Case Report

Severe generalized junctional epidermolysis bullosa in a newborn

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ABSTRACT

Epidermolysis Bullosa (EB) is a group of inherited skin fragility disorders. It characteristically presents as blisters formation over skin and mucosa. Epidermolysis bullosa simplex, junctional epidermolysis bullosa and dystrophic epidermolysis bullosa are three major subtypes of EB depending on the level of skin cleavage. Present case report describes a rare junctional epidermolysis bullosa in a newborn. Clinical manifestations vary from mild local blisters formation to severe one with multisystem involvement. Protection of skin, prevention of complications and supportive care are the only current treatment options.

Key words: Blisters, Epidermolysis Bullosa, Junctional epidermolysis bullosa, Skin fragility

INTRODUCTION

Epidermolysis bullosa (EB) is a rare group of inherited, mechano-bullous disorders characterized by extremely fragile skin and mucosa that blisters following minor trauma.[1] It was first reported by Von Hebra in 1870 as Erblichen Pemphigus.[2] The first classification of EB was described by Pearson in 1962 based on electron microscopic features.[3] Three major types of EB are epidermolysis bullosa simplex, junctional epidermolysis bullosa and dystrophic epidermolysis bullosa; and all are differentiated according to the level at which the tissue separates and blisters form.[4] Overall incidence of EB is around 19.60 per million live birth and that of junctional EB is 2.04 per million live birth.[5] The severity ranges from mild, localized disease to generalized devastating process and affected children are called butterfly children as their skin is very fragile. Here we are reporting a rare case of junctional epidermolysis bullosa in a new-born.

CASE HISTORY

A full-term male newborn, with birth weight of 2.13 kilograms, was born to 23 years old primigravida, with history of consanguineous marriage, by normal vaginal delivery. The newborn was admitted to the neonatal intensive care unit (NICU) of our institute with extensive erosions and blistering of the skin since birth. The lesions involved both lower limbs including dorsum of the foot, upper limbs extending below wrist, dorsum of both hands and trunk, oral mucosa and scrotal area (Figure-1). Intravenous fluids and empirical antibiotics were started through umbilical venous catheter. Trophic feeds were also started. Patient was nursed on thick foam pad to protect against trauma. Regular skin dressing was done using silver sulfadiazine, liquid paraffin and potassium permanganate crystal. During the hospital course there was appearance of new blisters at friction sites (Figure-2) and healing of older ones (Figure-3).

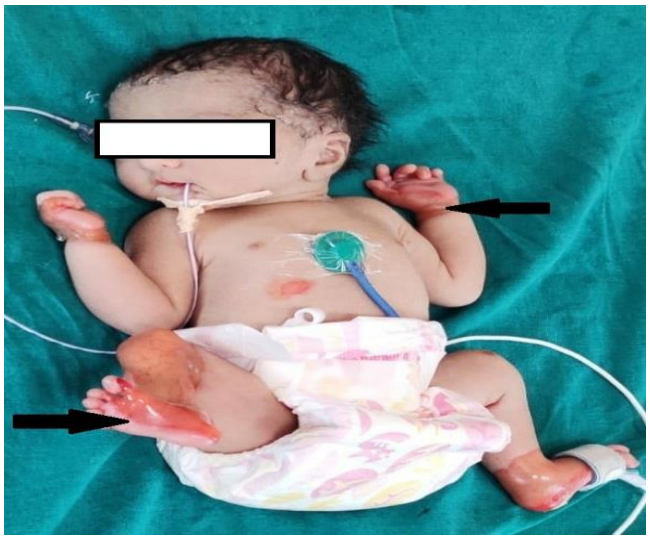

Figure-1: Extensive erosions and blistering of the skin (black arrows) at the time of birth

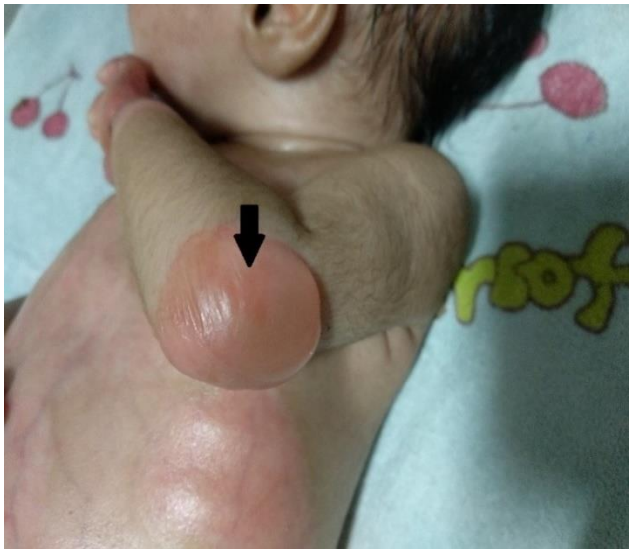

Figure-2: New bullae formation at the site of friction (black arrow)

