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# EPIDERMOLYSIS BULLOSA SIMPLEX: A CASE REPORT

Pharmacy

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## ABSTRACT

Epidermolysis Bullosa Simplex (EBS) is a rare genetic disorder characterized by the formation of blisters and skin lesions in response to minor trauma, such as friction or scratching. A 3-year-old female with a pre-existing diagnosis of Epidermolysis Bullosa Simplex (EBS) presented with fever and decreased oral intake, accompanied by concurrent dengue and suspected streptococcal infections. Her medical history was marked by recurrent respiratory tract infections, anemia, and severe acute malnutrition. The patient's condition was further complicated by severe anemia and pyrexia. A comprehensive treatment regimen was initiated, comprising intravenous fluids, broad-spectrum antibiotics, antifungals, antipyretics, proton pump inhibitors, vitamin and zinc supplements, and mucolytics. The patient's condition is characterized by fragile skin, recurrent infections, and nutritional deficits, which exacerbate her anemia and malnutrition. Multidisciplinary care is crucial to reduce infection recurrence and improve nutritional and developmental outcomes.

## KEYWORDS

Epidermolysis Bullosa Simplex, fragile skin, recurrent infections, nutritional deficits, anemia, malnutrition, dengue, streptococcal infection, and multidisciplinary care

## INTRODUCTION

Epidermolysis Bullosa Simplex, a rare genetic disorder with a prevalence of approximately 1 in 50,000 individuals worldwide, is characterized by the development of vesicles and cutaneous lesions in response to subtle trauma, including friction or scratching. This subset of Epidermolysis Bullosa, which comprises three primary subtypes, is distinguished by its propensity to induce blisters and skin lesions in response to minimal physical stimuli. [1]. The etiology of Epidermolysis Bullosa Simplex is attributed to mutations in the KRT5, KRT14, and COL7A1 genes, which encode proteins that are essential for the morphogenesis and homeostasis of the skin's epithelial layer [2]. The pathogenesis of Epidermolysis Bullosa Simplex is characterized by a perturbation in the normal adhesive relationships between the epidermal and dermal compartments, resulting in the formation of vesicles and cutaneous lesions. This disruption is posited to be a consequence of aberrant expression of keratin 5 and 14, which are indispensable for maintaining the integrity of the cutaneous epithelium. Furthermore, mutations in the COL7A1 gene have been correlated with EBS, implying that collagen VII may also play a crucial role in the development and progression of this disorder. [3].

The conventional treatment approach for Epidermolysis Bullosa Simplex (EBS) focuses on mitigating the manifestations of the condition and averting potential sequelae. This entails adopting a cautious approach to minimize skin trauma, employing gentle skincare regimens, and applying topical therapies to alleviate blistering and skin lesions. [4]. In instances of extreme severity, invasive therapeutic interventions may be necessitated to rectify aberrant skin lesions and restore functional integrity [5]. Genetic guidance and diagnostic evaluation may be advised for individuals with a familial predisposition to EBS. Research has demonstrated that EBS is accompanied by a profound influence on quality of life, with numerous patients experiencing affliction, anguish, and despair as a result of their condition [6]. Therefore, it is essential to develop effective treatment strategies that take into account the complex needs of individuals with EBS.

**Case Presentation:**

A 3-year-old female, second-born from a non-consanguineous marriage, presented with an 8-day history of fever and reduced feed intake for 2-3 days. Previously asymptomatic, her fever onset was sudden, low-grade, intermittent, nocturnal, and alleviated by antipyretics, accompanied by lethargy. Diagnosed with epidermolysis bullosa simplex at birth, her medical history includes hospitalization for bronchopneumonia at 4 months, recurrent lower respiratory tract infections, anemia requiring three transfusions, and severe acute malnutrition with coagulative negative staphylococcus aureus in July 2023. Her mother has diabetes, and her father has hypertension. Admitted to NICU three days post-birth for EBS, she was breastfed for 2 months and then switched to Lactogen. The child is not immunized and exhibits developmental delays, including an inability to sit unsupported, turn pages, scribble, identify facial features, and engage with her environment.

The child appeared moderately active. Vital signs included a heart rate of 130 beats per minute and a respiratory rate of 30 cycles per minute. A cardiovascular examination revealed normal heart sounds (S1S2+), and the respiratory system was clear with bilateral air entry (BAE+). The abdomen was soft and non-tender, with no organomegaly. The central nervous system examination detected no abnormalities.

