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Case Report

Neonatal epidermolysis bullosa simplex: a mechanobullous skin fragility disease

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ABSTRACT

Epidermolysis bullosa (EB) is a group of hereditary skin blistering disorders characterised by skin fragility and mechanically induced blistering. We present a case of a full-term female neonate with widespread, fragile blisters and erosions at birth, prompting early suspicion of EB. Rapid dermatological evaluation and immunofluorescence antigen mapping confirmed EB simplex (EBS), allowing prompt intervention. Early parental counselling plays a crucial role in ensuring confidence in home care. After six weeks, the mother managed feeding and wound care independently. This case highlights the importance of recognising neonatal skin fragility early, facilitating timely diagnosis and intervention to minimise complications and improve long-term outcomes in infants with EB.

Keywords: Epidermolysis bullosa, Newborn, Inherited skin disorder, Skin blistering, Skin fragility, Genodermatosis

INTRODUCTION

Epidermolysis bullosa (EB) is a severe, genetically inherited skin disorder characterized by the separation of the epidermis and dermis at the basement membrane due to structural protein defects.[1] It is classified into four major subtypes: EBS, junctional EB (JEB), dystrophic EB (DEB), and Kindler syndrome, each distinguished by the specific level of blister formation within the skin layers, genes involved affecting different proteins and producing features to differentiate between them clinically. This is summarized in the following table (Table 1).[2-5]

CASE REPORT

A full-term female neonate (birth weight: 3398 g) was born via normal vaginal delivery to a 39-year-old G3P2A0L1 mother with gestational diabetes mellitus and no family history of similar skin conditions. At birth, the infant was hemodynamically stable, with APGAR (8, 9, 9) and exhibited widespread, superficial erythematous blisters containing serosanguineous fluid, along with erosions over the hands, forearms, cheeks, and gluteal region. No mucosal involvement was noted. Koebners phenomenon was positive. Given the clinical presentation, the baby was admitted to the NICU with a presumptive diagnosis of EB.

The blisters were carefully punctured with a sterile needle, cleansed with normal saline, and dressed in non-adherent Vaseline gauze. Antibacterial cream was applied to prevent secondary infections, and a thin protective layer of cream was used to minimize friction in pressure-dependent areas.

Supportive measures included the following: Gentle handling, foam-padded bedding and limited heat exposure, soft clothing and pain relief.

Histopathology of the biopsied skin revealed-hyperkeratosis, acanthosis, and a subepidermal bulla with

hemorrhage along with mild chronic inflammation in the dermis, consistent with EB. Immunofluorescence antigen mapping showed negative staining for type IV collagen, Type VII collagen, Keratin 14, and Laminin 332, effectively ruling out JEB and DEB. Based on these findings, the final diagnosis was EBS. Sepsis was ruled out and appropriate precaution was taken to prevent nosocomial infection.

The blisters exhibited a waxing and waning pattern, with older lesions healing without contractures while new lesions continued to develop in pressure-dependent and friction-prone areas. Spontaneous limb movements in the neonate, observed from the early postnatal period, appeared to precipitate lesion formation over the upper limbs (Figure 1) and distal lower limbs (Figure 2). Over the course of management, further examination revealed multiple well-demarcated erosions involving the lower extremities and trunk (Figures 3 and 4). Daily nursing care was a real challenge as even minimal pressure or friction induced blisters and erosion. The disease course was dynamic, with concurrent healing of earlier erosions and emergence of new lesions at previously uninvolved or minimally involved sites during hospitalisation (Figure 5 A and B). The mother was initially apprehensive about handling the fragile infant, fearing further skin damage. She was taught about gentle handling, safe feeding, and wound care techniques. The newborn was started with expressed breast milk feeding with a katori-spoon until the mother was confident enough to handle the baby for direct breastfeeding. Regular family counselling sessions were conducted to help them understand the prognosis and provide emotional and psychosocial support to the mother. By six weeks, the mother was confidently managing feeds and assisting with wound care. The infant was discharged with a plan for long-term follow-up, with emphasis on the following: Ongoing wound care, infection prevention and gentle handling techniques,

At subsequent follow-up visits, the baby achieved normal growth parameters and developmental milestones. The patient received routine vaccinations as per the immunization schedule, with careful consideration to minimize skin trauma during administration. Genetic studies could not be performed because of financial constraints, and parents were advised for the same before planning their next pregnancy along with counselling about the course of disease.

