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PAEDIATRICS
Short Communication

Congenital Epidermolysis Bullosa

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Case

A 1.6 kg, preterm (32 weeks), female child was born through a normal vaginal delivery to a primigravida mother. The cause of prematurity being preterm premature rupture of membrane. On examination she was seen to have desquamating lesion around all limbs, face and also of left ear pinna [figure 1-4]. Baby was started on iv fluids and antibiotics in view of suspect sepsis. Dermatological consultation was sought for erosive lesion all over the body with keeping epidermolysis bullosa into consideration. A skin biopsy was sought which clinched the diagnosis of epidermolysis bullosa simplex (generalised). Nails were not involved .There was no scaring or alopecia. The infant had turbulent course in the nursery and infant expired at the age of 2 weeks because of Klebsiella sepsis

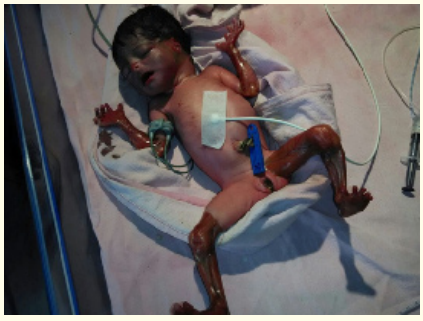

Figure 1: *Figure showing desquamation of the skin of four limbs of the neonates. Also note that there are prominent veins over the eroded area.*

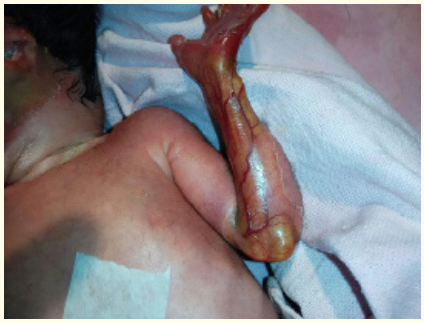

Figure 2: *Figure showing eroded area of left upper limbs with underlying erythematous skin. Also see the visible veins and eroded ear pinna.*

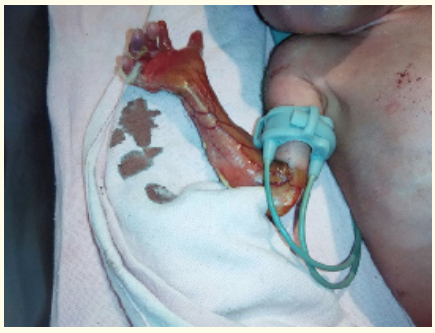

Figure 3: *Figure showing eroded area of left upper limbs with underlying erythematous skin.*

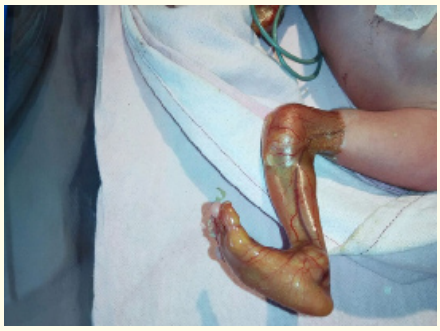

Figure 4: *Figure showing desquamated skin of the left lower limbs with erythematous underlying skin. There is also well demarcated line differentiating normal and eroded skin.*

Discussion

Epidermolysis bullosa (EB) is a group of dermatological hereditary bullous diseases, which are experiencing varying levels of skin and mucous fragility in common. They are a consequence of mutations in skin structural proteins. There are four major types of EB, that are grounded upon the ultrastructural level of tissue cleavage in the peel:

a. EB simplex (intra-epidermal)
b. Junctional EB (intra-lamina Lucida)
c. Dystrophic EB (sub-lamina densa)
d. Kindler syndrome (intra-epidermal, intra-lamina Lucida, and sub-lamina densa). [1-2]

The badness of the disease and its subtype is determined by the degree of blistering of skin and type of variation and is has varied presentation in different subtypes of EB.WHEN TO SUSPECT EPIDERMOLYSIS BULLOSA-EB should be counted as a differential diagnosis in any newborn infant who presents at the outpatient department with blisters and/or erosions in the absence of another plausible etiology (eg, staphylococcal infection).

