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Journal of Dermatology and Dermatitis
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Case Report

Hereditary epidermolysis bullosa: New description

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Received date: March 16, 2020; Accepted date: May 27, 2020; Published date: June 15, 2020

Citation: Kaoutar Laamari, Hanane Baybay, Samia Mrabat, Zakia Douhi, Sara Elloudi, Fatima Zahra Mernissi (2020). Hereditary epidermolysis bullosa: New description. Journal of Dermatology and Dermatitis.5 (1); Doi:10.31579/2578-8949/66

Abstract :

Epidermolysis bullosa (EB) is a heterogeneous group of genetically determined, mechano-bullous disorders characterized by blister formation in response to mechanical trauma. The blistering of the skin occurs in the varying degrees of severity and can severely incapacitate the life of the afflicted patient. Epidermolysis Bullosa Simplex (EBS), the most commonly occurring type, is dominantly inherited where treatment still remains a major challenge.

Keyword: epidermolysis bullosa; mechano-bullous disorders' mechanical trauma; epidermolysis bullosa simplex

Introduction:

Epidermolysis bullosa (EB), often referred to as the butterfly disease, is a group of rare genetic conditions characterized by skin that is delicate and fragile as butterfly wings. The skin blisters in response to friction, minor injury, or trauma. In certain types of EB, other organs, such as the esophagus, can also be affected, and secondary complications may require multiple interventions. While there has been significant progress in classifying the disease – identifying genes and proteins involved – there have been few advances in the treatment of the disease. We report here the case of a 4 mouths infant Epidermolysis bullosa.

Case report:

It was a 4 month old infant, who consulted in the emergency room for bullous lesions that appeared one week of life.The dermatological examination objectified the presence of multiple blisters sitting at the level of the pressure zones in particular the neck, the seat, and the feet and at the periungual level surmounted by place with mellic crusts. In view of this clinical aspect, a skin biopsy was carried out which confirmed the diagnosis of hereditary dystrophic bullous epidermolysis.The pathology was explained to the family, while specifying the measures to be taken.

Discussion:

Epidermolysis bullosa (EB) is a severe inherited skin disease with separation of the dermal epidermal component of the basal membrane [1]. There are four subgroups: Kindler syndrome, epidermolysis bullosa simplex (EBS), dystrophic epidermolysis bullosa and junctional epidermolysis bullosa. [2]The pathogenesis of these disorders is unknown. Bullae formation has to do with various basic defects including structural and biomechanical abnormalities of keratin, hemidesmosomes, anchoring fibrils, anchoring filaments, and altered skin collagenase. [3]Dystrophic epidermolysis bullosa of Hallopeau Siemens is the most severe form. It is generalized and transmitted genetically as a recessive. Diagnosis is often made of birth with the blisters leaving diffuse skin erosions. [6] Risk of death is significant. Moreover, it is often associated with severe mucosal involvement and skin appendage conditions. Clinical course is marked by complications with severe oesophageal stenosis, conjunctival involvement and the possibility of squamous cell carcinoma arising in the cutaneous lesions. [4,5]

There is presently no definitive cure for EB. The objective of treatment is to alleviate symptoms and provide supportive measures. Therapy is therefore focused on the prevention of lesions and complications which requires multidisciplinary approach involving pediatrician, dermatologist, surgeon, nutritionist, dentist, physiotherapist, nurse, psychologist, pain 18 specialist, and geneticist. Psychological support for parents and family members is vital. EB is not a contraindication for any vaccination. [6,7].

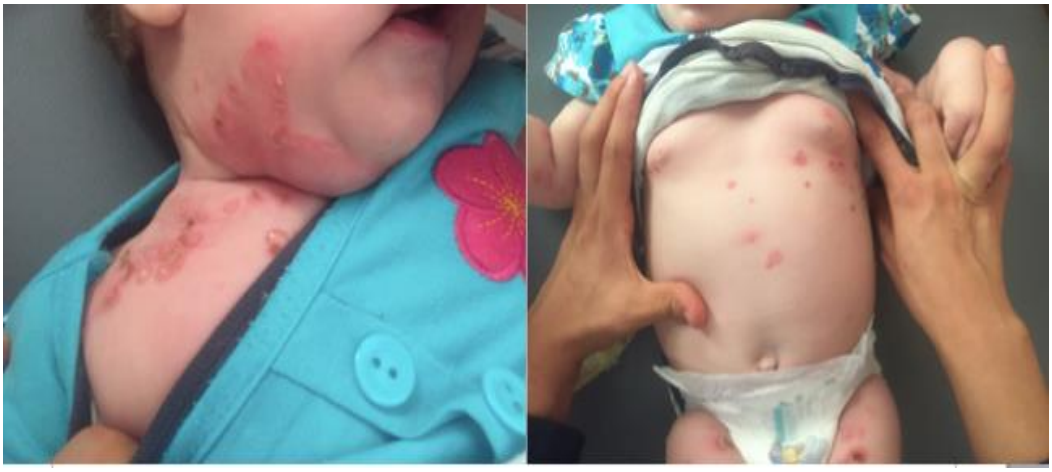

Figure 1: multiple blisters surmounted by place with melic crusts in the neck and the trunk

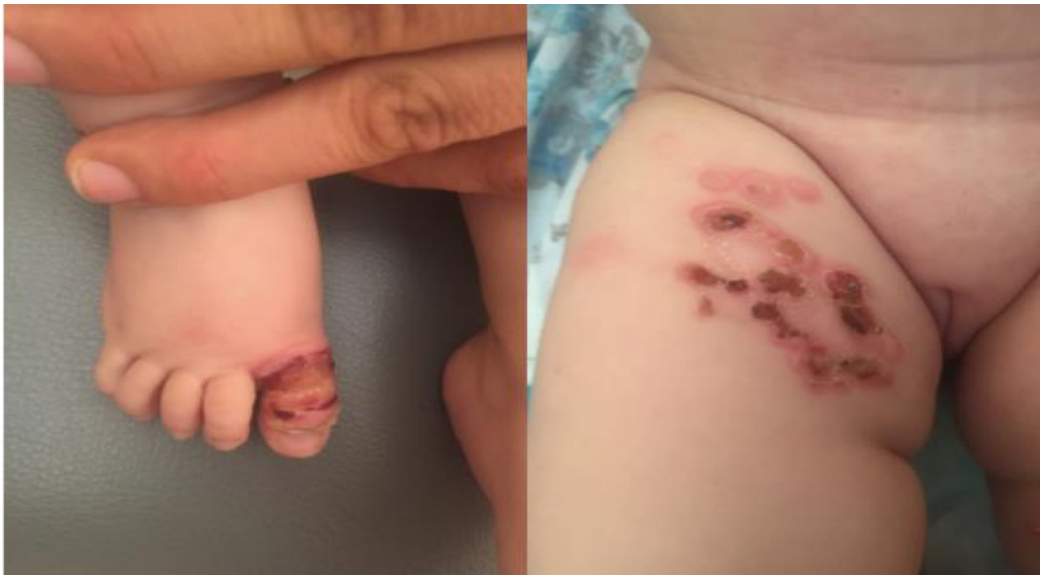

Figure 2 : multiple blisters surmounted by place with mellic crusts at the thigh and perungueale

Conclusion:

Dystrophic epidermolysis bullosa is a highly complex disease that requires appropiated diagnosis from the first signs of this disease, and also an adequate histopathological diagnosis in order to start treatment and reduce morbidity and mortality. Currently, with the advancement of technology, specific treatments are being carried out in relation to the genetic alteration and the results of the molecular studies, thus giving proper treatment for each of the sub-variants of this disease.

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