

Epidermolysis Bullosa Associated Septicemia in a Neonate-case Report

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<http://dx.doi.org/10.13005/bpj/969>

(Received: March 01, 2016; accepted: April 30, 2016)

ABSTRACT

Epidermolysis bullosa is a rare genetic connective tissue disorder that typically manifest at birth or early childhood or adults with various subtypes (simplex, recessive, dystrophic and junctional) 17 days old male baby born to primi consanguineous parent presented with serous blisters and atrophic scars on arms and body since birth. referred to our hospital as baby developed dystrophic nail and oral lesions, blistering of skin involving both upper and lower limb below knee joint including dorsum of foot and involving upper limb extending below elbow joint, dorsum of both hand. minimal trauma elicited fresh blisters. dermatologist advised skin biopsy, deferred due to financial constraints. systemic examination was normal. no family history of bullous skin lesion. Baby initially had bullous skin lesions over abdomen, hands and legs. oral antibiotics (amoxicillin), t.bact oint outside, since new lesions found antibiotics changed to augmentin. liquid paraffin for e/a. for super added infections linezolid and minimal handling advised, baby on breast — feed, increasing in weight 3.5kg on follow up at 60 days of life. There is no definite, approved management. avoid the baby from rough handling, breast feeds and vit e also suggested. stem cell based therapy is on trial

Key words: Epidermolysis bullosa, epidermolysis bullosa dystrophica, newborn, blistering.

INTRODUCTION

Epidermolysis bullosa comprises a group of genetically determined skin fragility disorders characterized by blistering of the skin and mucosa following mild trauma^{1,2,3}. Epidermolysis bullosa is a rare group of inherited disorders that manifest as blistering or erosion of the skin and in some cases the epithelial lining of other organs, in response to little or no apparent trauma, the exact prevalence of epidermolysis bullosa is unknown. mild variant have been estimated to occur as frequently as 1 in 50,000 births⁴. Approximately 400,000-500,000 are affected worldwide and no definitive treatment have yet been developed^{5,6}.

Case report

A term 17 days old female baby born to a 3rd degree consanguineous parents (primi mother, 24 years old with the history of gestational diabetes on meal plan, history of PROM more than 24 hours delivered by LSCS indication being CPD with foetal distress) was admitted to NICU for blisters, of skin of the hands and foot from birth onwards. on examination there was blistering and peeling of the skin involving abdomen and mouth also. Baby was treated outside with amoxicillin and breast feeds. As the lesions in the tongue were increasing there was difficulty to suck at the breast.



Fig.1: Blistering and Pleeing of the skin - hands

After initial septic work up, heamogram, x ray chest and abdomen, baby was continued breast feeds directly and also through paladi. Baby was started on syrup augmentin. Dermatologist suggested dressing with liquid paraffin. Birth weight of the baby was 2.8 Kg. admission weight in NICU was 2.47 Kg. For the next few days, lesions were started resolving but new lesions were appearing on and out but lesser in severity

Investigations showed haemoglobin 13.3 g /dl, total WBC count 26400 /cu mm neutrophils 68% lymphocytes 25 eosinophil 1% monocytes 6%, Pcv 40%, platelet 7.5 lakhs /cu mm. The blood culture showed methicillin resistant coagulase negative strephalococcus (MR_CONS) sensitive to amikacin, Clindamycin, Gentamicin, Rifampin, Linezolid, Teicoplanin, Vancomycin treated with continued breast feeding and paladai feeds syrup augment in 1.25 ml twice a day for one week. Syrup Linezolid 1.25 ml twice a day for one week justee drops 0.5 ml, zincovit drops 1 ml liquid paraffin and teaBactointment. After a stay of 12 days in nursery baby was discharged with a weight of 2.5 kg. baby was reviewed after one month with a weight of 3 kg the last visit was after one month the weight picked upto 3.5 kg advise on discharge with regular immunisation schedule with continuing breast feeds with vitamins use of white soft clothes less of trauma.



Fig. 2: Blistering and Pleeing of the skin - Abdomen chest

DISCUSSION

More than 30 types of epidermolysis bullosa have been described, rendering categorization of the types controversial and often confusing. An international consensus meeting in Vienna, Austria in 2008 reaffirmed the following currently used names for the four major types of epidermolysis bullosa⁷.

