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## Case Presentation

### Segmental lesions along blaschko's lines in an elderly man

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## Abstract

Darier disease (DD) is an autosomal dominant genodermatosis characterized by multiple keratotic and crusted papules over seborrheic areas, along with a variable involvement of oral mucosa, palmoplantar region, and nails. Segmental subtypes (type 1 and 2) are uncommon clinically limited forms of DD that usually present at middle age with few cutaneous lesions following Blaschko's lines. We report a case of extensive multi segmental DD type 1 that developed in an elderly man, an unusual clinical onset of DD that dermatologists should bear in mind.

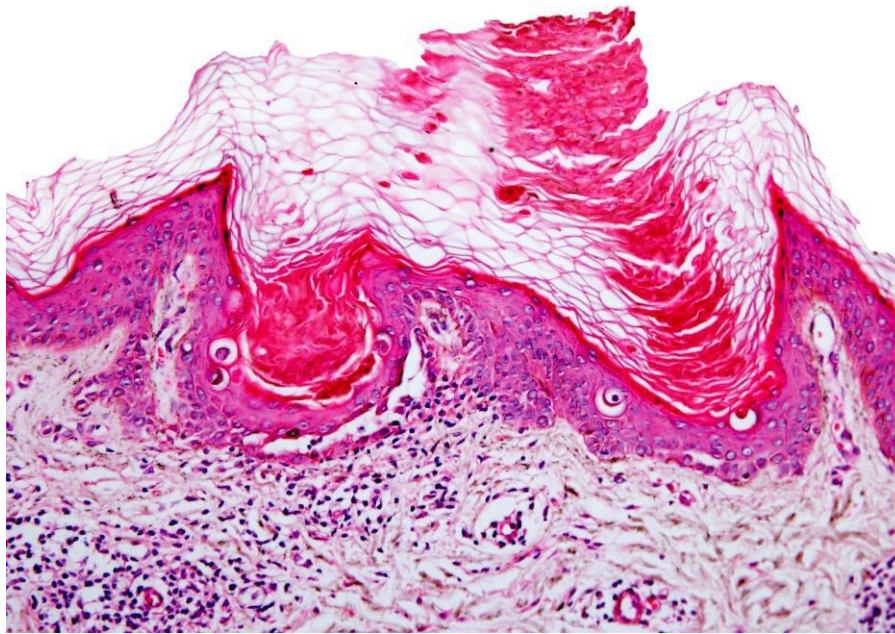
**Keywords:** Blaschko, Darier disease, acantholytic dyskeratosis.

## Introduction

Darier disease (DD) is an autosomal dominant genodermatosis caused by mutations in the ATP2A2 gene at chromosome 12q24.1, which encodes the sarco/endoplasmic reticulum calcium ATPase type 2 (SERCA2) [1]. Its most common presentation affects teenagers and young adults and consists of multiple keratotic, crusted, red to brown papules involving seborrheic areas. Palmoplantar lesions, nail abnormalities, and mucosal changes may occur as well. Segmental subtypes (type 1 and 2) are uncommon clinically limited forms of DD that usually present during middle age with few cutaneous lesions following Blaschko's lines [2]. Segmental type 1 is caused by a postzygotic mutation in the ATP2A gene during embryogenesis, which leads to a mosaic pattern of cutaneous involvement. In contrast, segmental DD type 2 develops in patients with generalized DD as a result of a heterozygous germline mutation compounded by a postzygotic mutation.

## Case synopsis

A 64-year-old man presented at our department in August 1996 because of the progressive appearance of brown pruritic papules on his left lower limb for one year. Interestingly, lesions were arranged following Blaschko's lines and he noted flaring during summer. There was no history of similar cutaneous problems in his family and mucosal surfaces, nails, and hair were normal. Histological examination from a skin biopsy specimen showed acantholytic dyskeratosis (Figure 1).



**Figure 1.** Haematoxylin and eosin staining of a skin biopsy demonstrates hyperkeratosis, with a parakeratotic column, and dyskeratotic cells in different strata of the epidermis. Acantholytic cells that give rise to a suprabasal slit.

With the diagnosis of segmental DD type 1, tretinoin 0.1% cream once daily was initiated. A noticeable improvement was observed at first, but new outbreaks over the left flank, left arm, and contiguous shoulder occurred throughout the summers of 1997-2008. During those years, additional biopsies confirmed previous histopathological findings and the patient received several cycles of oral retinoids with good response. He did not return to clinic until May 2014, when brown pruritic papules along Blaschko's lines had progressively reappeared on his left hemi-abdomen and left lower limb during the last ten months (Figure 2).



