Case for diagnosis*

Caso para diagnóstico

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CASE REPORT

Female patient, 42 years-old, without any noticeable personal or familial history, has presented, for the last four years, keratotic papules, some crusted, in a reddish-brown coloration, linearly distributed along the left mammary region (Figure 1). The patient presents similar lesions following the Blaschko lines from the left flank to the hypogastrium and also on the pretibial area of the left leg (Figure 2). These are pruriginous lesions that are exacerbated by the heat. There are no alterations to the palmar-plantar regions, mucosae or nails.

The histologic examination of an inter-mammary papule shows hyperkeratosis with areas of parakeratosis, acanthosis and suprabasal acantholysis associated to corps ronds and grains, besides a superficial inflammatory perivascular infiltrate formed by lymphocytes (Figure 3).

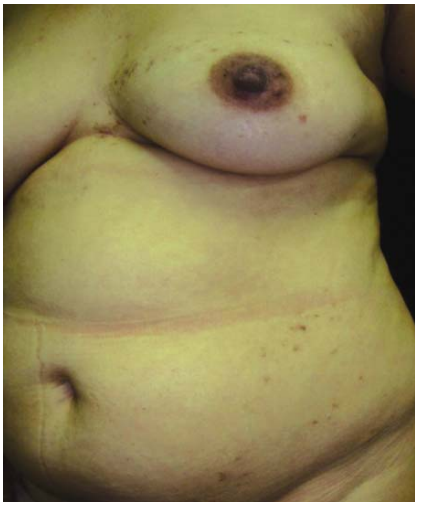

FIGURE 2: Keratotic papules, some crusted, with reddish-brown coloration, following the Blaschko lines from the left flank to the hypogastric area

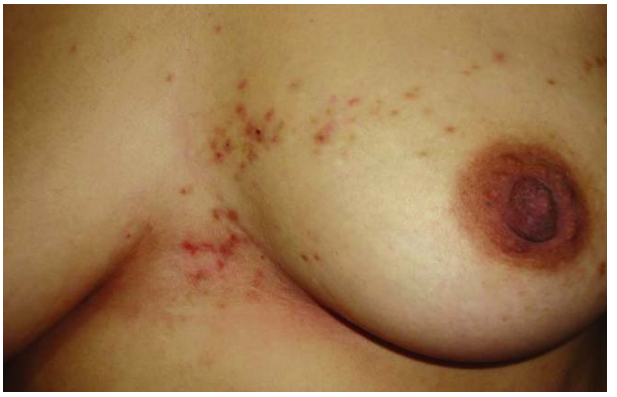

FIGURE 1: Keratotic papules, some crusted, with reddish-brown coloration, linearly distributed along the left mammary region

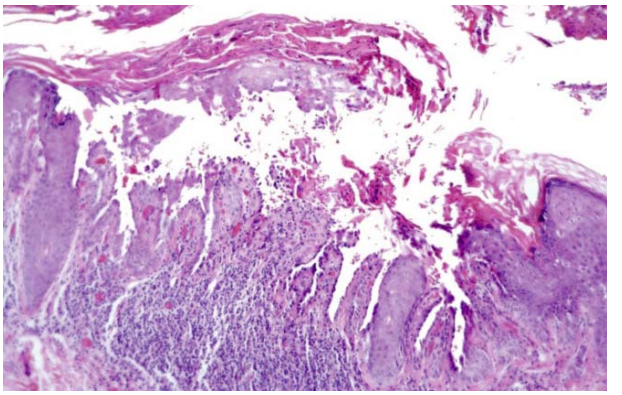

FIGURE 3: HE 100x: Hyperkeratosis with areas of parakeratosis, acanthosis and suprabasal acantholysis, associated to corps ronds and grains, besides a superficial inflammatory perivascular infiltrate formed by lymphocytes

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DISCUSSION

Linear Darier Disease type 1: Darier's Disease (DD) is an autosomal dominant genodermatosis, characterized by keratotic lesions some of which are crusted, with a reddish-brown coloration, mainly in seborrheic areas of the skin, symmetrically distributed and typically exacerbated by ultraviolet light. It usually appears during the second decade of life, but it may start later on, affecting skin and nails in a peculiar form.

