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**Case Report**

# Congenital epidermolysis bullosa presenting with extensive bullous lesions at birth: a rare neonatal case report

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## ABSTRACT

Epidermolysis bullosa (EB) is a rare inherited genodermatosis characterized by marked skin fragility and blister formation after minimal mechanical trauma. It includes a spectrum of disorders such as EB simplex, junctional EB, dystrophic EB, and Kindler syndrome, with variable severity ranging from localized to life-threatening disease. A 28-year-old gravida 2 para 1 woman at 40+3 weeks of gestation with oligohydramnios underwent emergency lower segment cesarean section following failed induction and fetal bradycardia, delivering a live male neonate. At birth, the infant had extensive erythematous raw areas, skin peeling, and hemorrhagic bullous lesions over the extremities. A clinical diagnosis of epidermolysis bullosa simplex was made. The neonate developed progressive widespread blistering with mucosal involvement, fever, and worsening clinical condition was referred to a tertiary centre despite intensive supportive care succumbed at 42 days of life. This case highlights severe neonatal EB with poor outcome, emphasizing early recognition, supportive care, and genetic counselling.

**Keywords:** Epidermolysis bullosa, Neonatal blistering disorder, Butterfly children, Rare genetic skin disorder, Congenital skin fragility

## INTRODUCTION

Epidermolysis bullosa (EB) comprises a rare group of inherited genodermatoses characterized by marked skin fragility and formation of blisters following minimal mechanical trauma.[1] Based on the level of tissue separation within the skin, EB is broadly classified into four major types: epidermolysis bullosa simplex, Junctional epidermolysis bullosa, dystrophic epidermolysis bullosa and kindler syndrome.[2] Epidermolysis bullosa simplex is the most frequently encountered variant and primarily involves the epidermal layer of the skin.[1] Junctional epidermolysis bullosa usually presents during infancy and may manifest with severe blistering lesions.[1,2] Dystrophic epidermolysis bullosa affects deeper layers of the skin and may involve mucosal surfaces, leading to various extracutaneous complications.[1-3] Kindler syndrome is a rare subtype characterized by acral blistering in infancy, photosensitivity and progressive poikiloderma, often associated with mucocutaneous and radiological abnormalities.[2] Bart syndrome is an uncommon clinical entity associated with epidermolysis bullosa and is characterized by the triad of aplasia cutis congenita, blistering skin lesions and nail abnormalities. It is considered a clinical manifestation observed in certain forms of EB.[1,2] The clinical severity of EB varies considerably, ranging from mild localized blistering to severe generalized disease depending upon the underlying histopathological subtype. Short-term complications include dehydration, malnutrition and secondary infections leading to sepsis, whereas long-term complications may include contractures, esophageal

strictures, laryngeal stenosis and an increased risk of squamous cell carcinoma.[2,4] Management of EB remains largely supportive and focuses mainly on wound care, prevention of infection, nutritional support and pain management.[1,2,5] It is an extremely rare inherited blistering disorder with an estimated incidence of approximately 1 in 50,000 live births worldwide. Reliable epidemiological data from India remain limited due to underdiagnosis and the absence of a national registry; however, only a small number of cases have been reported from tertiary care centers across the country, highlighting the rarity of the disease.[2,5,6] Epidermolysis bullosa is a genetically heterogeneous inherited disorder caused by mutations in genes encoding structural proteins responsible for dermo-epidermal adhesion. The disease may follow either autosomal dominant or autosomal recessive inheritance patterns, resulting in defective skin integrity, recurrent blister formation and impaired wound healing.[4,7] The management of epidermolysis bullosa lesions remains clinically challenging because of the varied and complex nature of the disease manifestations. Appropriate wound dressing should be selected according to the characteristics of the lesions, including dryness, excessive exudation, colonization or infection, associated pain, pruritus and the presence of hyper granulation tissue.[2-4,7]

## CASE REPORT

A 28-year-old pregnant woman, gravida 2 para 1 living 1 (G2P1L1), a booked case, was admitted at 40 weeks +3 days of gestation with oligohydramnios. The antenatal course was otherwise uneventful, and routine hematological, metabolic and endocrinological investigations were within normal limits. Ultrasonography with colour doppler revealed a single live fetus corresponding to 40 weeks +3 days gestation with an amniotic fluid index (AFI) of 5. Her previous pregnancy was complicated by gestational diabetes mellitus managed with insulin therapy, resulting in a full-term normal vaginal delivery of a 4 kg baby. There was no history of hypertension, thyroid disorder, fever, drug intake or consanguinity. Induction of labour was attempted at term however, adequate uterine contractions could not be achieved, resulting in failed induction followed by fetal bradycardia, necessitating emergency lower segment cesarean section (LSCS). A live male neonate weighing 3.5 kg was delivered with Apgar scores of 8, 9 and 8 at 1, 5 and 10 minutes, respectively. The amniotic fluid, umbilical cord and placenta were macroscopically normal. At birth, the neonate was noted to have raw, red areas, reddish discoloration with peeling of the skin over the fingers of the right hand and the toe of the left foot. The nails were long and horny. On examination, multiple hemorrhagic bullous lesions with surrounding erythema and superficial skin peeling were observed, suggestive of marked skin fragility. The lesions showed aggravation even with minimal handling and friction. Rest of systemic examination was normal and the neonate did not have any feeding or respiratory difficulty. Skin biopsy could not be performed because of financial constraints. After dermatological consultation, a clinical diagnosis of epidermolysis bullosa simplex (EBS) was made. Conservative management was initiated with emphasis on gentle handling, prevention of trauma and maintenance of skin integrity. Soft, loose-fitting clothing was advised to minimize friction and prevent the development of new lesions.

