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Case Report

Epidermolysis bullosa in newborn: a case report

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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 trauma in the varying degrees of severity and can severely incapacitate the life of the afflicted patient. Treatment still remains a major challenge. Daily wound care, pain management and protective bandaging are the only available treatment options.

Keywords: Blistering, Epidermolysis bullosa

INTRODUCTION

Epidermolysis bullosa is a heterogenous group of hereditary disorders characterized by fragility of the skin and mucous membrane that manifests as blistering of the skin in the varying degree of severity following mechanical trauma. The severity can range from a mild localized disease to a generalized devastating process. Blisters are distributed over those areas of skin which are vulnerable to pressure or irritation. Epidermolysis bullosa simplex, junctional epidermolysis bullosa and dystrophic epidermolysis bullosa are the three major types. These three subtypes are differentiated according to the level at which tissue separates and the blisters form, that is, depending on whether this happens above within or below the epidermal basement membrane.[1]

CASE REPORT

21 days old neonate was hospitalized with complain of blisters on various parts of body since birth. The baby was born to non-consanguineous parents. The late pre-term (36-week 5 days) male baby with birth weight of 3.5 kg was born to 23 years old second gravida mother by caesarean section with apparently uneventful perinatal period. Systematic examination was normal. The blisters were present on his neck, thorax, both upper limbs extending below elbow and dorsum of hands and both lower limbs involving buttocks, thighs and extending below knee joint including dorsum of feet.

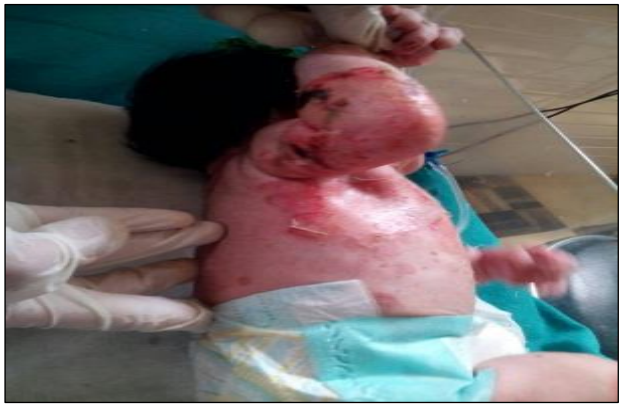

Figure 1: Blisters in right axilla extending to elbow.

Blisters were of variable sizes. Oral cavity, conjunctiva, cornea and scalp were normal. Nails were dystrophic. There was no family history of skin lesion. Nikolsky sign

was positive. Paraffin gauge dressing was applied on the wounds.

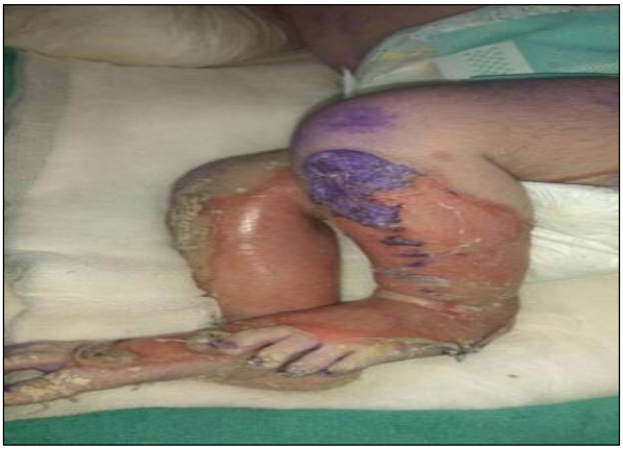

Figure 2: Blisters overs both lower limbs involving feet.

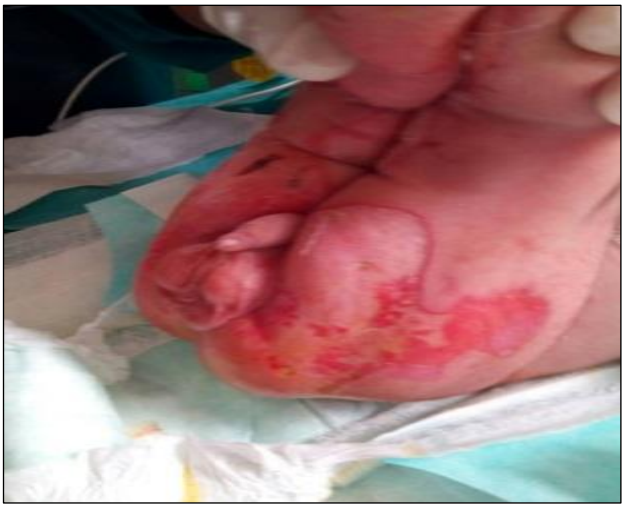

Figure 3: Blisters over scrotum and thigh region.

