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CASES FOR DIAGNOSIS

Misdiagnosis in Epidermolysis Bullosa: Yet Another Burden on Patients and their Families

Error diagnóstico en un caso de epidermólisis bullosa: una carga más para los pacientes y sus familiares

Case Presentation

A 39-week-old male infant developed several blebs on his thumbs within a few hours of birth, which were classified as ''suction blisters'' at his local hospital. In the following days, several extensive blisters appeared on his chest and arms, presenting positive Nikolski's sign. He was then diagnosed as ''Staphylococcal scalded skin syndrome'', so nasal and conjunctival exudate samples were taken for culture and empirical treatment with intravenous cloxacillin was started. No microorganisms were identified on culture, so antibiotic treatment was discontinued at one week of life, and a diagnosis of ''neonatal bullous pemphigoid'' was done. Blisters remained apparently stable, with no purulent or haemorrhagic secretions.

However, at 20-days of life, the baby started with fever and exudative blistering lesions. Due to the lack of response to treatment and to worsening of the skin lesions, he was referred to our institution at 44-days of life. Upon arrival, the baby presented extensive haemorrhagic lesions which involved 60% total body surface area, affecting all 4 extremities, head and back (Figs. 1 and 2). He presented painful erosions of the pharyngeal mucosa, which were aggravated by the nasogastric tube, finally requiring the performance of a gastrostomy due to impossibility of maintaining oral feeding.

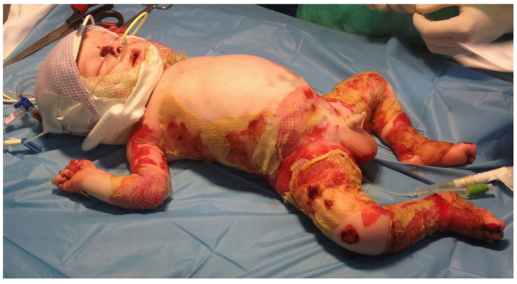

Figure 1

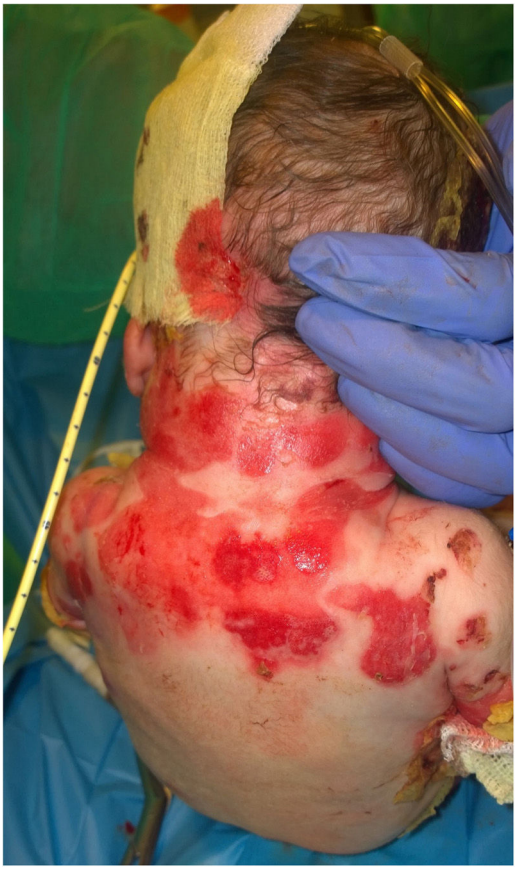

Figure 2

What is Your Diagnosis?

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Diagnosis

Herlitz junctional Epidermolysis Bullosa (H-JEB)

Diagnosis and Discussion

A skin biopsy was performed, which determined the diagnosis of Herlitz junctional Epidermolysis Bullosa (H-JEB), after antigen mapping showed a complete lack of laminin 332. He subsequently developed severe hydroelectrolytic and haematological disorders, requiring several blood transfusions. The child died at the age of 3 months because of cardiac and respiratory failure due to severe sepsis caused by *Pseudomonas aeruginosa*. Patient's parents required psychological support all through the course of the disease and have received genetic counselling.

H-JEB is attributed to homozygous mutations in the LAMA3, LAMB3 or LAMC2 genes, each encoding for one of the three chains of the heterotrimer laminin-332, resulting in the lack of expression of this protein in the skin.[1] This lethal subtype usually presents as severe and extensive non-healing mucocutaneous blistering due to skin cleavage at the lamina lucida, and are at increased risk of death from dehydration, metabolic derangement and sepsis, with 40–60% patients progressing to sepsis and 30–50% patients dying from sepsis-related complications.[2] Therefore, all efforts should be directed towards preventing these complications.[3]

The differential diagnosis for a neonate presenting with blisters must include bullous impetigo, Staphylococcal scalded skin syndrome, epidermolysis bullosa and bullous pemphigoid.[4] Any form of EB should be suspected if any mechanical skin fragility is observed at birth with extensive blistering associated with crusting and erosions. Skin biopsy should not be delayed in case of any suspected bullous disorder in the neonatal period.

H-JEB is ultimately fatal, even with the best of care. In the case we present here, the wrong initial diagnosis probably did not alter the outcome. However, an earlier diagnosis would have expedited the referral of the patient to a specialized EB centre, allowing for earlier and better supportive care. The handling of the newborn, until a final diagnosis and prognosis is assured, is a challenging and difficult time which poses complex ethical dilemmas, both to the family and healthcare professionals. Ethical questions and discussions regarding life expectancy, quality of future life and the utility of certain aggressive treatments will arise and must be addressed, for they are necessary when planning the management of these patients and their families. EB parents usually complain of the feeling of uncertainty when dealing with this disease.[5] A prompt and correct diagnosis is paramount to inform them as honestly as possible about the disease and its lethal prognosis, and to help both the patients and their parents to deal with such a desolating condition. Cases such as the one we present here should result in education and training of health personnel at all levels. As health-care providers, we have a responsibility to provide the best possible medical care, both to our patients and their families.

Conflict of Interests

The authors declare that they have no conflict of interest.

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