**Figure 2.** (a) Crusted brown papules, in the left hemiabdomen and the left lower limb, along Blaschko's lines (b) Detail of abdominal involvement. (c) Involvement of the left lower limb, following a linear distribution.

## Discussion

Although segmental DD type 1 is widely known as localized, segmental, zosteriform, linear, or unilateral DD, some authors consider this dermatosis a variant of epidermal nevus with acantholytic dyskeratosis, rather than a segmental form of DD; consequently some authors prefer to name it *acantholytic dyskeratotic epidermal nevus*. This issue remains under discussion

since the typical mutation of DD in the ATP2A2 gene has been found in lesional skin of patients with acantholytic dyskeratotic epidermal nevus, but not in leukocytes or unaffected skin, supporting the designation of segmental DD. [3, 4]. Moreover, two cases of blaschkoid cutaneous lesions have been described in association with ipsilateral nail and palmar changes manifestations similarly to the generalized form [5, 6].

Segmental DD is estimated to occur in about 10% of patients with DD and patients characteristically have a later onset compared to the generalized variant, appearing during the third or fourth decade of life. According to scientific literature, most cases of type 1 segmental DD report a limited cutaneous involvement with only one or a few segments affected. The extensive multi-segmental distribution of cutaneous lesions observed in our patient, along with the strikingly late appearance of them represent two unusual features rarely described in type 1 segmental DD; 4 cases have been reported in individuals over the age of 60 [7, 8, 9, 10] and only two of them exhibited a multi-segmental involvement [9, 10].

The best known clinical picture of DD is an eruption of hyperkeratotic brownish papules on seborrheic areas. However, keeping in mind unusual and misleading presentations of this entity is essential to consider DD within the differential diagnosis of acquired blaschkoid dermatosis.

## References

1. Sakuntabhai A, Ruiz Pérez V, Carter S, Jacobsen N, Burge S, Monk S, et al. Mutations in ATP2A2, encoding a Ca<sup>2+</sup> pump, cause Darier disease. *Nat Genet* 1999;21:271-7. [PMID:10080178].
2. Happle R. A rule concerning the segmental manifestation of autosomal dominant skin disorders: Review of clinical examples providing evidence for dichotomous types of severity. *Arch Dermatol* 1997;133(12):1505-9. [PMID: 9420534].
3. Sakuntabhai A, Dhitavat J, Burge S, Hovnanian A. Mosaicism for ATP2A2 mutations causes segmental Darier's disease. *J Invest Dermatol*. 2000;115:1144-7. [PMID:11121153].
4. Murakami S, Iizuka H, Suzuki H, Hashimoto K, Takahashi H, et al. A Japanese case of segmental Darier's disease caused by mosaicism for the ATP2A2 mutation. *Br J Dermatol*. 2003;49:185. [PMID:12890216].
5. Munro CS, Cox NH. An acantholytic dyskeratotic epidermal naevus with other features of Darier's disease on the same side of the body. *Br J Dermatol* 1992; 127: 168–171. [PMID:1390147].
6. Cambiaghi S, Brusasco A, Grimalt R, Caputo R. Acantholytic dyskeratotic epidermal naevus as a mosaic form of Darier's disease. *J Am Acad Dermatol* 1995; 32: 284–286. [PMID:7829719].
7. Barfield RL, Barrett KR, Moon CM, David-Bajar K. Pruritic linear papules on a 75 year old woman: a case of localized Darier-White disease. *Cutis* 2002;70:225-8. [PMID:12403314].
8. Ghorpade AK. Unilateral Blaschkoid Darier's disease over the forehead. *Indian J Dermatol Venereol Leprol* 2014;80:189-190. [PMID: 24685875].
9. Gilaberte M, Puig L, Vidal D, et al. Acantholytic dyskeratotic naevi following Blaschko's lines: a mosaic form of Darier's disease. *J Eur Acad Dermatol Venereol*. 2003; 17(2):196-199. [PMID:12705751].
10. Milobratovic D, Vukicevic J. Localized Darier's disease. *Acta Dermatovenereol Alp Pannonica Adriat*. 2011;20(4):213-5.[ PMID:22367378].