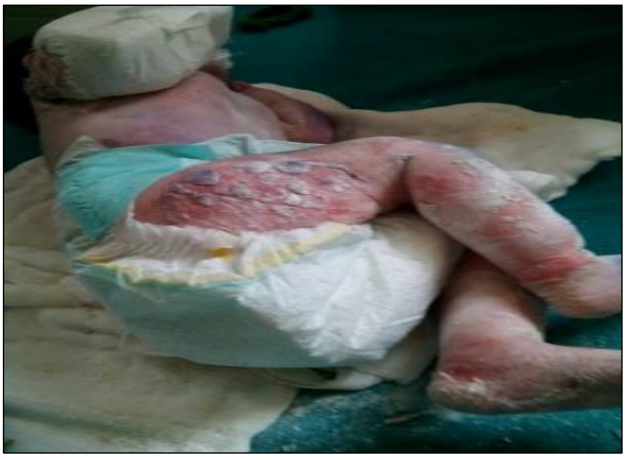

Figure 4: Blisters over right lower limb extending from thigh to involve feet.

Skin biopsy was performed. Biopsy showed sub-epidermal blisters that contains few neutrophils and eosinophils. Floor of the blisters was partly covered by fibrin, plasma and a few inflammatory cells. The papillary dermis shows edema and mixed perivascular and interstitial infiltrate of lymphocytes and neutrophils with occasional eosinophils.

On the basis of biopsy report and clinical features diagnosis of epidermolysis bullosa simplex was confirmed. Cefotaxime and Linezolid was given to patient.

DISCUSSION

Epidermolysis bullosa comprises a group of genetically determined skin fragility disorders, which are characterized by blistering of the skin and mucosa, in response to little or no apparent trauma.[1,2] These disorders represent heterogeneous phenotypes, and are associated with a variable range of complications, from localized skin fragility to neonatal death.[3] This group is classified on the basis of the mode of inheritance, clinical, laboratory and epidemiological studies into three major forms: epidermolysis bullosa simplex, junctional epidermolysis bullosa and dystrophic epidermolysis bullosa.

This disease was first to be explained by Von Hebra in 1870 as ‘Erblichen Pemphigus’, which was later modified to its recent name ‘epidermolysis bullosa hereditaria’ by Koebner in 1886. The first classification of the disease was done even much later in 1962 by Pearson.[5]

Epidermolysis bullosa simplex is an autosomal dominant disorder, although the mode of transmission is recessive in some subtypes.[7] Sites of predilection are the hands, feet, elbows, knees, legs and scalp.[6] Epidermolysis bullosa simplex blisters typically heal with minimal or no scar and do not result in skin atrophy.[8] Most cases of Epidermolysis bullosa simplex are due to mutation in the cytokeratin genes 5 (KRT5) or 14 (KRT14). These genes encode for intracellular proteins, K5 and K14 respectively, which are responsible for maintaining tissue integrity. They largely affect the mechanical integrity of the basal keratinocytes, which causes a substantial trauma to induce a blister.[9]

CONCLUSION

There is presently no definitive cure for epidermolysis bullosa. The objective of treatment is to alleviative the symptoms and provide supportive measures. Therapy is therefore focused on the prevention and halting the progression of skin lesions and complications. Nursing the babies on thick foam pads protects them from undue trauma induced blistering. The erosions should be cleaned with sterile normal saline and covered with non-adherent dressing. Non-adhesive dressing pads or Vaseline impregnated gauge covered by soft, bulky

dressing are ideal. Epidermolysis bullosa is not a contraindication for any vaccination.[9] Psychological support for parents and family members is vital.[10] Nutritional support is important for adequate growth and development and to promote optimal wound healing. To families of affected children, prenatal diagnosis using molecular techniques offers genetic counseling.

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