CLINICAL CASES

Bullous epidermolysis in newborn

Epidermólisis bullosa en recién nacido

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Abstract

Epidemolysis bullosa is a genetic disease. Clinical picture include blistering and ulcers after friction or trauma. Skin lesions may be cutaneous or extracutaneous. Family history, clinical findings, lab tests and microscopic examination are needed to make the diagnosis of the disease. A diagnosed case is presented in this paper, it is a 25 days female baby who was assisted at the Teaching Pediatric Hospital "Jose Luis Miranda "in Santa Clara. Clinical findings are described in the paper, as well as, comments about different types of Epidemolysis and their clinical manifestations, since this disease is not frequent in the pediatric age.

Keywords: epidemolysis, bullosa, genetics, pediatric age (source: MeSH-NLM).

Resumen

La epidermólisis bullosa es una enfermedad genética. Se manifiesta por la aparición de ampollas, úlceras posteriores a roces o traumatismos. Las lesiones pueden tener una localización cutánea y extra cutánea. Para realizar el diagnostico se debe tener presente los antecedentes heredofamiliares, la clínica, la microscopia y exámenes de laboratorio. Presentamos un caso de Epidermólisis Bullosa diagnosticado en recién nacido, femenino de 25 días en el Hospital Pediátrico Docente José Luis Miranda de Santa Clara. Se describen los hallazgos clínicos del caso. Se comenta acerca de los diferentes tipos de Epidermolisis y las formas de presentación por considerarse una entidad poco frecuente en la edad pediátrica.

Palabras clave: epidermólisis, bullosa, genética, edad pediátrica (fuente: DesCS-BIREME)

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Introduction

Epidermolysis bullosa, also known as butterfly skin, is an autoimmune disease of genetic origin that affects between 15 and 17 births per million people. It is characterized by the presence of blisters, ulcers, and wounds on the skin, especially on mucous membranes. Other extra-cutaneous locations can also be affected, with ontogenic ocular, gastrointestinal, and musculoskeletal alterations. The skin of those affected by epidermolysis bullosa is fragile, weak, extremely sensitive, and very vulnerable, as delicate as glass, as even the slightest physical contact can cause the skin to detach[1-4]. It can be congenital or acquired. The types of epidermolysis bullosa are more severe in the neonatal period and can even be fatal in the first few months. There are two ways in which the disease can be inherited: dominant inheritance, where one parent carries the dominant gene and has a 50% chance of passing it on to their offspring, and recessive inheritance, where both parents carry the disease and have a 25% chance of having a child with normal genes, a 50% chance of having a child who is a carrier of the disease, and a 25% chance of having a child who has the condition[1,2].

There are approximately thirty subtypes of Epidermolysis Bullosa, which are often grouped into three categories: Simple type, which is located in the epidermis, mainly on the hands and feet, is very painful, and tends to heal well. It is the most common and least lethal, appearing at birth. Junctional type, which is located between the epidermis and the dermis and can affect the mucous membranes. This variety appears less frequently. Dystrophic type, which is located in the dermis, is the most common and severe type and can leave sequelae, producing deformities in the upper and lower limbs. Kindler syndrome is a variety where several layers fuse, appearing in early childhood and causing photosensitivity, giving the skin a varying appearance from person to person[5-8].

There are civil associations worldwide called DEBRA (Dystrophic Epidermolysis Bullosa Research Association) that aim to support and help families and patients with Epidermolysis Bullosa. It has expanded to 32 countries around the world[8].

Case presentation

A term neonate, born to a 17-year-old primigravida with obstetric history of 1 pregnancy and no abortions, with a gestational age of 39 weeks and personal history of chronic hypertension, mild anemia, and vaginitis which were treated during pregnancy. The delivery was uneventful with an Apgar score of 8/9 and a birth weight of 3000 grams. The newborn presented with skin lesions at birth and was transferred to the José Luis Miranda Pediatric Hospital neonatology ward. The lesions were characterized by extensive and deep erythematous blistering, covering most of the body surface (65%), with skin fragility and loss of epidermis (Image: 1A, 1B, 2A, 2B, 3A), located on the face, chest, abdomen, upper and lower extremities.