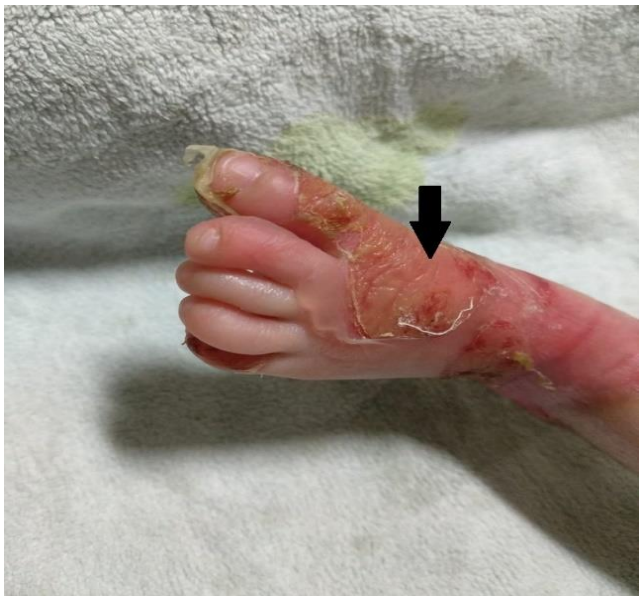

Figure-3: Single, large, well defined, bulla ruptured to form erosion with diffuse erythema and crusting over dorsum of foot (black arrow) with sparing of nails

Initially, bullous impetigo, staphylococcal scalded skin syndrome, bullous congenital ichthyosiform erythroderma and epidermolysis bullosa were considered as differential diagnosis. All blood investigations, including sepsis screening tests, were normal. Two blood cultures and local skin lesions cultures were sterile. Skin biopsy was taken from new blister which showed sub epidermal blister at dermo-epidermal junction containing inflammatory infiltrates of few neutrophils and few lymphocytes with fibrotic stroma on Hematoxylin & Eosin staining (x40) (Figure-4). Direct immunofluorescence of skin biopsy showed multifocal IgG and C3 deposits at basement membrane with sub epidermal blister, confirming the diagnosis of junctional epidermolysis bullosa (Figure-5). Oral feedings were gradually increased then full feeds were achieved from 8th day onwards. For pain relief, paracetamol was given orally. Patient was discharged on 32nd day of life (Figure 6). Counselling of parents about the disease course and gentle handling of baby was done before discharge. At 3-months follow up, multiple erosions were present over trunk, face and limbs and were managed as before. However, the baby expired at 4 months of life due to sepsis.

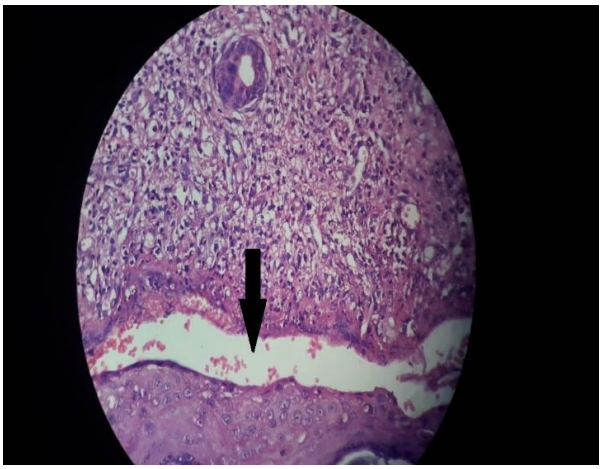

Figure-4: Skin biopsy showing sub epidermal cleft (black arrow) at dermo-epidermal junction on Hematoxylin & Eosin staining (x 40)

