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Case Report .

Generalized Epidermolysis Bullosa Simplex, Dowling Meara in Saudi Family, Case Report and Review of Literature, Saudi Arabia 2017-2018

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ABSTRACT

A 4 years old Saudi girl, her neonatal period was characterized, since first 2 days of life by nails dysplasia and yellow pigmentation then progress to well demarcated, Serous and hemorrhagic, vary in size blisters. That improved gradually with age. The blisters started from the nails which lead to nail shedding, extremities then became generalized. Dermatological examination revealed multiple herpetiform hemorrhagic blisters and erosive lesions, slight scars and milia mainly on palm and sole. There is a history of similar skin lesion in his father. Confirmation of genetic mutation in KRT14 gene.

Key words: Generalized Epidermolysis Bullosa Simplex, Dowling Meara Saudi Arabia.

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INTRODUCTION

Epidermolysis bullosa (EB) is a grouping of rare genetic conditions in which bullous affecting primarily the skin arise after exposure to mechanical trauma.[1] Four major types of epidermolysis bullosa (EB) are recognized on the basis of the ultrastructural level of skin cleavage: simplex (intraepidermal); junctional (intra-lamina lucida); dystrophic (sub-lamina densa); and Kindler syndrome (intraepidermal, intra-lamina lucida, and sub-lamina densa).[1] Epidermolysis bullosa simplex (EBS) is the most common type of EB, accounting for 75 to 85 percent of all cases of EB in Western countries.[2] EBS is almost always inherited in an autosomal dominant fashion, but rare autosomal recessive forms have been reported.[2]

CASE REPORT

A 4 years old Saudi girl, was born full term via uncomplicated vaginal delivery. Her neonatal period was characterized, since the second day of her life by nails dysplasia and yellow pigmentation then progress to well demarcated, Serous and hemorrhagic, vary in size blisters (Fig.1), That improved gradually with age. The blisters started from the nails which lead to nail shedding, extremities then became generalized, associated with pruritus, the blister exacerbate with hot weather and minor trauma. The blister also appeared spontaneously (Fig.2-3). They ruptured and formed superficial erosion, then healed with recurrence lesion. There is a history of similar skin lesion in his father.

A clinical examination: showed normal somatic and visceral status. Dermatological examination revealed multiple herpetiform were hemorrhagic blisters and erosive lesions, slight scars and milia mainly on palm and sole. No evidence of infection, no draining wound. No active bleeding. There was also a marked nail dystrophy on the fingers and toenails.

Genetics Testing For Epidermolysis Bullosa

Epidermolysis bullosa is an inherited disorder associated with skin fragility and blistering among other symptoms. The type and severity of the symptoms depends on the specific genetic cause of the disease.

RESULT

Heterozygous for (one copy of) the variant c.374G>A (p.Arg125His) in the KRT14 gene.

This child was found to carry a single copy of the variant called p. arg125 His in her KRT 14 gene. This common variant that has been reported in multiple patient with the autosomal dominant generalized severe form of epidermolysis bullosa simplex.

DISCUSSION

EBS is characterized by trauma- or friction-induced skin blistering with localized or disseminated anatomic distribution. The most common subtypes of EBS are:

- EBS, localized (formerly known as EBS Weber-Cockayne)
- EBS, generalized severe (formerly known as EBS Dowling-Meara)
- EBS, generalized intermediate (includes the type formerly known as EBS Koebner).

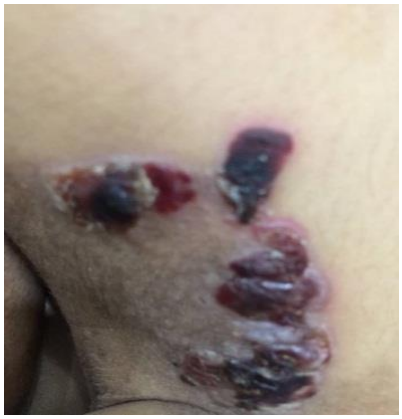


Fig 1: Hemorrhagic herpetiform blisters in trunk.

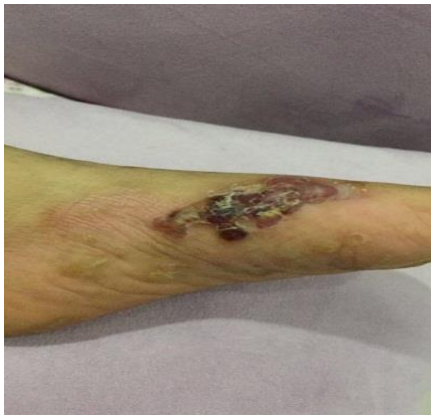


Fig 2: Hemorrhagic herpetiform blisters in foot.