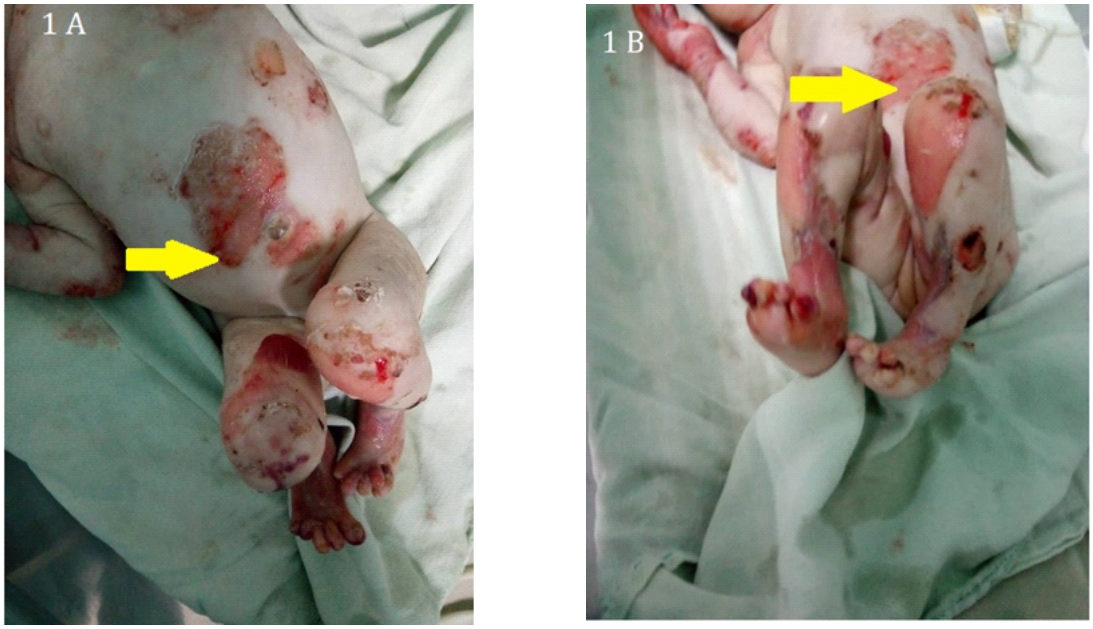

Image 1. 1A shows fragile, sensitive and painful skin with the appearance of localized burns on the abdomen and 1B shows wounds on the lower limbs when the blisters are broken.

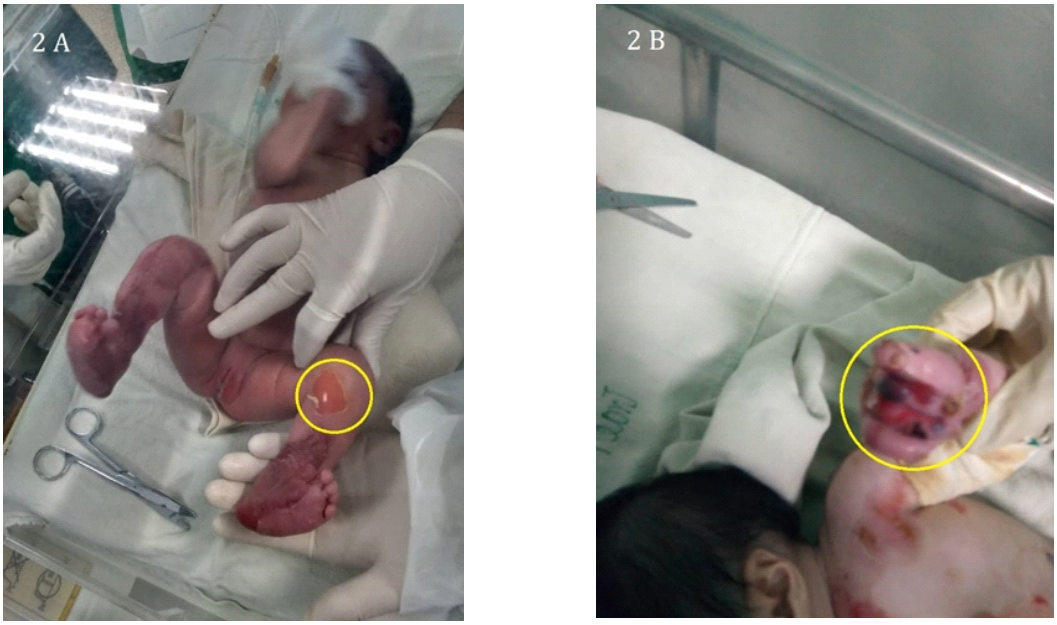

Image 2. Epidermolysis bullosa with necrotic lesions located on upper limbs 2B and blistering on lower limbs 2A

