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Letter to the Editor

Epidermolytic keratinocytic nevus as an expression of cutaneous mosaicism

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Dear editor,

Ichthyoses are genetic disorders of keratinization and are divided into three large groups: non-epidermolytic ichthyoses, ichthyosiform dermatitis and epidermolytic ichthyosis. The latter is an autosomal dominant disease with complete penetrance and may be familial or sporadic (1, 2). In epidermolytic epidermal nevus, the same mutations as in epidermolytic ichthyosis are found, but in a pattern of cutaneous mosaicism (3).

We received a 6-year-old girl in our outpatient clinic with a history of linear hypochromic spot on the left upper limb noticed in the first year of life. The mother reported the appearance of other similar linear spots on the left dorsal, lumbar and sacral region in the last 2 years and an area of marked hyperkeratosis in the region of the cubital fossa in the last few months. She was born vaginally, without complications and the family history was negative for similar lesions. On examination, he presented hypochromic linear lesions, with a mildly warty appearance (following Blaschko's lines) on the left upper limb, shoulder, scapular and sacral region, all ipsilateral (Figures 1 and 2).

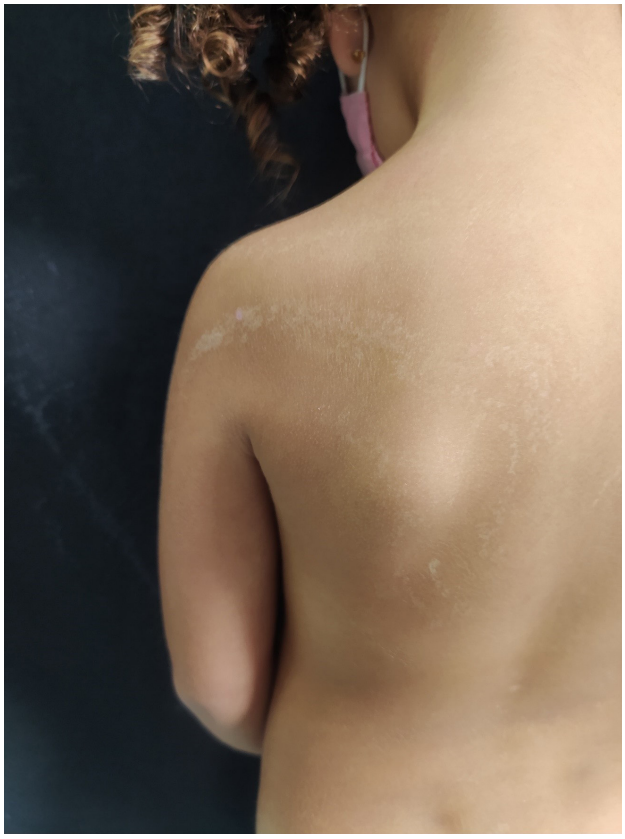

Fig. 1. Linear lesions with a discreetly warty appearance (following Blaschko's lines) on the shoulder and scapular region.

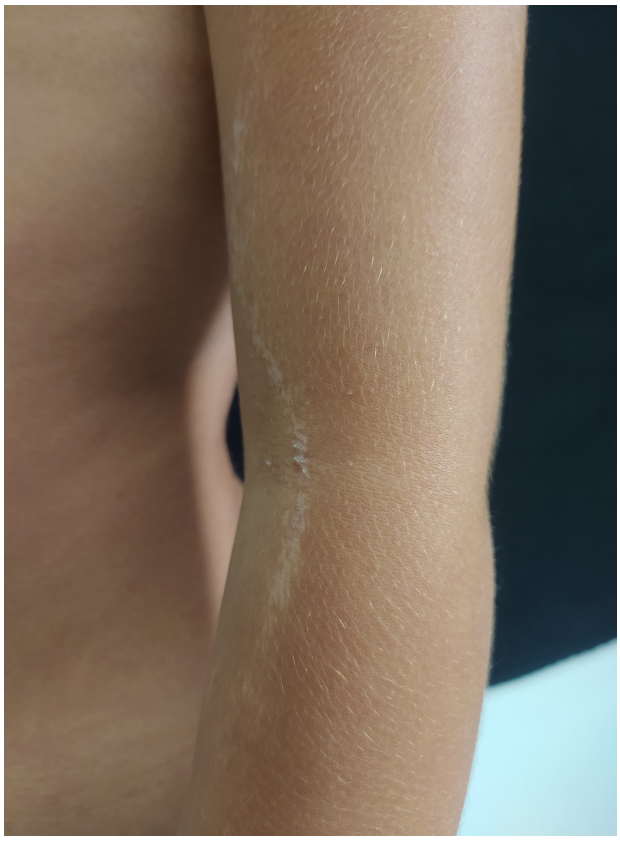

Fig. 2. Hypochromic and hyperkeratosis linear lesion in the left cubital fossa.

The pathological examination demonstrated skin with hyperkeratosis, papillomatosis, acanthosis, associated with intraepidermal vacuolation with keratohyaline granules. The histopathological findings, associated with clinical information, confirmed epidermolytic hyperkeratosis in an epidermal nevus (Figures 3 and 4).