**Laboratory Findings:** Hemoglobin: 6.4 grams% (decreased), White Blood Cells (WBC): 4.59/microliter, Platelets: 300,000/µL, Neutrophils: 46%, Lymphocytes: 41.6%, Monocytes: 10.6%, Eosinophils: 1.5%, Mean Corpuscular Hemoglobin (MCH): 21.3 pg (decreased), Mean Corpuscular Hemoglobin Concentration (MCHC): 28.4 g/dL (decreased), Red Blood Cells (RBC): 3.01 x 10^6/uL (decreased), Mean Corpuscular Volume (MCV): 74.8 fL (decreased), Hematocrit (HCT): 22.8 (decreased), Blood Urea: 14 mg/dL, Creatinine: 0.28 mg/dL, Sodium: 135 mEq/L, Potassium: 4.1 mmol/L, Chloride: 105 mEq/L, Total Protein: 7.5 g/dL, Dengue IgM: Negative, Dengue NS1: Positive, Antistreptolysin O (ASO): Positive, 400 IU/ml, Ultrasound: No serological abnormalities in the abdomen

**Diagnosis :**

Based on the clinical presentation and findings, the patient was diagnosed with a known case of **Epidermolysis bullosa simplex**, complicated by severe anemia and pyrexia.

**Treatment:**

The patient's treatment regimen includes several medications administered for different purposes. For short-term fluid replacement, an intravenous infusion of dextrose and sodium chloride (DNS) is administered at a rate of 32 ml per hour. To combat bacterial infections, the patient receives Piptaz (a combination of piperacillin and tazobactam) at a dose of 100 mg/kg/day, specifically 800 mg diluted in

20 cc of normal saline (NS) and given slowly three times a day (TID) for five days.

To manage fever, the patient is given intravenous paracetamol at 8 ml four times a day (QID). For gastric issues, particularly erosive gastritis and gastric acid hypersecretion, pantoprazole is administered as an 8 mg dose in 5 cc NS via slow IV once daily (OD). To address vitamin deficiency, a B complex with folate tablet is taken orally once daily (OD). Additionally, to treat zinc deficiency, the patient takes a zinc supplement syrup at 5 ml per oral (PO) once daily.

For eye dryness and irritation, methylcellulose eye drops are used four times a day (QID). The treatment also includes several antibiotics for severe bacterial infections: meropenem at 60 mg/kg/day (160 mg in 20 cc NS, given slowly TID for 12 days), vancomycin at 40 mg/kg/day (100 mg in 20 cc NS, administered slowly TID for 12 days), and linezolid at 20 mg/kg/day (80 mg given slowly twice daily (BD) for 10 days). To manage respiratory tract infections with thick mucus, the patient takes ambroxyl syrup at 2.6 ml PO TID. Lastly, for fungal infections, fluconazole is administered at 6 mg/kg/day (48 mg in 24 ml, given slowly once daily).

Each medication in this regimen is carefully chosen and dosed to address specific health issues, ensuring comprehensive and targeted treatment.

## CONCLUSION:

Epidermolysis bullosa simplex (EBS), a genetic disorder causing fragile skin, predisposes a 3-year-old female to recurrent infections and nutritional deficits, worsening her anemia and malnutrition. She presented with fever and decreased oral intake, complicated by dengue and a possible streptococcal infection. Treatment included intravenous fluids, broad-spectrum antibiotics, antifungals, antipyretics, proton pump inhibitors, vitamin and zinc supplements, and mucolytics. This intensive regimen addresses both her acute infections and the chronic effects of EBS. Comprehensive, multidisciplinary care is essential to reduce infection recurrence and improve her nutritional and developmental outcomes.

## DISCUSSION:

This case exemplifies the complex challenges in managing genetic disorders that predispose patients to recurrent infections, malnutrition, and anemia. Similar to cases of other chronic genetic skin conditions, such as Xeroderma Pigmentosum (XP), which involves extreme sensitivity to UV light leading to skin cancers and other complications, EBS requires a multifaceted treatment approach. Both conditions necessitate meticulous skin care to prevent trauma and subsequent infections, along with aggressive management of secondary complications.

However, while XP management focuses heavily on preventing UV exposure and treating skin cancers, EBS care emphasizes preventing and treating infections, nutritional support, and managing skin integrity. The use of a broad-spectrum antibiotic regimen, mucolytics, and antifungals in this EBS case parallels the comprehensive approach needed in XP to mitigate infection risks and support overall health. This underscores the importance of a tailored, multidisciplinary approach in managing rare genetic disorders to improve patient outcomes and quality of life.

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