Mutations on *ATP2A2* gene have been identified as the underlying cause of this disease. Such mutations are localized at the chromosome 12, region 12q2324.1, leading to a mal-function of the calcium-ATPase pump in the endoplasmatic / sarcoplasmatic reticulum, with consequent premature keratinization and loss of cellular adhesion (acantholysis) between keratocytes.[1]

Until recently, over 90 different mutations have already been reported, including deletions, insertions and substitutions.[2]

On ocasion, mitotic errors or mutations during the early phases of the developmental process give origin to two or more cell populations on the same individual, a condition referred to as mosaicism. In mosaicism, the disease presents a linear pattern, both phenotypically and histologically similar to classic Darier's, however in unilateral form, following the Blaschko lines, as described in our patient.

Two types of linear distribution were described in DD. In type 1, DD lesions follow the Blaschko lines unilaterally, without other manifestations of the disease. In type 2, these lesions represent focal areas of greater severity, superimposed to disseminated DD. The prevalence of the linear variant is unknown, but the estimation is that it may correspond to 10% of all DD cases.[3]

Ungueal manifestations in DD are often absent in the linear variant, as well as familiar antecedents of the disease.[4] The patient presented in this article has normal nails e no familiar history of DD, in agreement to the medical literature. The anatomopathological examination of the inter-mammary lesion confirmed the diagnostic hypothesis of DD, however, it did not allow for the distinction between types 1 and 2 of the disease. This differentiation is purely clinical, based upon the distribution of the lesions on the skin.

The differential diagnoses would be Grover's Disease and acantholytic dyskeratotic epidermic nevus. The epidermic nevus is already present at birth. There is much controversy in the literature if it would be, actually, a linear variant of DD.[5] Grover's Disease presents in a sudden form following a febrile illness and it is more frequent in elderly patients.

Our patient was diagnosed with type-1 linear DD, because she had characteristic DD lesions following Blaschko lines, without other signs or symptoms. Topic retinoic acid is a valid therapeutic option in such cases, but the results are often mixed. ❑

Abstract: Linear Darier is a rare variant of Darier's Disease. The keratotic papules follow Blaschko's lines, characterizing the cutaneous mosaicism. We report the case of a 42-year-old woman who presented with small, linearly distributed, reddish-brown papules on the left mammary region, from the left flank to the hypogastrium and on the pretibial area of the left leg. Histologically, suprabasal acantholysis and dyskeratotic cells were seen, confirming the diagnosis.
Keywords: Darier disease; Mosaicism; Skin diseases, genetic

Resumo: Darier linear é uma variante rara da Doença de Darier. As pápulas ceratósicas se distribuem seguindo as linhas de Blaschko, caracterizando o mosaicismo do envolvimento cutâneo. Relatamos o caso de uma mulher de 42 anos que apresentava pápulas acastanhadas, distribuídas linearmente na região mamária esquerda, do flanco esquerdo ao hipogástrio e na região prétibial esquerda. Histologicamente, foram observadas acantólise suprabasal e células disceratósicas, confirmando a suposição diagnóstica.
Palavras-chave: Dermatopatias genéticas; Doença de Darier; Mosaicismo

REFERENCES

1. Sakuntabhai A, Ruiz-Perez V, Carter S, Jacobsen N, Burge S, Monk S, et al. Mutations in ATP2A2, encoding a Ca2+ pump, cause Darier's disease. Nat Genet. 1999;21:271-7.
2. Wada T, Shirakata Y, Takahashi H, Murakami S, Iizuka H, Suzuki H, et al. A Japanese case of segmental Darier's disease caused by mosaicism for the ATP2A2 mutation. Br J Dermatol. 2003;49:185-8.
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.
4. Sanderson EA, Killoran CE, Anita PL, Wilkel CS. Localized Darier's disease in a Blaschkoid distribution: Two cases of phenotypic mosaicism and a review of mosaic Darier's disease. J Dermatol. 2007;34:761-4.
5. Tarle RG, Tarle SF, Neto JF, Shibue JRT, Mello SM. Doença de Darier unilateral ou nevo epidérmico disceratósico acantolítico? A propósito de um caso. An Bras Dermatol 1997;72:37-9.

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