Mosaic epidermolytic ichthyosis

Sónia Raquel Mendes, Ana Rita Gameiro, José Carlos Cardoso, José Pedro Reis

Dermatology Department,
Coimbra University Hospital,
Coimbra, Portugal

Correspondence to
Dr Sónia Raquel Mendes;
soniaraquel23@gmail.com

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DESCRIPTION

We report the case of a 51-year-old female patient observed for papillomatous, pigmented, confluent and slightly reticulated lesions, some of which were linear, located mainly in the armpits and extending to the trunk and cervical region and present since birth (figure 1). The lesions seasonally changed to hyperkeratotic and verrucous in the winter. She also had hyperkeratotic lesions in a linear pattern in the palms.

She reported no previous diseases, use of medication, previous hospitalisations, surgeries, drug allergies or similar diseases in the family.

Histopathological examination revealed an epidermis with extensive foci of hyperkeratosis, under which there were hypergranulosis and vacuolar degeneration of the granular layer and the upper layers of the stratum spinosum (figure 2). The pattern described confirmed the clinical hypothesis of mosaic epidermolytic ichthyosis (EI).

The patient started treatment with acitretin 25 mg per day, later reduced to 10 mg per day, with good response.

The genetic study of keratins 1 and 10 in skin biopsy tissue and in peripheral blood revealed the mutation in the keratin 1 gene variant c.982A>T (p.Thr328Ser) in heterozygosity.

Figure 2 Hyperkeratosis, reticular degeneration of the keratinocytes of the upper half of the epidermis with coarse kerato-hyaline granules and eosinophilic inclusions (H&E 100x).

EI is a rare autosomal dominant disorder, and spontaneous mutation may occur in about 50% of all cases.^{1 2} Mutations have been reported in the keratin 1 and 10 genes; when associated with keratin 1 mutations, it may be related to palmoplantar keratoderma.^{1 2} The recognition of this disease becomes important considering its differential diagnoses.¹⁻³ Once the disease is diagnosed, affected parents can receive genetic counselling; information about the expected course of the disease and more severe forms of inherited ichthyosis can be excluded.²

Learning points

- ▶ The first manifestations of the disease occur at birth, in the form of multiple erosions that subsequently originate hyperkeratotic lesions.
- ▶ The histopathology is typical, standing out the marked hyperkeratosis, with reticular degeneration of keratinocytes in the upper layers of the epidermis.
- ▶ The treatment of epidermolytic ichthyosis, as well as that of other forms of ichthyosis, includes the use of emollients, keratolytic agents, topical retinoids, topical vitamin D analogues and oral retinoids.

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Competing interests None declared.

Figure 1 Papillomatous, pigmented, confluent lesions, some of which were linear, which evolved to hyperkeratotic and verrucous lesions.

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