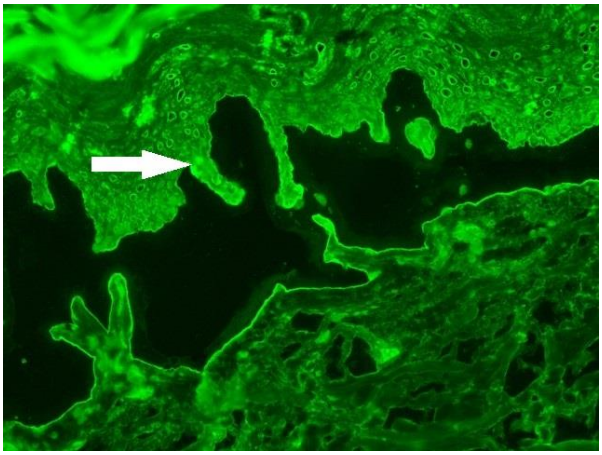

Figure-5: Direct immunofluorescence of skin biopsy showing multifocal IgG and C3 deposits at basement membrane (white arrow) with sub epidermal blister

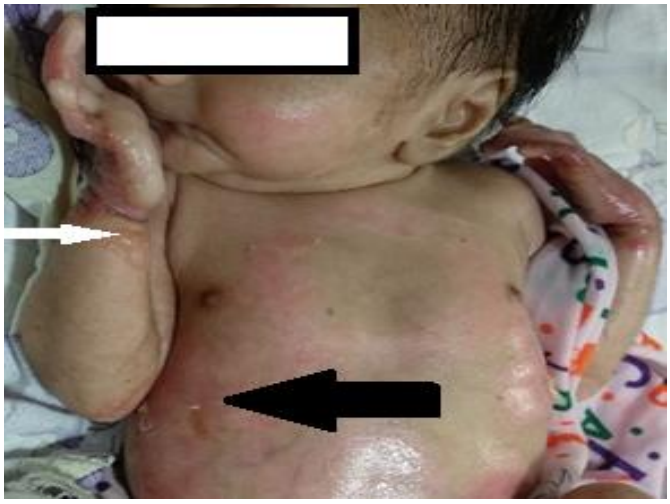

Figure-6: At the time of discharge, multiple ill defined, coalescent, erythematous patches over abdomen and cheek (black arrow), erosions with crusting over dorsum of both hands and bulla with clear fluid over ulnar border of hand (white arrow)

DISCUSSION

EB is a heterogeneous group of hereditary disorders characterized by fragility of skin and mucous membranes. EB is classified into three major types: EB simplex (EBS), Junctional EB (JEB) and Dystrophic EB (DEB), on the basis of specific cleavage plane within the dermo-epidermal basement membrane zone.[6] JEB is an autosomal recessive disorder due to mutation in laminin 332, BP 180 or α6β4 integrins genes.[7] Three subtypes of JEB are Herlitz subtype, non-Herlitz subtype and the subtype with pyloric stenosis.[8] The Herlitz subtype of JEB is a severe phenotype, in which the affected infants have intractable skin and mucous membrane erosions and blisters since birth and die early in infancy due to complications. Our case had Junctional epidermolysis bullosa, most probably of Herlitz subtype as it presented in a severe form. The non-Herlitz subtype of JEB is much milder phenotype, in which the affected patients survive up to adulthood. JEB with pyloric stenosis subtype has generalized blistering at birth with congenital atresia of pylorus, or any other congenital anomalies of genitourinary tract, and they die early in infancy.[7] In JEB, nails eventually become dystrophic and then permanently lost. Mucous membrane involvement may be severe and ulceration of respiratory, gastrointestinal, and genitourinary epithelium may also be there. In our case also, erosions were present in oral cavity.

EB can be confirmed by using immunofluorescence mapping or electron microscopy to identify the plane in which separation occurs and blisters form. In JEB, epidermis separates from basal lamina, forming a blister cavity in a plane of lamina Lucida, where hemi desmosome structure and density are frequently diminished.[7,8] In our case also, electron microscopy and immunofluorescence of skin biopsy showed blisters at dermo-epidermal junction. Final diagnosis can be done by mutation analysis.

Currently, there is no curative and definitive treatment of EB. The treatment given is mainly supportive, focused on prevention of progression of skin lesions, prevention of secondary bacterial infection, prevention of dehydration, maintenance of temperature and to provide adequate nutrition. Protein therapy, gene therapy and cell therapy are currently under trials.[9]

CONCLUSION

Epidermolysis bullosa is a hereditary skin disorder in which there is fragility of skin and mucous membrane which varies in severity from mild to life threatening. Meticulous skin care and prevention of complications are the mainstay of treatment.

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