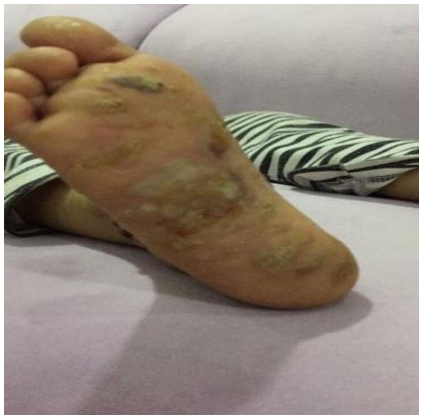
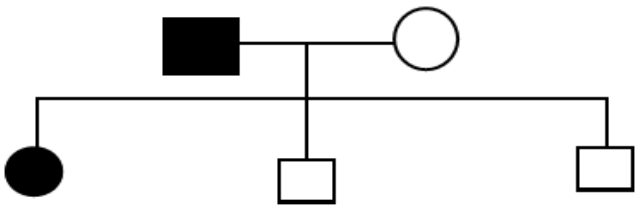


Fig 3: Multiple herpetiform blisters and erosive lesions, slight scars in the sole.



In most cases, EBS is caused by dominant negative missense mutations in KRT5 and KRT14 genes encoding keratins that are mainly expressed in the basal keratinocytes.[3]

Rarely, EBS may be caused by autosomal recessive inheritance of either one of these genes (more often KRT14).[3] EBS, generalized severe — generalized severe EBS, formerly known as Dowling-Meara EBS, is the most severe form of EBS. It presents at birth with disseminated trauma or friction-induced blistering. Grouped blisters with an arcuate, "herpetiform" arrangement may appear spontaneously on the trunk, upper limbs, or neck. Involvement of the oral mucosa is common. Hyperkeratosis of the palms and soles appears during infancy and can progress over time to confluent keratoderma. Additional clinical features include nail dystrophy, nail shedding, and hair loss (telogen effluvium).[4]

EBS-DM is the most severe form of EBS and can be life threatening in neonates. Improvement usually occurs during middle to late childhood, just lake in our case. However, some adults remain substantially disabled by Dowling-Meara EBS throughout their lives as a result of persisting blistering of the hands and feet and palmar and plantar keratoderma.[5]

The management of patients with EB involves a multidisciplinary team usually composed of a dermatologist, an EB nurse, primary care provider, occupational therapist, nutritionist, and social worker. Specialists, including gastroenterology, ophthalmology, nephrology, hematology, endocrinology, cardiology, pain management, plastic surgery, and specialized dentistry, are consulted as needed. This multidisciplinary approach is emphasized by the 2014 multicenter consensus recommendations for skin care in inherited epidermolysis bullosa.[6] The differential diagnosis includes also incontinentia pigmenti, bullous ichthyosiform erythrodermia, staphylococcal scalded skin syndrome, neonatal or congenital varicella, neonatal pemphigus, pemphigoid, aplasia cutis, pachyonychia congenita. Microscopically The Dowling-Meara variant of EBS is characterized by cytolysis of the basal cells. Ultrastructurally it is characterized by clumping of the keratin tonofilaments within areas of incipient blistering.[7]

No treatment of any of the groups of EBS is available so far. It is only symptomatic and the primary aim is to protect the skin and stop blister formation. Fresh blisters should be drained after puncturing them with a sterile disposable needle and then dressed with nonadherent dressings. Topical antiseptic and antimicrobial agents should be used to protect from secondary bacterial infection. It is very important to avoid the use of tight dressings and footwear, avoidance of high enviromental temperatures, nasogastric feeding in the cases of severely affected with Dowling-Meara neonates.[8]

CONCLUSION

EBS is one of the most well understood diseases of the EB category, but nevertheless despite knowing its etiology and pathogenesis we still lack the therapeutic options to cure the disease. The disease may be lethal if the lesions are widespread as the infection can spread to internal organs. Genetic counselling is the sole method to prevent the disease. The standard care for EB simplex is generally supportive and preventive, including avoidance of trauma, nutritional support and infection control.

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