Residência RP Pediátrica
Publicação Oficial da Sociedade Brasileira de Pediatria

Submitted on: 16/08/2016
Approved on: 29/04/2017

CASE REPORT

Congenital junctional epidermolysis bullosa: A case report

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Keywords:
Epidermolysis Bullosa, Epidermolysis Bullosa, Junctional, Congenital Abnormalities.

Abstract

Objective: To describe a case of junctional epidermolysis bullosa in a newborn with clinical manifestations starting at birth and present a literature review on this subject. **Case description:** a 3-day-old female was admitted due to skin lesions with spontaneous tense bullae with citrine-yellow content in the palate, umbilical region, and buttocks. She progressed with an increase in the number of bullae and erosions all over the body, with involvement of the hands and feet, including onychodystrophy. After the clinical diagnosis of epidermolysis bullosa, a biopsy of an abdominal blister was performed, and immunomapping revealed characteristics of junctional epidermolysis bullosa. The initial therapy was topical application of essential fatty acids and education regarding the management and routine care of the patient. She was discharged with scheduled monthly follow-up visits for clinical observation. **Comments:** This case highlights the importance of the awareness about epidermolysis bullosa. Pediatricians must know how to conduct the investigative approach and how to follow-up the patient. This report can contribute to the dissemination of knowledge about this disease, enabling the identification of new cases; emphasizing the importance of a multidisciplinary approach; and aiming to anticipate, prevent, and treat the most common complications associated with this entity.

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INTRODUCTION

Hereditary epidermolysis bullosa (EB) is a disease secondary to the mutation of genes responsible for encoding proteins that enable adhesion of one skin layer to another. It is characterized by the appearance of bullae and lesions on the skin and mucous membranes, either spontaneously or in response to minimal trauma. This disease can be manifested at birth or during the first years of life.[1] There are several clinical types, depending on the depth of skin at which adhesion defects occur. The severity and extent of skin lesions and the involvement of other mucocutaneous organs vary considerably among the types of EB, which is mainly determined by the nature of gene mutations and penetrance, resulting in different phenotypic expressions.[4] There is no definitive treatment for EB, only symptomatic relief.[7]

EB is classified into four main types: EB simplex (EBS), junctional EB (JEB), dystrophic EB (DEB), and Kindler syndrome. These four types differ not only phenotypically and genotypically but also especially according to the level of dermal-epidermal junction cleavage. In EBS, cleavage occurs intraepidermally and may be basal or suprabasal, whereas in DEB, it occurs intradermally. In JEB, cleavage affects the dermal-epidermal junction, whereas in Kindler syndrome, it can occur at any level. EB classification also includes the hereditary mode of disease transmission: autosomal dominant or recessive. The recessive forms are usually associated with the most severe EB.[4]

The objective of this study is to report the case of a patient with clinical suspicion of EB from birth and a subsequent lesion biopsy confirming JEB as well as review the literature available on the conduction of the case.

CASE DESCRIPTION

A 3-day-old female patient was referred to the Juiz de Fora University Hospital (HU-UFJF), Brazil due to spontaneously appearing skin lesions and tense bullae of citrine-yellow content in the palate, periumbilical, and intergluteal regions. Her clinical course progressed with an increase in the number of bullae and erosions spread throughout the body, with involvement of the hands and feet as well as onychodystrophy (Figure 1). Neonatal history shows cesarean delivery at 39 weeks; birth weight of 3,310 g (appropriate for the gestational age); 1-minute and 5-minute Apgar scores of 8 and 9, respectively; and delivery without complications.

The bullae ruptured, resulting in superficial ulcerations and subsequently atrophic and hyperchromic lesions. No similar case had been observed in the familyAfter the diagnostic hypothesis of EB, the patient was referred to the São Paulo Clinics Hospital, where a biopsy was performed of a bulla in the abdominal region. Immunomapping showed that the lesion was compatible with junctional EB.

The initial therapy was the prescription of essential fatty acids (EFA) and guidance on the management and routine care of the patient. The patient was discharged without signs of infection within the lesions; however, vitamin D3 was prescribed to prevent hypovitaminosis D because sun exposure is contraindicated. Breastfeeding was maintained. In the follow-up visit at the age of 1 month, the lesions had worsened (Figure 2), and there were signs of secondary infection in the hands and feet. The use of a mupirocin ointment on the infected lesions was initiated, and application of EFA was maintained, improving the infectious process. The patient's family was reinstructed on basic skin hygiene and the importance of skin protection, in order to avoid trauma. Monthly follow-up visits were scheduled to monitor the clinical picture. At the age of 5 months, the patient was hospitalized with pneumonia, bronchospasm, and severe sepsis, progressing to respiratory failure and death.



Figure 1. Ulcerated lesions after bullae rupture in the thoracic region.

DISCUSSION

For the diagnosis of EB, the clinical history, physical examination, and bulla biopsy are important. Optical microscopy facilitates the differentiation of EB from other bullous diseases, such as pemphigus. Electron microscopy or direct immunofluorescence can reveal the level of cleavage of the bullae in the subepidermal region, thereby allowing the differential diagnosis of EB subtypes.[1]



Figure 2. Onychodystrophy in the left thumb and ulcerated lesion resulting from bullae rupture.

Precise diagnosis in the neonatal period can be difficult and must rely on a combination of clinical, histological, and molecular findings. The progression of the clinical picture and the lesions in the first 30 days of life may confuse physicians. The differential diagnosis of bullae formation in this age group includes friction blisters, staphylococcal pyoderma, staphylococcal scalded skin syndrome, toxic epidermal necrolysis, EB, congenital syphilis, intrauterine herpes simplex virus infection, and bullous pemphigoid.[2]

From birth, children need continuous effort from caregivers, who must completely rearrange their daily activities to fulfill this requirement.[3]

Supportive care for the treatment of wounds and early recognition of complications are essential in treating patients with EB, which is currently an incurable disease. The main types of complications are secondary bacterial infection, followed by sepsis (a frequent cause of neonatal death), defective healing, and aggressive cutaneous neoplasms (the most frequent cause of mortality from adolescence onwards).[4]

Currently, no specific therapy is available for EB. The treatment of patients with EB should be multifactorial and based on a series of principles. It must be aimed at preventing skin trauma to avoid the formation of new bullae, preventing secondary bacterial infection and providing aggressive infection treatment when it occurs, taking measures to improve wound healing, maintaining good nutrition, correcting complications, and finally, rehabilitation. The formation of bullae can be prevented by an attempt to handle the child carefully, using seamless clothes without tags; padding bony prominences; and avoiding skin friction, excess heat, and patches on the skin. Infection is prevented by changing bandages daily, applying topical antibiotics on the lesions, and draining the bullae.[2]

Dental interventions must be performed in all children with EB in order to emphasize and initiate health promotion and prevention of oral diseases as early as possible.[5]

A multidisciplinary approach is required to address problems such as esophageal strictures, contractures, diseases of the larynx, urethral stricture, conjunctival scarring, and psychosocial problems. A registered dietitian should be consulted for the nutritional need of a child with EB.[4,7]

CONFLICTS OF INTEREST

The authors declare no conflict of interest.

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