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Blistering Skin and Blood Loss: A Rare Case of Epidermolysis Bullosa with Severe Anemia

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ABSTRACT: This case report examines an infant presenting with Epidermolysis Bullosa (EB) and severe anemia, highlighting the complexities of managing this condition. Despite receiving comprehensive care, EB significantly impacts patients' quality of life due to chronic pain and psychosocial challenges. Effective management requires a multidisciplinary approach, with emphasis on wound care, pain management, and nutritional support. Targeted therapies, such as gene editing and protein replacement, offer promising avenues for improving outcomes in EB patients, emphasizing the importance of understanding underlying genetic mutations. Collaboration among healthcare professionals, researchers, and advocacy groups is essential for advancing EB management and enhancing patient care. This case underscores the urgent need for continued research efforts to address the unique challenges posed by EB and provide better support for affected individuals.

KEYWORDS: Epidermolysis Bullosa, severe anemia, multidisciplinary care, quality of life, chronic pain, psychosocial challenges, wound care, pain management, nutritional support, gene editing, protein replacement, genetic mutations, collaborative research, and patient

INTRODUCTION

Epidermolysis Bullosa (EB) is a group of rare, inherited disorders characterized by fragile skin that blisters and tears in response to minor trauma or friction. This condition significantly impacts the quality of life of those affected and often leads to severe complications.

The lesions usually appear as thick blisters that have the potential to burst and leave scars. They can occur after birth or at any time up until early adulthood [1]

Epidermolysis bullosa has been classified into four types that are simplex, junctional, dystrophic, and Kindler syndrome. Although the three kinds of EB have distinct causes, all three presents with the same symptom's painful blisters and sores

The most common form, EB simplex, stems from mutations in the genes responsible for keratins K5 and K14, predominantly impacting the epidermis and leading to blister formation at friction sites. Dystrophic EB, linked to mutations in the COL7A1 gene, disrupts type VII collagen production, crucial for anchoring fibrils that maintain epidermis-dermis connection. Junctional EB, associated with mutations in genes encoding laminin-332, type XVII collagen, or integrin α6β4, presents with severe symptoms affecting the basement membrane skin zone. Kindler syndrome, a less common variant, involves mutations in the FERMT1 gene, characterized by blistering, photosensitivity, and progressive skin atrophy.

Symptoms of EB can vary in severity, influenced by type/subtype and inheritance pattern, with even milder cases causing moderate to severe pain, pruritus-related discomfort, and complications such as infections, sepsis, dehydration, malnutrition, anemia, and skin malignancies [2]. Prenatal diagnosis via methods like amniocentesis or chorionic villus sampling is feasible as early as the first trimester [3].

Diagnosing EB typically entails clinical assessment, family history review, skin biopsy, and genetic testing. Treatment strategies, employing a multidisciplinary approach, focus on wound care, pain management, and complication prevention. Epidermolysis Bullosa Acquisita (EBA) involves autoantibodies targeting type VII collagen, causing epidermal detachment from the dermis. Although EBA onset can occur at any age, common peaks are observed in the first three and seventh to eighth decades of life [4, 5]. Diagnosis and categorization rely on thorough personal and familial history, complemented by findings from immunomapping, transmission electron microscopy, and molecular techniques searching for defective genes. Recent advancements have enhanced understanding of the disease's clinical and molecular basis, fostering innovative clinical trials exploring novel cell- and gene-based therapies [6].

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CASE REPORT

A 3-year-old female child, weighing 8 kg, was admitted to the hospital with a one-week history of sudden-onset, low-grade intermittent fever, which was not associated with chills and rigors and was relieved by antipyretics. She exhibited decreased activity and reduced feed acceptance over the past 2-3 days. Physical examination revealed the child was moderately active with a heart rate of 130 beats per minute, a respiratory rate of 30 breaths per minute, normal heart sounds, and clear lung fields. Laboratory findings showed a hemoglobin level of 5.1 g/dL, a white blood cell count of 29,700 cells/µL, and a platelet count of 9,000 cells/µL. The child has a history of epidermolysis bullosa simplex from birth and multiple hospitalizations for bronchopneumonia and lower respiratory tract infections, with three previous blood transfusions. The family history includes a mother with diabetes mellitus and a father with hypertension.

Upon referral to Dermatology, examination revealed multiple erosions, skin atrophy, dyspigmentation, scarred areas, loss of nails from multiple digits, and milia over the face, neck, and upper trunk. The oral cavity showed erosions on the hard palate, but the teeth and scalp were normal. Ophthalmologic evaluation was normal except for epidermolysis bullosa in the right cornea.

