GENETICS AND GENODERMATOSES

EPIDERMOLYSIS BULLOSA SIMPLEX WITH MOTTLED PIGMENTATION: REPORT OF A SPORADIC CASE

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Background: Epidermolysis bullosa simplex with mottled pigmentation (EBS-MP) is an uncommon subtype of Epidermolysis bullosa simplex (EBS) caused by a mutation in the KRT5 gene. Clinically it is characterized by non-cicatricial blisters mainly at the distal extremities and progressive mottled hyperpigmentation, that often disappears in adulthood. There is also palmar and plantar hyperkeratosis. Small acral verrucous papules, onychodystrophy and mild involvement of the oral mucosa can be observed during childhood. EBS-MP diagnosis is based on typical clinical findings, family history, imunomapping and/or electron microscopy, as well as molecular/ mutation analysis when possible. The differential diagnosis includes Naegeli-Franceschetti-Jadassohn (NFJ) ectodermal dysplasia, other forms of dyschromia, Dowling-Degos disease, and even atypical cases of Darier's disease with mutations in ATP2A2.

Observation: Two-year-old girl, phototype III, with a history of blistering skin since birth. On dermatological examination she had desiccated blisters on the feet and hyper and hypochromic macules scattered over the tegument with mottled appearance. Normochromic papules on the dorsal region of the fingers and onychodystrophy were also observed. The blisters appeared spontaneously or after minimal trauma, according to the mother's report and were located mainly at the distal extremities. At two months of age, the hyper and hypochromic macules began. The mother also referred oral mucositis. A skin biopsy was carried out and the material was sent for immunomapping, that resulted EBS (deposition of all antigenic markers on the blister floor – dermal side). Considering the patient's clinical and laboratorial findings, the diagnosis of EBS-MP was concluded, possible a sporadic case, by the absence of a family history for EB or other bullous diseases.

Key message: The classic clinical features of EBS-MP, its benign evolution (improvement in adulthood), its main differential diagnoses and the importance of immunomapping, a simple and easily executed tool that corroborated the diagnostic conclusion of this case.