Image 3. Shows the extent of the lesions with involvement of the face, thorax, abdomen and upper and lower limbs where the use of wet dressings was necessary

According to previous evidence, a consultation with dermatology was requested and a diagnosis of Epidermolysis Bullosa and Aplasia Cutis was made. Laboratory tests were requested, from which the following results were obtained: positive blood culture for Pseudomonas Aeruginosa, positive urine culture for Escherichia Coli with high sensitivity to meropenem. The patient received antibiotic treatment for 10 days. As new lesions appeared and the patient's condition worsened, further tests were performed.

The new blood culture showed a positive result for Klebsiella, and vancomycin was added to the antimicrobial treatment. An epicutaneous catheter was placed, and the patient began to exhibit extreme bradycardia, palmoplantar cyanosis, bradypnea with thermal dissociation, and marked pallor. Endotracheal intubation and mechanical ventilation were performed, with the support of vasoactive drugs, and blood transfusions were administered simultaneously. Despite the therapeutic measures taken, the patient's clinical course was complicated, leading to death.

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Therefore, diseases such as neonatal pemphigus and gestational herpes were ruled out, especially because there were no family history of Epidermolysis Bullosa.

The ethical principles contained in the Declaration of Helsinki were complied with, and informed consent was obtained from the patient's family.

The clinical evolution of our patient from the beginning of his treatment in the diagnostic and treatment hospital is described below (Figure 1).

