

EPIDERMOLYSIS BULLOSA SIMPLEX OGNA – CASE REPORT

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Summary

A 27 year old woman with no family history of epidermolysis, presents trauma-induced acralhemorrhagic blisters, palmoplantar keratoderma, posttraumatic violaceous macules and erosions on the lower legs. Based on clinical findings, histopathology and immunofluorescence, we conclude that the diagnosis is Epidermolysis bullosa simplex Ogna. The treatment is based on reducing provoking factors and prevention of infection. The evolution is benign.

Key words: *Epidermolysis bullosa simplex Ogna, hemorrhagic blisters, plectin.*

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Rezumat

O femeie în vârstă de 27 ani, cu nici un istoric familial de epidermoliză, prezintă vezicule induse de traumatisme acralhemorrhagice, palmoplantar keratoderma, macule violacee posttraumatice și eroziuni pe picioare mai mici. Pe baza constatărilor clinice, histopatologice și imuno-fluorescența, putem concluziona că diagnosticul este Epidermolysis-bullosa simplex Ogna. Tratatamentul se bazează pe reducerea factorilor declanșatori și prevenirea infecției. Evoluția este benignă.

Cuvinte cheie: *Epidermolysis bullosa simplex Ogna, vezicule hemoragice, plectina.*

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1. Introduction

Epidermolysis Bullosa comprises a group of genetically blistering diseases, genodermatoses, present at birth or appear during infancy or later in the second or third decade. The common clinical findings are the blisters on skin or mucosa, which can arise spontaneously but more often it is associated with minor trauma; for this reason, this is also known as the mechanobullos disorder. This abnormal fragility of the skin is determined by the ultrastructural level of cleavage in the skin: within the epidermis, at the dermoepidermal junction or in the upper dermis and determines the classification of this complex and heterogeneous group into simplex, junctional and dystrophic subtypes [4]. The

symptoms and prognostic are variable, but the slow healing and secondary skin infections are common for all groups. In the case of epidermolysis bullosa simplex (EBS) subtype, at the electron microscopy level, is characterized by intradermal blister formation, the split appears due to the collapse of the cytoskeleton or due to its dissociation from hemidesmosomes and to cytolysis of the basal keratinocytes, mainly caused by dominant-negative mutations in the genes encoding keratins 5 and 14, but also desmoplakin, plakophilin-1, plectin or alpha6 beta4-integrin [11]. A rare disorder in this subtype is Epidermolysis bullosa simplex Ogna form, with only few references in the speciality literature [6].

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a.



b.

Fig. 1. a., b. Hemorrhagic blisters of the soles

2. Clinical case:

We present the case of a Caucasian female patient, aged 27, with no family history of epidermolysis, who is registered in the National Program for Epidermolysis Bullosa. The disease starts in the first year of her life with skin fragility, she tends to have trauma-induced acral blisters, often hemorrhagic, which worsen in warm weather. In time, blistering diminished, mostly of the palms and a mild focal palmoplantarkeratoderma appears. Also, with increasing age, posttraumatic violaceous macules on the extremity as well as spotty hypopigmented macules on the back appear. Every visit confirm the hemorrhagic aspect of the blisters. At the last visit, she presents only few hemorrhagic blisters on the soles (fig.1), violaceous macules and erosions on the lower legs.

Cardiological and neurological examination excluded cardiomyopathy and muscular dystrophy. Histopathology reveals intraepidermal microblisters, electron microscopy shows a split just above the hemidesmosomal plaque. Indirect immunofluorescence was performed three years ago and based on this exam, the hemorrhagic aspect of the blisters and no other symptoms: muscular or any other kind, we conclude that the diagnosis is Epidermolysis bullosa simplex Oagna. The treatment is based on reducing the provoking factors: trauma and prevention of infection, thus the evolution is benign.

3. Discussion:

This rare disorder was first described in Norway and named after the village where the first affected family originated [6]. The disease is inherited in an autosomal dominant pattern of the mutation in PLEC1 gene on chromosome 8q24, encoding plectin, a hemidesmosomal protein which is distributed among a variety of tissues, including skin, nerve and muscle [1], [2], [9]. In contrast to the severe autosomal recessive transmission forms of this mutation, associated with muscular dystrophy or pyloric atresia [12], the clinical finding of EBS Oagna are only cutaneous and has a good prognosis. In the few cases reported in literature, the clinical findings of the patients are very similar to our patient symptoms: the mild skin fragility, manifesting with acral blisters, occasionally elsewhere, that are often hemorrhagic and erosions which heal with violaceous macules [8], [9]. The careful clinical examination and microscopic analysis of a skin biopsy, seem to be the important features that distinguish this rare subtype from the frequent keratin-associated, the mildest and most common type Weber-Cockayne EBS, which is also characterised by blisters of palms and soles, but without the hemorrhagic aspect [5], [11]. Also, in contrast with another keratin-associated EBS subtype: Dowling-Meara, with more severe course of disease, in EBS Oagna we don't find in addition to palmoplantar distribution, extensive herpetiform blistering, oral mucosal involve-

ment, milia or loss of nails [5], [7]. The Kobner type is associated with milder generalized blistering and here is not the case [11]. Hematoxylin and eosin staining revealed intra-epidermal microblisters, and electron microscopy and immunofluorescence provide additionally useful diagnostic information: reduction and interrupted staining for the hemidesmosomal components plectin [3], [8], [10]. The specific plectin antibodies: 5B3, 10F6, 31/Plectin, GP21 [9], [13], was not available for our case.

4. Conclusions:

EBS Oagna is a rare form in the EBS group, which is characterized by acral blisters, often hemorrhagic. Diagnosis is confirmed by electron microscopy: the split is intraepidermal, and immunofluorescence: reduction or absent staining for plectin. The clinical findings of our patient are very similar to those described in the publications mentioned in this article and electron microscopy and immunofluorescence confirm diagnosis for our case.

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Conflict de interese
NEDECLARATE

Conflict of interest
NONE DECLARED

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