Management of neonates with EB should be performed in an intensive care unit, with health care personnel having the necessary expertise and resources necessary to handle the extensive erosions or potential complications related to widespread skin sloughing. [1] Expert nursing care is an important key to successful treatment of newborn infants with EB.

Diagnosis-Skin biopsy for immunofluorescence microscopy (IFM) is the beginning footfall in the diagnosis of Epidermolysis bullosa (EB). A biopsy of unaffected, non-rubbed skin may be required in patients with extreme mechanical skin fragility (a characteristic feature of recessive EB). In these patients, performing a punch biopsy may be sufficient to get the cleavage plane necessary for diagnosis. [3, 4]

Newborns presenting with skin lesions suspicious of EB should be treated with utmost precision. They should be set along a thick foam pad and the lodgings should be utilized for sending the baby. High temperature may induce blisters in some newborn infants with EB; the dangers and benefits of using an incubator must be learned separately. If suction is necessary, a soft catheter should be picked out and minimal suction pressure exerted. [5] In newborns with severe dystrophic EB, over-zealous suction can strip the mucus, resulting in scarring and earlier development of pharyngeal strictures. It is preferable to delay bathing until lesions present at birth have healed. The erosions can be cleaned by gentle irrigation with warm sterile normal saline and incubated with non-adherent dressings. [6, 7] Dressings are changed on one arm at a time to prevent self-inflicted trauma from giving up the bare skin along the opposite limb if the baby is shaken during the modification. Disposable nappies can be used, but should be lined with a soft cloth to reduce trauma from the flexible boundaries. Erosions on the diaper area can be protected by liberal application of a variety of white soft paraffin and liquid paraffin in equal shares. [5] Clothing helps to prevent self-inflicted skin damage from kicking or rubbing. Clothes should be turned inside out to prevent skin damage from the beds

Learning Points

a. EB should be suspected in neonates presenting with blisters and erosions without other plausible etiology.
b. EB is a visual diagnosis and needs to be identified early to prevent complications.
c. Judicious and meticulous nursing care is the corner store of therapy.
d. Rule out other cause causing blisters like Bullous congenital ichthyosiform erythroderma, Incontinentia pigmenti , Aplasia cutis congenita , Congenital erythropoietic porphyria and Bullous mastocytosis

Bibliography

1. Fine JD., *et al*. "The classification of inherited epidermolysis bullosa (EB): Report of the Third International Consensus Meeting on Diagnosis and Classification of EB". *Journal of the American Academy of Dermatology* 58.6 (2008): 931-950.
2. Fine JD., *et al*. "Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification". *Journal of the American Academy of Dermatology* 70.60 (2014): 1103-1126.
3. Fine JD. Hereditary epidermolysis bullosa (EB) biopsy sites.
4. Intong LR and Murrell DF. "How to take skin biopsies for epidermolysis bullosa". *Dermatology Clinics* 28.2 (2010): 197-200.
5. Petronius D., *et al*. "A comparative study of immunohistochemistry and electron microscopy used in the diagnosis of epidermolysis bullosa". *The American Journal of Dermatopathology* 25.3 (2003): 198-203.
6. Pigors M., *et al*. "Lack of plakoglobin leads to lethal congenital epidermolysis bullosa: a novel clinico-genetic entity". *Human Molecular Genetics* 20.9 (2011): 1811-1819.
7. Groves RW., *et al*. "A homozygous nonsense mutation within the dystonin gene coding for the coiled-coil domain of the epithelial isoform of BPAG1 underlies a new subtype of autosomal recessive epidermolysis bullosa simplex". *Journal of Investigative Dermatology* 130.6 (2010): 1551-1557.

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