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CASE REPORT

CONGENITAL EPIDERMOLYSIS BULLOSA IN A FEMALE CHILD WITH ONE AFFECTED SIBILING: A RARE CASE REPORT

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ABSTRACT

Epidermolysis bullosa (EB) is a rare group of hereditary disorders, which are characterized by blistering of the skin and mucosa due to little or no apparent trauma. The severity can range from mild, localized skin blisters to generalized, systemic life-threatening disease and the treatment is mainly supportive. We reporting a case of congenital EB in a 5months female child born to a gravid five mother by normal delivery, the child was with blisters on both upper and lower limbs since birth. There was history of second degree consanguinity and history of bullous lesions in previous child and the child was expired on 5th day because of respiratory infection. Based on the clinical features of the child we diagnosed it as a dystrophic epidermolysis bullosa.

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.[1],[2] The incidence and prevalence of EB According to the National EB registry project from USA were estimated to be 19.60 per million live births and 8.22 respectively.[3] Different types of epidermolysis bullosa are there, among that epidermolysis bullosa simplex, junction EB, dystrophic EB are the major types. These three major types are based on the blister formation that is above, within, or below the epidermal basement membrane. In 2008 another EB type kindler syndrome was included in major types that is characterized by blistering that arises in multiple levels within and/or beneath the basement membrane zone (BMZ), rather than within a discrete plane, as occurs in all other EB types.[4]

Major EB type	Level of blister formation
EB simplex	Intraepidermal
Junctional EB	Intra-lamina lucida
Dystrophic EB	Sub-lamina densa
Kindler syndrome	Multiple levels (intra-lamina lucida and sub-lamina densa).

Treating epidermolysis has become a challenge. Gene therapy is used in treating epidermolysis bullosa, it include in vivo, ex vivo and in-utero gene therapy.[5] Complications of epidermolysis bullosa include chronic anemia, dialated cardiomyopathy, osteopororis/ osteopenia, corneal abrasions, squamous cell carcinoma, malnutrition, dysphagia, chronic constipation.[6]

Case report

A 5months female child of weight 4.3kgs was born to a gravid five mother by normal delivery, the child was with blisters on both upper and lower limbs since birth. The blisters were gradually grown and spread throughout the body. There was history of second degree consanguinity and history of bullous lesions in previous child and the child was expired on 5th day because of respiratory infection.

The bullous lesions on present baby were spread throughout the body as baby growing. Multiple bullous lesions were present over lower abdomen, lips, both upper and lower limb and back side of the body. Milia formation was also there on the baby's upper limbs and lower abdomen region. The bullous lesions were filled with fluid and got ruptured, leading to red colour wound on the affected area and were healed by forming a scar. Based on the clinical features we estimated it as dystrophic EB Frequent scaring had lead to roughening of the skin. The diagnostic tests for type of bullous were not done because of financial problem. The infant was discharged with tropical emollients, skin protective measures and nutritional supplements to catch up the growth.

