes, such as the hypopigmented areas observed in our patient, not seen in other inflammatory dermatoses [2].

Furthermore, it helps guide biopsy site selection and allows retrospective diagnosis. In our patient, the combination of clinical, dermoscopic and histologic findings established a straightforward diagnosis despite the severity of the flare. Systemic retinoids remain the most effective therapy for widespread lesions, reducing hyperkeratosis by acting on the epidermal component [1].

CONCLUSION:

This case illustrates the typical dermoscopic and onychoscopic features of Darier's disease, a rare autosomal dominant genodermatosis. Dermoscopy is a valuable, accessible tool that supports clinical diagnosis, guides biopsy site selection and helps monitor disease activity, paving the way to faster and more confident recognition of this rare disorder.

FIGURES:

Figure 1: Clinical presentation of Darier's Disease: erosive plaques on the neck (A and B) and keratotic papules on the chest, neck and shoulders (B) and the dorsum of the hands with nail involvement (C).

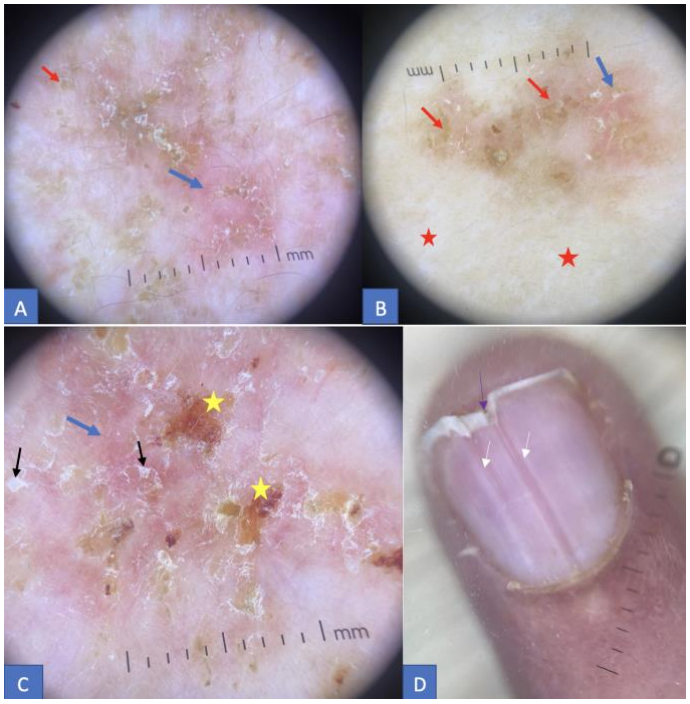

Figure 2: Dermoscopy of Darier's disease (A, B, C): Pinkish background (Blue arrow), Scattered brown crusts (yellow star), White scales (Black arrow), Post-inflammatory hypopigmented areas (red star). D: Onychoscopy of the fourth finger: V-shaped notch (purple arrow), alternating red and white longitudinal bands (white arrow).

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