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Case Report

Severe Epidermolysis Bullosa Simplex in a Neonate: A Case Report

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Abstract

Epidermolysis bullosa (EB) is an inherited rare genetic connective tissue disorder. Present with widespread life-threatening blisters and fragile skin, making treatment and handling of the newborn with EB challenging. Treatments include blister care, daily skin dressing, Sepsis including with only minor skin infection, to death in the neonatal period, with severe skin infection.

Keywords: *Blisters; Epidermolysis Bullosa (EB); Pseudomonas aeruginosa; Sepsis; Neonatal*

Abbreviations

EB: Epidermolysis Bullosa; WBC: White Blood Cell; HB: Hemoglobin Level; CRP: C-Reactive Protein

Introduction

Epidermolysis Bullosa (EB) is a genetic skin defect that causes skin fragility in which the slightest friction or damage separates the skin layers, causing blisters and open wounds [1] and, in some cases, damages the mucosal membranes and organs. Children with EB typically have skin as fragile as a butterfly wing. Of every one million live childbirths, 50 are diagnosed with epidermolysis bullosa, and there is an estimated 500,000 patients with EB worldwide [2]. The severity can range from a mild localized disease to a generalized devastating process. Blisters are distributed over those areas of skin which are vulnerable to pressure or irritation. Epidermolysis bullosa simplex, junctional epidermolysis bullosa and dystrophic epidermolysis bullosa are the three major types. These three subtypes are differentiated according to the level at which tissue separates and the blisters form, that is, depending on whether this happens above within or below the epidermal basement membrane [3].

Case Report

A 15 days old neonate was hospitalized with complain of blisters on various parts of body since birth. The baby was born to non-consanguineous parents. The term (40-week) male baby with birth weight of 2.9 kg was born to 21 years old primi gravida mother by caesarean section with apparently uneventful perinatal period. The Systemic examination was normal. The blisters were present on his nose, ear pinna neck, thorax, both upper limbs extending below elbow and dorsum of hands and both lower limbs involving buttocks, thighs and extending below knee joint including dorsum of feet. There was no family history of skin lesion. Skin biopsy was performed.

Biopsy showed sub epidermal blisters and the roof of which shows full depth necrosis. Blister cavity shows fibrinous exudate with RBC's and few inflammatory cells. The Papillary dermis shows edema and mixed perivascular and interstitial infiltrate of lymphocyte, histocytes and occasional neutrophils. Base of blister shows several capillaries at the dermoepidermal junction. Initial laboratory results were as follows: white blood cell (WBC) count, 22,000/µl hemoglobin (HB) level, 13.4 mg/dl, C-reactive protein (CRP) level, 47.5; creatinine level, 0.5; The patient was diagnosed with sepsis with wound culture was positive for *Pseudomonas aeruginosa*.

The treatment was initiated with topical medications, mupirocin were used. When moving his extremities, the skin became eroded with blistering, requiring him to be wrapped with Paraffin gauze and bandages. Initially, the baby was fed on his mother's milk, using a nasogastric tube, and he was orally stimulated by short time breastfeeding. Primary pain medication dosed in three divided doses with paracetamol and sepsis was treated with IV amoxycillin and clavulanate and linezolid.

Figure 1: *Blisters over neck, upper limb, thorax, and thigh.*

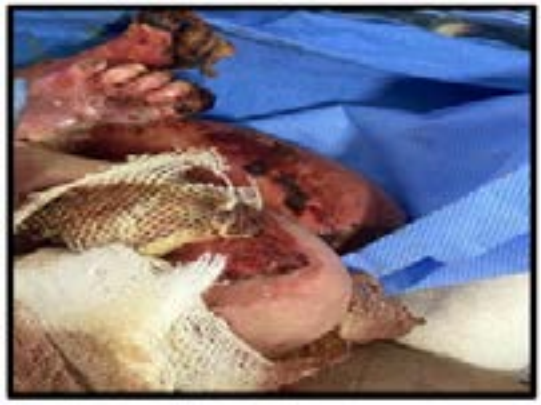

Figure 2: *Blister of lower limb.*

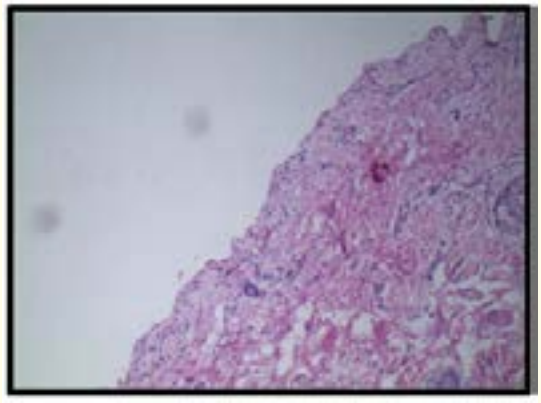

Figure 3: *Histopathology sub epidermal blister consistent with epidermolysis bullosa.*

Discussion

Epidermolysis Bullosa (EB) comprises a group of genetically determined skin fragility disorders, which are characterized by blistering of the skin and mucosa, in response to little or no apparent trauma [1,4]. These disorders represent heterogeneous phenotypes, and are associated with a variable range of complications, from localized skin fragility to neonatal death [5]. EB types are divided into four main groups according to the depth below the skin surface at which the blisters occur. Approximately 20% of EB cases are dystrophic (DEB), 10% junctional (JEB), and 70% simplex (EBS); Kindler syndrome is very rare. The genetic errors in EB result in defects in the proteins that make the outer skin layer (epidermis) adhere to the deeper layer (dermis). Some types of EB are inherited dominantly, others are inherited recessively. There are more than 30 clinical subtypes [6]. The uncertain outcome and crying of a child in pain is stressful for both the mother and healthcare professionals, calling for special handling and treatment. The collaboration between the department of dermatology having special knowledge on diagnosing and skin management of EB and the NICU, was essential in this case.

Conclusion

There is presently no definitive cure for epidermolysis bullosa. The objective of treatment is to alleviative the symptoms and provide supportive measures These patients were exposed to various types of infections, including sepsis. Daily wound care, pain management and protective bandaging are the only available treatment options. Therapy is therefore focused on the prevention and halting the progression of skin lesions and complications. Timely diagnosis and treatment of such patients are critical to reduce risk of infection.

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Conflict of Interest

The authors declare that they have no conflict of interest regarding the publication of this paper.

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