Table 1: Four major subtypes.

Types	Skin separation level	Genes	Affected protein	Features	Mode of inheritance
EBS	Basal layer of the epidermis (above the dermal-epidermal junction)	DSP	Desmoplakin	Blisters on mild trauma, affects friction prone areas	AR
JEB	Lamina lucida of the basement membrane (within the dermal-epidermal junction)	PKP1, JUP, KRT5, KRT14, LAMA3, LAMB3, LAMC2	Plakophilin1, Plakoglobin, Keratin 5, Keratin 14, Laminin332, Laminin332, Laminin332	Blisters at birth involves mucosal surfaces	AR, AR, AD, AR, AR, AR, AR
DEB	Below the dermal-epidermal junction	COL7A1 gene, which encodes type VII collagen (anchoring fibrils)	Collagen VII	Blisters heal with scarring and contracture, can involve esophegus	AD AR
Kindler syndrome	Mixed cleavage planes, occurring at multiple layers of the dermal-epidermal junction	FERMT1 gene, affecting cell adhesion	Kindlin-1	Blisters at birth Progressive poikiloderma, risk of cancer	AR

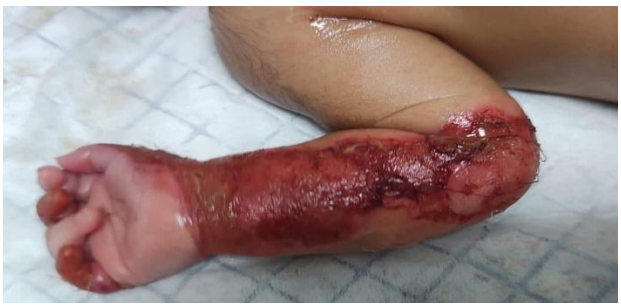

Figure 1: Extensive blistering with denuded erosions involving the distal upper limb.

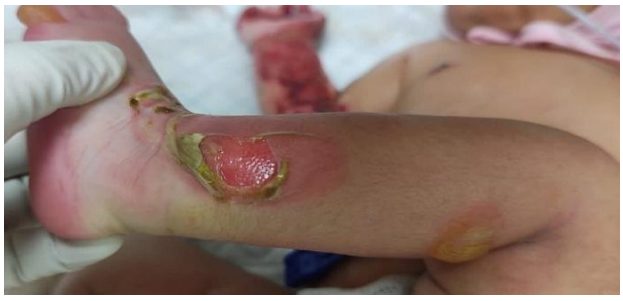

Figure 2: Erosions with surrounding erythema over ankle and lower leg following minimal mechanical trauma.

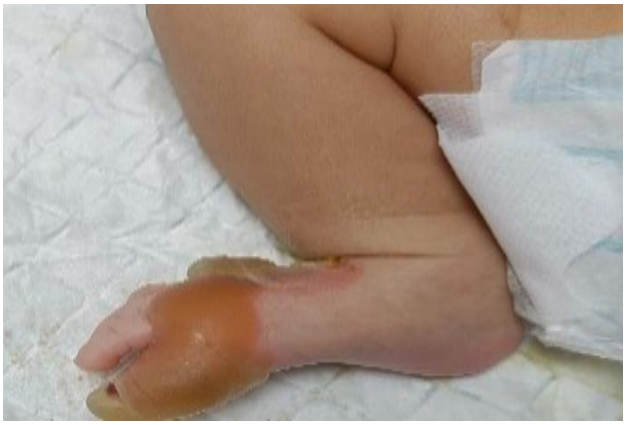

Figure 3: Tense blister over the dorsal surface of the foot in the neonatal period.

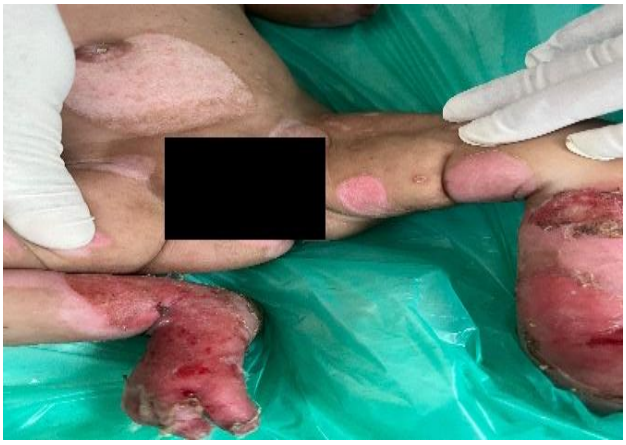

Figure 4: Extensive erosions and areas of skin denudation over the trunk and lower extremities in the neonatal period, predominantly affecting friction-prone areas.

