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CASE REPORT

EPIDERMOLYSIS BULLOSA IN NEWBORN: A RARE CASE

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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. The severity can range from a mild, localized disease to a generalized, devastating process. Its 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.

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INTRODUCTION

Epidermolysis bullosa (EB) is a rare group of inherited disorders that manifests 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 (Uitto and Pulkkinen, 2000). An international consensus meeting in Vienna, Austria in 2008 reaffirmed the following currently used names for the four major types of epidermolysis bullosa:

- Epidermolytic - Epidermolysis bullosa simplex (EBS)
- Lucidolytic - Junctional epidermolysis bullosa (JEB)
- Dermolytic - Dystrophic epidermolysis bullosa (DEB)
- Multiple levels of blistering - Kindler syndrome

The leading authority on epidermolysis bullosa added Kindler syndrome as the fourth major epidermolysis bullosa type in 2008 with a unique clinical phenotype—photosensitivity (Fine *et al.,* 2008). EBS is the most common amongst them (Morelli, 2007; Cooper *et al.,* 1984). EBS may manifest either at birth or during the neonatal period and there is no treatment or cure. Daily wound care, pain management and protective bandaging are the only available treatment options. Herewith we described a male neonate with blistering of the skin during the immediate neonatal period.

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CASE REPORT

- A preterm male child (36 weeks) with epidermolysis bullosa, was born to a 24 year old Primigravida female with a regularly supervised and apparently uneventful antenatal period in our hospital in January 2016.
- She came in preterm labour with absent membranes and cervical dialataion of 3cm and contracted pelvis.
- The baby was delivered by caesarean section with Apgar score of 6/10 without any adverse neonatal events.
- There was no history of similar disorder in the families of either of the non-consanguineously married parents.
- Mother's level II ultrasound and Doppler were normal. There was no family history of genetic disorders, skin disorders and diabetes.
- The birth weight of baby was 2.5 kilograms.
- Baby had blistering of the skin involving both the lower limbs below knee joint including dorsum of foot and involving the upper limb extending below the elbow joint, dorsum of both the hands (Figure 1) . Similar lesions were also present of buttocks (Figure 2)
- Baby had blistering involving the lower lip also. Minimal trauma elicited fresh blisters.
- Oral cavity, conjunctiva, cornea, nails, scalp and genitalia were normal. Systemic examination was normal.
- The bullae peeled off on second day of life.

- There was no family history of bullous skin lesions. The diagnosis of Epidermolysis bullosum was considered.
- Baby was treated conservatively; empirical antibiotic coverage was given to treat secondary bacterial infection .Good nursing care of skin was taken.
- On 10th day of life, the baby had aspirated while he was being fed and was put on Ventilator support.
- Parents were explained of the condition of the baby.

Clinically peeling appeared more superficial without bleeding suggestive of epidermolysis bullosa simplex.

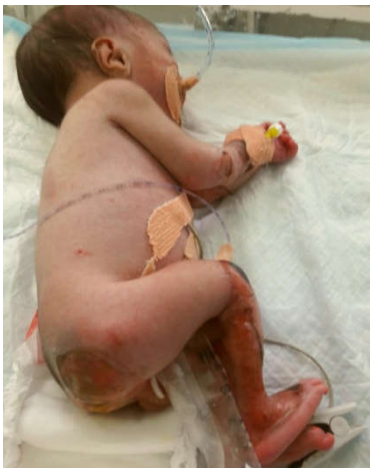

Figure 1. Baby had blistering of the skin involving both the lower limbs below knee joint including dorsum of foot and involving the upper limb extending below the elbow joint, dorsum of both the hands