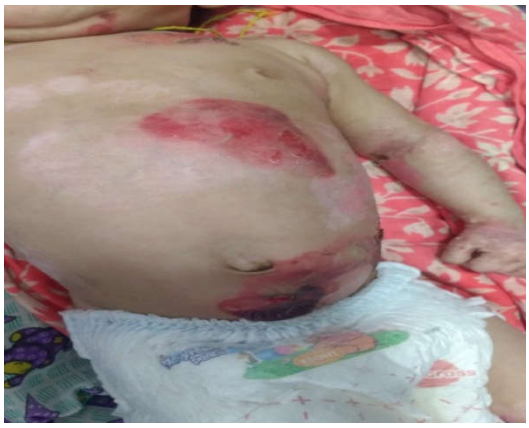

Fig.no 1 Haemorrhagic blisters on lower abdomen and scar at the healing area

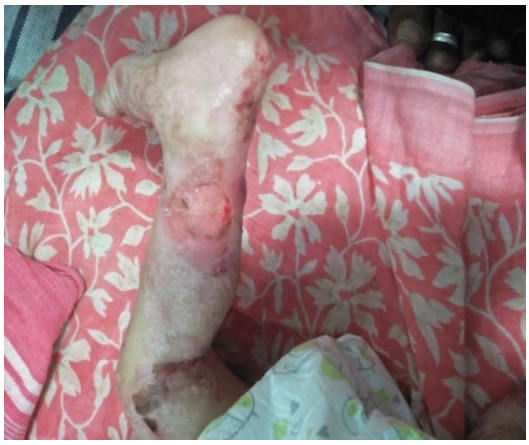

Fig.no 2 Reddish wound on the lower limb region after blister breakdown

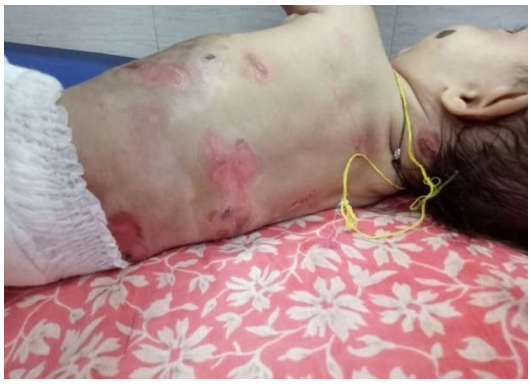

Fig.no 3 Blister healing and Scar formation on the back side of the body

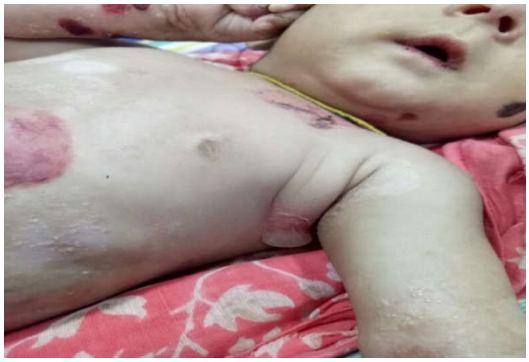

Fig.no 4 Blister healing on the lips and above the chest region and Milia formation on upper limbs and Lower abdomen region

DISCUSSION

Epidermolysis bullosa is a group of hereditary disease it affects the patient's quality of life. Based on plane of separation, Pearson classified 3 major EB by using electron microscope in 1962. In the major forms of EB, EB simplex is the most common form, next common form is dystrophic EB followed by junctional EB.[7]

Genetic abnormalities in epidermolysis bullosa:[7]

Type of epidermolysis bullosa	Gene affected	Protein encoded
EB simplex	PKP1	Plakophillin 1
	DSP	Desmoplakin
	KRT5	Keratin 5
	KRT14	Keratin 14
	PLEC1	Plectin
	ITGA6,ITGB4	Integrin, α6β4
Junctional EB	LAMA3, LAMB3, BLAMC2	Laminin 322
	COL7A1	CollagenVII
	ITGA6, ITGB4	Integrin, α6β4
Dystrophic EB	COL7A1	collagenVII
Kindler syndrome	KIND1	Kindlin-1

Inheritance and clinical features of major EB [3]

Type	Inheritance	Clinical features
EB simplex	Autosomal dominant, rarely recessive	Localised blisters or grouped vesicles, limited mucosal involvement, plamoplanatar hyperkeratosis, nail dystrophy, normal teeth and hair.
Junctional EB	Autosomal recessive	Wide spread blistering, scarring, significant granulation tissue, severe mucosal involvement, dental pitting, Alopecia, nail dystrophy.
Dystrophic EB	Both autosomal dominant and recessive	Hemorrhagic blisters, scarring, milia, pseudo syndactyly, severe mucosal involvement, physical and sexual retardation, significant morbidity and mortality.

Based on the clinical features we classified this case as dystrophic epidermolysis bullosa. This EB is both dominant and recessive, in dominant EB skin and mucosal involvement is less. When compared with dominant, in recessive EB the large portion of the body surface is affected. The skin and mucosal involvement is more that is extended to some of the internal mucosa and eyes. In all dystrophic forms the gene which encodes for protein collagen Vll is affected; this is main component for fibrils which play a role in attachment of epidermis and dermis. Less quantity or lack of these fibrils can cause separation of both epidermis and dermis.[6] Severe forms of dystrophic EB have multiple problems including digital fusion, contractures, chronic wounds, nutritional deficiencies and are at a high risk of squamous cell carcinoma in adulthood.[8]

Treatment of the EB has become challenge to clinicians. Gene therapy, A choice in the treatment of EB, it include in-vivo, ex –vivo and in-utero gene therapy. In in-vivo gene therapy transgene introduction and genetic incorporation is quick and economically cheap. But it is more difficult to monitor and control the extent of transgene introduction and genetic incorporation.

In ex-vivo gene therapy the patient own cells are used as a vector to gene incorporation by this we can reduce the possible immune reactions. In this therapy we can easily observe the effects of the transgene.

In-utero gene therapy the foetus is treated in the developmental stage so they prevent the early damage of the epidermis and other affected organs, significantly improving patient survival.[5]

In one study they used onabotulinum toxinA to treat EB simplex. Holaban *et al* reported that 50U of onaboutlinumA toxin was taken in 4ml ns and it was given as 1U per site at the weight bearing plantar regions of a 6years old African-American boy. After the first administration next 100U of onaboutlinumA toxin was given as 2U per each site for 4months. The patient was well tolerated with the both procedures without complication. He experienced decreased pain, bullae, malodor, and less hyperhidrosis, first noted approximately 2 weeks after each treatment.[9]

A prospective phase-2 clinical trial was done by Agnes Schwieger-Briel *et al* in dystrophic EB patients. He used oleogel S10 as a tropical agent to treat the wounds, it contain a betulin-rich triterpene extract from birch bark. They observed faster re-epithelialization of wounds in patients with dystrophic EB when treated with Oleogel-S10.[10]

CONCLUSION

Epidermolysis bullosa is affecting the patient quality of life, proper treatment should be needed. There is no proper treatment for EB so further research should be needed in the treatment area.

Aknowledgement

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Conflict of Interest: None

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