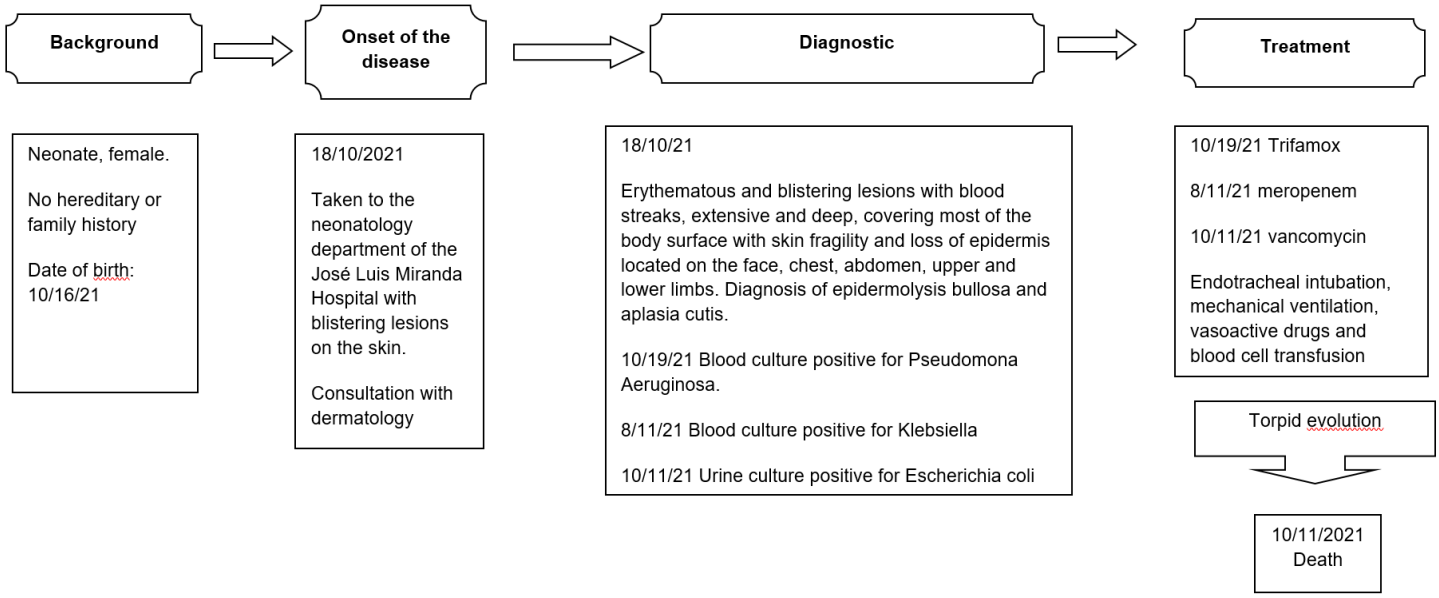

Figure 1. Clinical evolution of the patient[1]

Discussion

Epidermolysis bullosa is a genetic, hereditary, chronic, and complex condition. Its diagnosis and clinical manifestations are not well understood. It is a disease with low prevalence worldwide. Simple epidermolysis bullosa has a higher incidence in countries such as Norway, Scotland, and Northern Ireland. However, dystrophic epidermolysis bullosa is more common in Northern European countries, while junctional epidermolysis bullosa appears in two newborns per million inhabitants in the United States[1,2]. In Costa Rica, the DEBRA Latin American association was created, represented by several countries such as Mexico, Chile, Argentina, and Brazil. The DEBRA UK association is one of the best organizations focused on research, support, and care for patients with epidermolysis bullosa. According to statistical data until 2019, it is estimated that 200 people with epidermolysis bullosa live in Cuba[3-6].

To diagnose this disease in newborns, family history should be taken into account to determine the type of autosomal dominant or autosomal recessive inheritance. To rule out other pathologies, clinical methods, including a detailed medical history, physical examination, lesion description, and histopathological techniques should be used. To reach a comprehensive diagnosis in a patient with epidermolysis bullosa, the onion skin scheme should be used, determining the major type of epidermolysis bullosa, phenotype, transmission mode, structural separation site, associated findings, involved protein, gene and mutation type, and specific mutation. Acquired epidermolysis bullosa is rare in children[7].

The cutaneous findings (blisters) in different types of epidermolysis bullosa occur due to skin fragility. The severity of the disease varies with clinical manifestations, ranging from the appearance of some blisters affecting the hands and feet (Images: 1A,1B, 2A,2B, and 3) to death. The presence of blisters and abnormal scarring on the skin and mucous membranes contributes to worsening the disease. The most frequent extra-cutaneous manifestation is the presence of lesions in the oral mucosa. Secondary infection by Staphylococcus aureus can lead the patient to a fulminating septicemia[9,10].

In addition to this, it is necessary to highlight the complications that arise from the disease when lesions become generalized as shown in Figure 1, causing dehydration, malnutrition, and consequently, the death of the baby[11]. Studies by other authors showed that malnutrition prevails, along with various degrees of musculoskeletal and mucosal conditions[11,12].

Our patient with epidermolysis bullosa presented blisters with scabs (Figure 2A and 2B); however, the consulted literature also reports skin lesions such as milia cysts, scars, and pigmentation disorders[12].

Parents should be trained in the care and protection of extremely vulnerable skin and maintain hygienic measures. In the case we are presenting, these aspects were not met due to the early hospitalization of the patient and the unfavorable evolution of

the disease.

Conclusion

Although it is a rare and uncommon disease, epidermolysis bullosa should be known to enable early diagnosis and timely initiation of more effective therapy to avoid complications. The main focus should be to avoid mechanical trauma and cure lesions to prevent their growth and spread.

Authors' contributions

1. conceived the idea for the manuscript and analysis of the study: Dana Molina Piedra
2. Wrote the first draft of the article: Dana Molina Piedra
3. Methodology and data collection: Dana Molina Piedra, Yailin Perez Diaz y Yunier Cruz Rodriguez
4. Performed critical editing of the article: Dana Molina Piedra, Yailin Perez Diaz
5. Obtained photographs: Dana Molina Piedra y Yunier Cruz Rodriguez
6. Accepted the final content of the article and approval of the final version for publication: Dana Molina Piedra, Yailin Perez Diaz y Yunier Cruz

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