

Case Report:

Epidermolysis bullosa in newborn: a rare case report

¹Dr. R Somdeepak Reddy*, ²Dr. (Col) D Y Shrikhande, ³Dr. Prashant Nigwekar, ⁴Dr. Shashank Panwar ,
⁵Dr.Prathmesh Patil

¹Junior Resident, Pediatrics, Pravara Institute of Medical sciences,Loni,Maharashtra

²HOD and Professor of Pediatrics, Pravara Institute of Medical sciences,Loni,Maharashtra

³Professor of Pediatrics, Pravara Institute of Medical sciences,Loni,Maharashtra

⁴Junior Resident, Pediatrics, Pravara Institute of Medical sciences,Loni,Maharashtra

⁵Junior Resident, Pediatrics, Pravara Institute of Medical sciences,Loni,Maharashtra

*Corresponding author : email somu4reddy@gmail.com

Abstract:

Epidermolysis bullosa is a rare genetic connective tissue disorder. It has many genetic and symptomatic variations but all share the prominent symptom of extremely fragile skin that blisters and tears from minor friction or trauma. It is always painful, is often pervasive and debilitating. It affects 1 out of every 50,000 live births and those born with it are often called 'Butterfly Children'. There is no treatment or cure. Daily wound care, pain management and protective bandaging are the only available treatment options. We are reporting a case of epidermolysis bullosa brought to us on 16th hour of life, patient was managed conservatively.

Key words: Epidermolysis bullosa, butterfly children, mechanobullous disorder

BACKGROUND:

Epidermolysis bullosa (EB) is a heterogeneous group of hereditary disorders characterized by extreme fragility of the skin and mucous membranes, which gives rise to the formation of blisters and ulcers following minor trauma. [1]As the areas of the body most often affected are sites subjected to frequent pressure or friction, these conditions are also called mechanobullous disorders.

Three major types are :

- 1) Epidermolysis bullosa simplex
- 2) junctional epidermolysis bullosa
- 3) dystrophic epidermolysis bullosa

Epidermolysis bullosa simplex (EBS) is the most common among them [2,3]

The disease is always painful, is often pervasive and debilitating. It affects 1 out of every 50,000 live

births and those born with it are often called 'Butterfly Children' because as the analogy goes, their skin is as fragile as the wings of a butterfly. There is no treatment or cure. Daily wound care, pain management and protective bandaging are the only available treatment options.

Herewith we described a female neonate with blistering of the skin during the immediate neonatal period.

CASE REPORT:

Herewith we reported a case of Epidermolysis bullosa simplex (EBS) :

- A term female child with epidermolysis bullosa, born to a 29 year old lady with P5 D4 L1 ,delivered vertex vaginally without any adverse perinatal events, baby birth weight was 2802 grams.

- Mother has a bad obstetric history , 1st baby at 3 month of age and 4th baby in neonatal period died due to similar illness with pustular and vesicular lesions of skin, 2nd baby was a term still birth, 3rd died at 7 month of age baby was born with imperforate anus for which colostomy was done in neonatal period.
- The present Baby was brought to our hospital at 16th hour of life with following findings.
- Baby had blistering of the skin involving both the lower limbs and upper limbs predominantly on right lower limb involving the dorsum of foot. [Fig.1,2], arising most frequently over pressure points,
- Oral cavity, conjunctiva, cornea, nails, scalp and genitalia were normal. Systemic examination was normal.
- The bullae peeled off on 48th hour of life.
- Baby was treated conservatively, empirical antibiotic coverage was given to treat secondary bacterial infection , good nursing care of skin was taken.
- Baby was nursed on thick foam pads to protect from undue trauma inducing blister formation.
- Parents were explained of the condition of the baby and discharged on persistent request on 4th day of life.
- Clinically peeling appeared more superficial without bleeding suggestive of epidermolysis bullosa simplex.



Figure- 1



Figure-2



Figure 3

DISCUSSION

"Butterfly children" is the term given to those baby born with this disease, as their skin is seen to be as delicate and fragile as that of a butterfly. Epidermolysis bullosa refers to a group of inherited disorders that involve the formation of blisters following trivial trauma. Over 300 mutations have been identified in this condition. These disorders represent heterogeneous phenotypes and are associated with a variable range of complications, from localized skin fragility to neonatal

death[4]. There are three types of genetically inherited EB: Simplex, Dystrophic, Junctional. These three major types of EB differ phenotypically (what physical manifestations look like) and genotypically (the genetic make up) as well as by the area of the skin where there is blistering, otherwise known as

“the site of ultra-structural disruption or cleavage”. There is also an autoimmune form of the disorder called, EB Acquisita.

On May 19, 2007, 18 leading EB authorities met to review the classification system of EB and update it to reflect current knowledge.

Level of skin cleavage	Major EB type	Known targeted protein(s)
Intraepidermal (“epidermolytic”)	EB Simplex	Keratins 5 and 14
Intra-lamina lucida (“lamina lucidolytic”)	Junctional EB	Laminin-332, type XVII collagen
Sub-lamina densa (“dermolytic”)	Dystrophic EB	Type VII collagen
Mixed	Kindler syndrome	Kindlin-1

EB simplex (EBS), is a non scarring, autosomal dominant disorder [2], although the mode of transmission is recessive in some subtypes [5]. EB is caused by mutations in the DNA code that make up genes. Genes are responsible for making and expressing proteins. Genes identified are those that encode keratins 5 and 14 in Epidermolysis bullosa simplex (EBS), which makes up intermediate filaments of the basal keratinocytes[2,6]. In Epidermolysis bullosa simplex (EBS)-Koebner, blisters are usually present at birth or during the neonatal period. Sites of predilection are the hands, feet, elbows, knees, legs and scalp [2, 7]. Which is similar in our case. Intraoral lesions are minimal. Nails rarely become dystrophic and usually regrow even when they are shed. The dentition is usually normal. Epidermolysis bullosa simplex (EBS) blisters typically heal with minimal to no scar and do not result in skin atrophy [2, 7]. Secondary infection is the primary complication. The propensity to blister decreases with age, and the long term prognosis is

good. Blisters should be drained by puncturing, but the blisters top should be left intact to protect the underlying skin [2].

Localized EBS of the hands and feet (Weber-Cockayne type) often presents when a child begins to walk.[8]. There is presently no definitive cure for EB and the objective of treatment is to alleviate symptoms and provide supportive measures. Therapy is therefore focused on the prevention of lesions and complications. EB is not a contraindication for any vaccination [9]. A key to successful management is expert nursing care. Nursing the babies on thick foam pads protects them from undue trauma induced blistering. Genetic counseling is recommended for prospective parents who have a family history of any form of epidermolysis bullosa. During pregnancy, chorionic villus sampling to test the fetus. For couples at high risk of having a child with epidermolysis bullosa, the test can be done as early as 8 – 10 week of pregnancy.

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