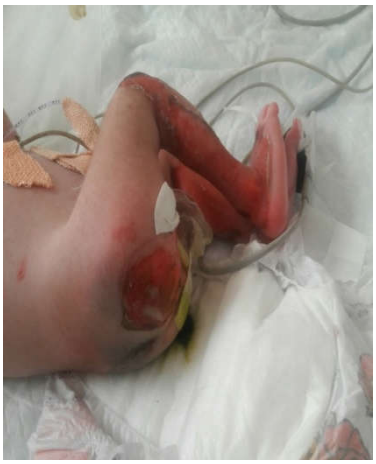

Figure 2. Similar lesions were also present of buttocks

DISCUSSION

Epidermolysis bullosa (EB) is the term given to a group of inherited mechanobullous disorders involving blistering of the skin and sometimes mucous membranes in response to minor frictional trauma. Each form has a specific cleavage plane within the epidermal-dermal basement membrane zone, as well as specific clinical manifestations. Each form of EB has distinguishing pathophysiology, mode of inheritance, and prognosis. These disorders represent heterogeneous phenotypes and are associated with a variable range of complications, from localized skin fragility to neonatal death (Johnson *et al.,* 1999). “Butterfly children" is the term given to those babies born with this disease, as their skin is seen to be as delicate and fragile as that of a butterfly. The exact prevalence of epidermolysis bullosa is unknown. Mild variants have been estimated to occur as frequently as 1 per 50,000 births. The more severe varieties are believed to occur in 1 per 500,000 births annually. The onset of epidermolysis bullosa simplex is at birth or early infancy while that of junctional epidermolysis bullosa is at birth. The onset of dystrophic epidermolysis bullosa is at birth or early childhood and the onset of Kindler syndrome is usually within the first year of life. 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 dermoepidermal junction, and anchoring fibrils may appear abnormal, reduced in number, or altogether absent. Epidermolysis Bullosa Simplex (EBS), the mildest and most common type of EB, is characterized by the lysis of basal keratinocytes above the basement membrane zone. Most subtypes of EBS are autosomal dominant mutations in Keratin 5 or 14 (such as Weber-Cockayne, Koebner, and Dowling-Meara); however, autosomal recessive mutations and mutations in other genes have been reported. Keratin 5 and 14 function in the adhesion of cells to the hemidesmosome through plectin, another EBS associated protein.

Most EBS children experience an increased tendency to blister as temperature increases; however, their overall prognosis is excellent with normal life expectancy and decreased tendency to blister with time. The Weber-Cockayne (WC) EBS subtype is the mildest, most common subtype of EBS where the bullae, though temperature sensitive, are confined predominantly to the hands and feet (De Kanter, 2004). In Koebner (K) EBS, patients experience generalized blistering with little to no mucosal involvement. 20% of patients experience nails involvement; however, all symptoms improve with advancing age. Dowling-Meara (DM) EBS is most severe in the neonate and infant, and can be fatal in the neonatal period. Large generalized blistering predominates; however, blisters do decrease with age. DM children experience profound mucosal involvement, as well as nail involvement and mild acral blistering. Unlike other major EBS disorders, EBS with muscular dystrophy is characterized by an autosomal recessive mutation in the gene coding plectin (Hintner *et al.,* 1981). These patients experience early onset generalized blistering with laryngeal and mucosal involvement, as well as progressive muscular dystrophy. Finally, EBS with mottled pigmentation is a rare autosomal dominant subtype characterized by mechanically induced blistering, unaffected mucosa, and a mixture of hyper- and hypopigmented macules (Featherstone *et al.,* 2007). There is no specific treatment for any of the variants of EB. Care for patients with EB is fundamentally preventative and symptomatic. The current treatment strategies focus on preventing the formation of new

blisters, preventing and treating infections, facilitating wound healing, providing nutritional support, managing extracutaneous complications, Optimum management of this disease can only be achieved by a multidisciplinary team, which should include the following specialists: dermatologist, surgeon, nutritionist, dentist, physiotherapist, nurse, psychologist, pain specialist, and geneticist (Sarkar *et al.,* 2011). The treatment plan must be individualized and optimal communication among team members is a vital factor in obtaining good results. Psychological support for parents and family members is vital (Sianez‑Gonzalez *et al.,* 2009). The 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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I am grateful to the parents for giving permission to publish this image for academic purpose.

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