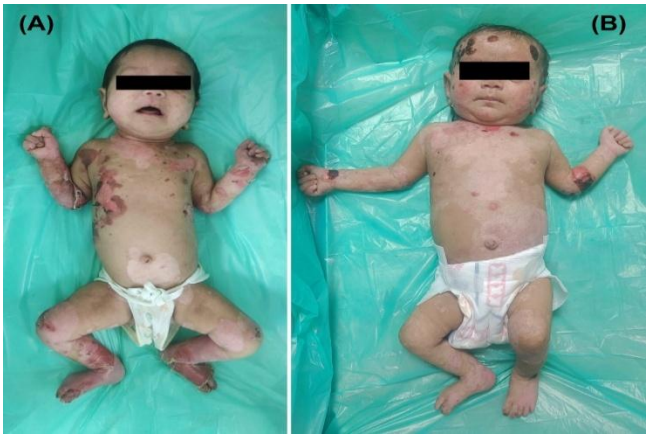

Figure 5 (A and B): Clinical progress during hospitalisation (A) Widespread erosions and denuded areas involving the trunk and extremities during the early neonatal period. (B) Follow-up image showing marked re-epithelialization of previously affected areas, with appearance of new erosions over the face not noted at during earlier presentation.

DISCUSSION

EB is a rare but severe genetic skin disorder that requires meticulous care. The key insights from this case highlight essential aspects of EB management that improve the quality of life of affected infants:

Early diagnosis and subtype identification

Precise diagnosis through skin biopsy and immunofluorescence antigen mapping is crucial for determining a specific subtype, guiding prognosis, and tailoring appropriate management strategies.

Meticulous wound care

Effective wound management helps to prevent secondary infections, and minimizing mechanical friction and pressure is essential to reduce blister formation and promote optimal wound healing.

Counselling

Comprehensive caregiver education is vital to empower parents in the handling of infants with EB. Providing psychosocial support and regular counselling helps families navigate the challenges, manage expectations regarding prognosis, and foster a supportive home environment.

Nutrition and immunization

Routine childhood immunization can be safely administered to infants with EB with appropriate precautions.

Newer treatment modalities

Recent research has identified a significant inflammatory component in EBS, generalized severe (EBS-gen sev), characterized by elevated T helper 17 (Th17) cytokines in affected skin. This insight has led to the exploration of targeted anti-inflammatory therapies.

Diacerein

As an interleukin-1β (IL-1β) inhibitor, diacerein has demonstrated efficacy in reducing blister formation and recurrence in EBS patients. Topical application of diacerein significantly decreased the number of blisters and their recurrence.[8]

Apremilast

This phosphodiesterase 4 (PDE4) inhibitor modulates the Th17 pathway. In a study involving three adult EBS-gen sev patients, apremilast treatment resulted in a marked reduction in the skin blistering and the was well tolerated.[9,10]

These findings underscore the potential of anti-inflammatory treatments targeting the Th17 and IL-1β pathways in managing EBS.[9,10]

There has been a study in which cisplatin shows its anti inflammatory action has been found to be efficacious, and has seem to reduce the blistering in a patient treated for nasopharyngeal carcinoma and had EBS.[11]

CONCLUSION

EB is a severe genetic skin disorder requiring early diagnosis, multidisciplinary care, and precise subtype identification. Gentle wound care, infection prevention, and nutritional support were associated with improved outcomes in the present case. Importantly, this case underscores the role of immunofluorescence antigen mapping as a rapid and reliable tool for early subtype differentiation in neonatal EB, thereby guiding appropriate management and providing meaningful prognostic counselling to families. Family education and counselling empower caregivers, while routine immunizations ensure protection against infections. Emerging therapies, including gene-and cell-based treatments, offer hope for future disease-modifying interventions.

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