Epidermolytic: epidermolysis bullosa simplex (EBS) - where blistering occurs in the upper layer of the skin (the epidermis)[4]. This is the most common type of EB, accounting for 70% of cases and tends to be milder than the other types[8]. This intraepidermal type is divided into 2 major subtypes: suprabasal and basal. Suprabasal EB targets transglutaminase 5; plakophilin 1; desmoplakin; plakoglobin proteins. Basal EB targets keratin 5 & 14; placenta; exophilin 5 (slac 2-b); bullous pemphigoid antigen 1⁹.

Dermolytic

Dystrophic epidermolysis bullosa (DEB) - where blistering occurs below the basement membrane zone in the upper part of the dermis⁴. DEB accounts for around 25% of cases. The dystrophic forms of EB are characterized by

deformities of the skin including coalescence of the fingers, blistering, scarring, nail changes and milia formation and have either autosomal recessive or autosomal dominant inheritance. In the most severe cases, scars may cause either ankyloglossia or microglossia [1, 10]. This affects the sublamina densa layer and is divided into 2 types: dominant dystrophic epidermolysis bullosa (DDEB) and recessive dystrophic epidermolysis bullosa (RDEB). Both the dominant and recessive types target the collagen VII protein

Lecidolytic

Junctional epidermolysis bullosa (JEB)- where blistering occurs at the junction between the epidermis and dermis (lower layer of the skin) in a layer of skin known as the basement membrane zone [4]. JEB accounts for around 5% cases and is usually considered the most severe type of EB [8]. This affects the intralamina lucida layer and is divided into 2 major subtypes, generalized and localized. Generalized JEB targets collagen XVII; lamina-332, $\alpha 4$ integrin proteins [9].

Kindler syndrome

Multiple levels of blistering, a unique clinical phenotype-photosensitivity [4]. Kindler syndrome affects multiple layers of skin while targeting the fermitin family homolog 1 (kindling-1) gene⁹.

The identified genes include those that encode keratin 5 & 14 in epidermolysis bullosa simplex, collagen VII in dystrophic epidermolysis bullosa and laminin 5 in Herlitz junctional epidermolysis bullosa. Toward the end of millennium as the complex structure of desmosomes and hemidesmosome was unravelled, the genes responsible for the rare subtypes were found, including those that encode $\alpha 4$ integrin in epidermolysis bullosa with pyloric atresia, plectin in epidermolysis bullosa with muscular dystrophy and plakophilin in epidermolysis bullosa with ectodermal dysplasia [4]. In our case, ultrasound abdomen the pylorus of the stomach was normal. During the past few years, systemic data collection and analysis have been performed on several thousand of patients with epidermolysis bullosa worldwide and more than 1000 mutations,

encompassing more than 10 structural genes have now been documented. More has been learned about the molecular basis of epidermolysis bullosa a group of diseases that share clinical or molecular features with several other genodermatoses.

Cytolysis causes blisters in the epidermis or basement membrane zone of the skin. In epidermolysis bullosa simplex, cytolysis causes blisters in the basal or spinous layers of the epidermis and keratinocytes often have abnormal density and organization of keratin filaments. In junctional epidermolysis bullosa the epidermis separates from the basal lamina forming a blister cavity in the plane of the lamina lucida where hemidesmosome structure and density are frequently diminished. In dystrophic epidermolysis bullosa the basal lamina remains attached to the epidermis but the blister cavity forms beneath the lamina densa of dermal-epidermal junction, and anchoring fibrils may appear abnormal, reduced in number or altogether absent⁴.

Epidermolysis bullosa simplex Ogna has been described in Norwegian individuals. Epidermolysis bullosa is an autosomal inherited disorder, the incidence does not differ by sex⁴.

This revised classification of EB incorporated several new genetic subtypes. Our proposed systemic onion skin approach using successive layers of skin approach using successive layers of clinical, immunohistochemical and molecular findings will prove useful to both clinicians and researchers and adaptable to future discoveries⁹. Special dental concern involving the use of soft toothed brushes and irrigation techniques. Pure diets are recommended because the lesions are affecting the oral mucosa and esophagus. There is also a need for diet supplements, such as vitamins, proteins and iron in order to prevent anemia. The use of corticoids, vit E and immunosuppressive drugs have also been suggested for the treatment of epidermolysis bullosa.

ACKNOWLEDGEMENTS

DR.P.P thanks DR.Raja gopal hod of dermatology, SRM medical hospital for the help in management of this case.

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