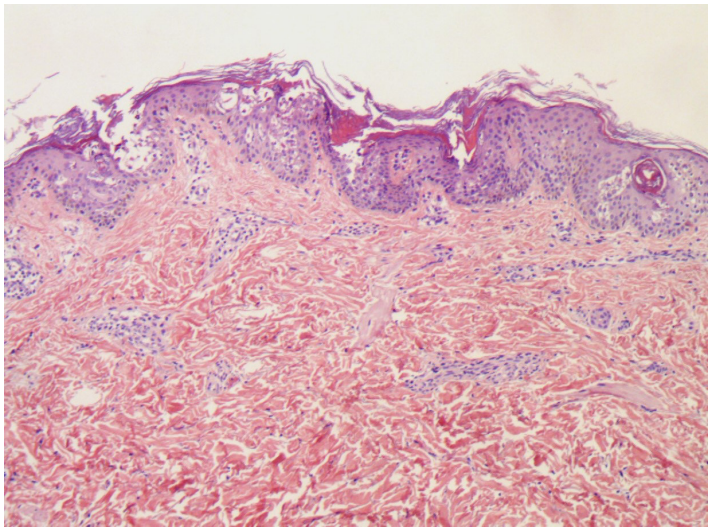

Fig. 3. Skin with hyperkeratosis, papillomatosis and acanthosis. H-E 200x.

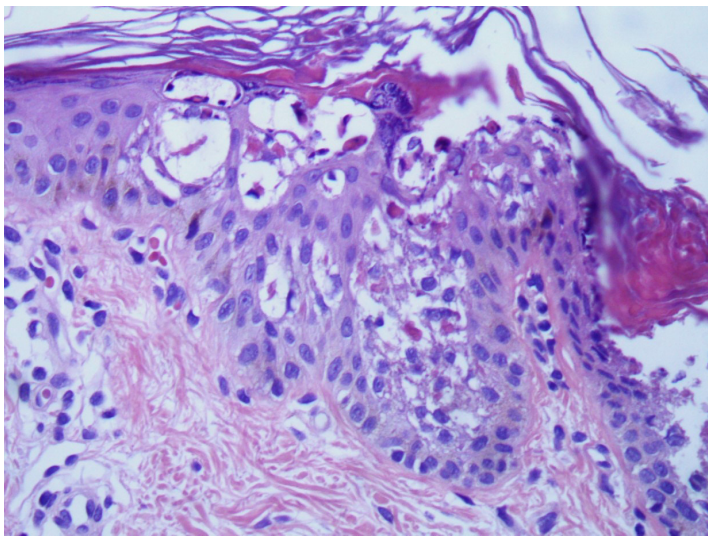

Fig. 4. Intraepidermal vacuolation with keratohyaline granules. H-E 400x.

Topical treatment with 15% urea cream was prescribed, as well as outpatient follow-up and referral to medical genetics. Epidermolytic ichthyosis (OMIM #113800) occurs as a consequence of mutations in the keratin 1 and 10 genes (KRT1 and KRT10) (1), located on chromosomes 17q12-21 and 12q11-13, respectively (3). "De novo" mutations occur in half of the cases (2). The prevalence varies from 1 in every 100,000 to 1:400,000 children (4) with equal affects of both sexes (3). It is possible to clinically classify epidermolytic ichthyosis according to the site involved as plantar/palmar or non-plantar/palmar (5, 2), also according to the extent of the skin involved as linear/localized (nevoid) (3) or presenting as the generalized form (4).

Epidermolytic keratinocytic nevus corresponds to epidermolytic ichthyosis in a cutaneous mosaic pattern. It appears as warty lesions, following a linear path. It may be present at birth and often follows Blaschko's lines. It can appear in any body segment, but it commonly affects the folds. The foul odor that appears in the maceration areas is characteristic of this disease (5).

There are several published reports of families of parents affected with the nevoid form and children with the complete form of epidermolytic ichthyosis (6, 7, 8). Therefore, we can infer that the mutations involved in epidermolytic keratinycytic nevus are usually somatic and rarely germline.

From a histopathological point of view, the findings are the same as those of epidermolytic ichthyosis: Vacuolar degeneration in the upper portion of the epidermis, basal layer that can be normal or hyperproliferative, suprabasal cellular degeneration, thin granular layer and cornea, with hyperkeratosis (1, 5, 9).

It is possible to identify perinuclear vacuolation with irregular cell borders, mainly in the spinous and granular layers. In electron microscopy, it is possible to identify the agglomeration of tonofilaments and their perinuclear layer in suprabasal cells.

The histopathological study is essential to exclude differential diagnoses such as linear lichen striatum, incontinentia pigmenti, sebaceous epidermal nevus and keratinocytic epidermal nevus. The last two may be associated with systemic changes comprising epidermal nevus syndrome (Schimmelpenning Syndrome) and keratotic pigment phakomatosis, which are associated with skeletal, neurological and ophthalmological changes.

In addition to these, papular epidermal nevus with "skyline" basal cells (PENS) can also be associated with extracutaneous findings (PENS Syndrome) such as neurological symptoms in 50% of cases. It is important to perform a biopsy for the correct diagnosis, multidisciplinary evaluation and referral to a geneticist.

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DISCLOSURE

All authors report no conflict of interest.

INFORMED CONSENT

Informed consent was obtained from the parents of the children included in this case report.

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