Five days after admission, the patient tested positive for dengue NS1 antigen and developed peripheral cyanosis, leading to her transfer to the Pediatric Intensive Care Unit (PICU). She was treated with intravenous Avil (2 mg) and Ringer's lactate at 5 mg/kg. Follow-up laboratory findings indicated a white blood cell count of 19.4 x 10^3/µL, hemoglobin level of 9.4 g/dL, hematocrit of 31.2%, and a platelet count of 234,000/µL.

The treatment regimen included:

Drug	Dose	Frequency
DNS	32 ml per hour	Continuous
Piptaz	100 mg/kg/day	Continuous
Paracetamol	8 ml four times daily	Every 6 hours
Pantoprazole	1 mg/kg/dose	Every dose
Meropenem	60 mg/kg/day	Continuous
Vancomycin	40 mg/kg/day	Continuous
B-complex tablet	Once daily	Daily
Multivitamin syrup	5 ml once daily	Daily
Zinc syrup	5 ml once daily	Daily
Lacrigel ointment	4-5 times daily	Every 4-6 hours
FML ointment	Twice daily	Every 12 hours
Tobramycin ointment	Five times daily	Every 6 hours
Soframycin ointment	As needed	As needed

DISCUSSION

This case report details a patient with Epidermolysis Bullosa (EB), characterized by skin fragility and blistering from minor trauma. The diagnosis was confirmed through clinical evaluation, skin biopsy, and genetic testing. Treatment included comprehensive wound care, pain management, and nutritional support. Advanced therapies, such as gene therapy and protein replacement, were considered to address the genetic cause. The patient's response to treatment was monitored and adjusted to manage complications and improve quality of life. This case emphasizes the importance of a multidisciplinary approach and ongoing research to develop more effective treatments for EB.

Epidermolysis Bullosa (EB) arises from genetic mutations affecting proteins critical for skin integrity. In EB simplex, mutations in keratin genes compromise the epidermal cytoskeleton's stability, leading to skin fragility [7]. Dystrophic EB results from mutations in COL7A1, hindering type VII collagen production and anchoring fibril formation, disrupting the dermal- epidermal junction [8]. Junctional EB stems from mutations in genes encoding laminins, integrins, or collagen XVII, disrupting basement membrane attachment [9]. Kindler syndrome involves FERMT1 mutations, affecting kindlin-1 and impairing cell adhesion [10]. These molecular defects result in blistering, erosions, and impaired wound healing. Chronic inflammation and repeated trauma exacerbate tissue damage, impacting patients' quality of life. Understanding EB's pathophysiology aids in developing targeted therapies to alleviate symptoms and improve patient outcomes.

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Epidermolysis Bullosa (EB) profoundly diminishes patients' quality of life due to chronic pain, physical limitations, and psychosocial challenges [7]. Daily blistering, wound care, and social isolation contribute to emotional distress and financial strain [10]. Understanding the clinical implications of Epidermolysis Bullosa (EB) underscores the importance of multidisciplinary care, genetic counseling, and patient education [11]. Early diagnosis, comprehensive wound management, and psychological support are vital for optimizing patient outcomes and improving quality of life [12].

Future research in Epidermolysis Bullosa (EB) should focus on developing targeted therapies to address the underlying genetic mutations and pathophysiological mechanisms. Investigating novel gene editing techniques, such as CRISPR/Cas9, may offer potential strategies for correcting defective genes and restoring normal protein function [13]. Additionally, clinical trials exploring the efficacy of protein replacement therapies and stem cell-based treatments could provide valuable insights into improving wound healing and skin regeneration in EB patients [14]. Ongoing studies investigating the role of immunomodulatory agents and anti-inflammatory drugs in mitigating disease severity and reducing blistering episodes are also promising avenues for enhancing EB management [15]. Collaborative efforts between researchers, clinicians, and patient advocacy groups are essential for advancing understanding and treatment options for EB, ultimately improving patient outcomes and quality of life.

CONCLUSION

In conclusion, this case emphasizes the challenges of Epidermolysis Bullosa (EB) and highlights the importance of multidisciplinary care and ongoing research. Despite comprehensive treatment, EB significantly impacts patients' quality of life. Understanding genetic mutations and developing targeted therapies are crucial for improving outcomes. Collaboration among clinicians, researchers, and advocacy groups is essential for advancing EB management and supporting affected individuals.

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