**Figure 1: Early neonatal presentation of epidermolysis bullosa showing haemorrhagic bullae and extensive skin erosion over the fingers of right hand.**

Within 12 hours of birth, additional bullae containing sterile straw-coloured fluid appeared over the sacral region. Subsequently, widespread blistering and erosions developed over multiple areas of the body, including the buccal mucosa.

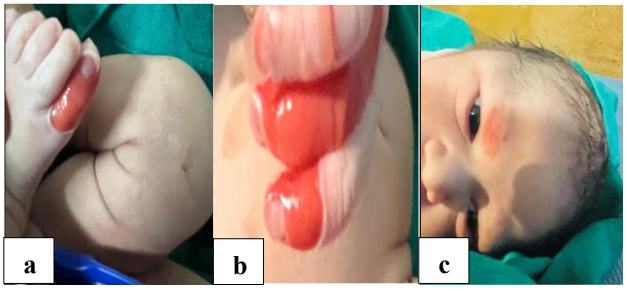


**Figure 2: Clinical manifestation of epidermolysis bullosa, (a) haemorrhagic bullae and epidermal denudation involving the digit and lower limb, (b) progressive acral blistering with tense haemorrhagic bullae over fingers, and (c) facial erosions with periocular skin involvement.**

During the course of hospitalization, the neonate developed high-grade fever reaching 103°F, along with hoarseness of cry and dysphagia, which were clinically attributed to involvement of the laryngeal and pharyngeal mucosa. In view of progressive clinical deterioration and increasing areas of denuded skin, the neonate was referred to a higher specialized centre with NICU facilities, where conservative management, prophylactic antibiotics, enteral gavage feeding and partial parenteral nutrition were continued. On follow-up, despite intensive supportive care, the condition progressively worsened, and

the infant unfortunately succumbed to the disease at 42 days of life.

## DISCUSSION

EB is a rare inherited blistering disorder with an estimated incidence of nearly 1 in 50,000 live births worldwide.[8] Indian epidemiological data remain limited due to underreporting and lack of a national registry, with only sporadic cases reported from tertiary care centres.[9] Owing to extreme skin fragility, affected children are often referred to as "butterfly children".[10] EB is broadly classified into four major types: EBS, junctional EB, dystrophic EB and kindler syndrome.[1,3,4,6,10] Among these, EBS is the most common subtype and is caused by mutations affecting keratin proteins responsible for dermo-epidermal stability, resulting in blister formation even after minimal trauma or friction.[3-5,10] In the present case, the neonate was born with extensive erythematous erosions and skin peeling involving the extremities. Progressive blister formation and superficial skin denudation were noted even with minimal handling during hospitalization, consistent with the characteristic clinical features of EBS. Mucosal involvement was also observed during the course of illness. Diagnosis of EB is primarily based on clinical presentation and may be further confirmed by skin biopsy, immunofluorescence mapping and genetic analysis.[1,3,11] However, due to financial and technical limitations, these investigations could not be performed in the present case, and the diagnosis was established clinically following dermatological consultation. Management of EB remains largely supportive and includes prevention of trauma, wound care, infection control and nutritional support.[10-12] Despite intensive conservative management at a higher specialized centre with NICU facilities, the infant developed progressive widespread blistering with extensive skin denudation and eventually succumbed to complications of the disease at 42 days of life. Although the exact cause of death could not be conclusively established, secondary infection, sepsis and metabolic or biochemical complications associated with severe EB were considered possible contributing factors. Postmortem examination could not be performed as consent for autopsy was not granted by the family.

## CONCLUSION

Congenital epidermolysis bullosa is a rare and potentially life-threatening inherited blistering disorder characterized by extreme skin fragility and recurrent bullae formation following minimal trauma. The present case highlights the aggressive neonatal presentation and poor clinical outcome associated with severe forms of the disease despite intensive supportive care. Early recognition, meticulous wound management, prevention of trauma and timely referral to specialized centres remain essential for improving patient care. As no definitive curative therapy is currently available, genetic counselling and prenatal diagnostic screening play an important role in early diagnosis and prevention in high-risk families. Furthermore, advances in molecular diagnostics, gene therapy and cell-based therapies offer promising future strategies for the management of epidermolysis bullosa. Early prenatal diagnosis through non-invasive genetic screening may also help identify severely affected fetuses in high-risk families.

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