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Case Report

Familial epidermolysis bullosa simplex– A case report

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Abstract Epidermolysis bullosa is a rare disorder with frequent blistering at trauma prone sites. We report a case of 10 days old female infant presenting with flaccid yellow colored fluid filled blisters over the extremities. On histopathological examination, a diagnosis of epidermolysis simplex was made. After treatment with appropriate antibiotic, the infant was discharged in satisfactory condition. The treatment options include optimal wound care and prevention of trauma. Newer therapies like gene therapy are in the pipeline, but still have a long way to go before some specific treatment can be advocated.

Key words
Fragility, treatment, blisters, genetic, trauma, epidermolysis.

Introduction

Epidermolysis bullosa is a skin fragility disorder with blisters on skin and mucosa due to trauma or sometimes even without any obvious trauma**.** It is of three types depending upon the level of split.[1] Out of all the three main variants, epidermolysis simplex type carries a relatively better prognosis especially with the localized variant. As of now, this disorder has no cure.[2] So, the main focus of the treatment is optimal wound care, prevention of trauma induced blisters by gentle handling of the child and adequate nutritional support.

Case report

We report a case of 10 days old female infant presenting with flaccid yellow colored fluid filled blisters over the hands, feet, legs, arms, nose and chin. No mucosal involvement was there. There was no history of fever. The size of the bulla ranged from 0.5-3.5 cm. The bigger bullae were drained under aseptic conditions. Biopsy was taken from a fresh blister and was sent for histopathological examination. The histopathology showed vacuoles in the basal layer of epidermis with a cleft in the lamina lucida and the diagnosis of epidermolysis bullosa simplex was made. Antigen mapping could not be performed due to non availability and unwillingness of the patient.

There was family history of blistering in the older sibling, the record of which was unavailable. The neonate was a product of non consanguinous marriage. The neonate was initially put on systemic antibiotic Syrup linezolid before the report of culture and sensitivity came and subsequently was put on Inj. Vancomycin after the culture and sensitivity report showed the growth of MRSA. No topical antibiotic was given to avoid bacterial resistance and only topical 1% gentian violet paint was given. The infant showed improvement in 8 days and was discharged home satisfactorily. The parents of the neonate were advised to come for follow up regularly and were advised Vaseline gauze dressings. The mother was advised gentle handling of the infant especially during bathing

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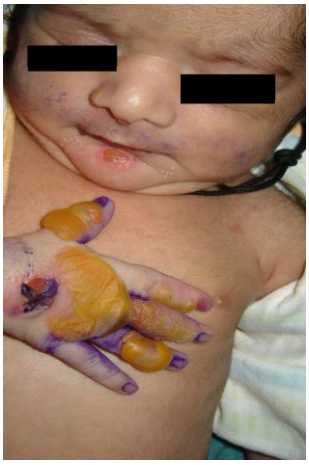

Figure 1 10 days old infant showing blisters over hands, chin and nose.

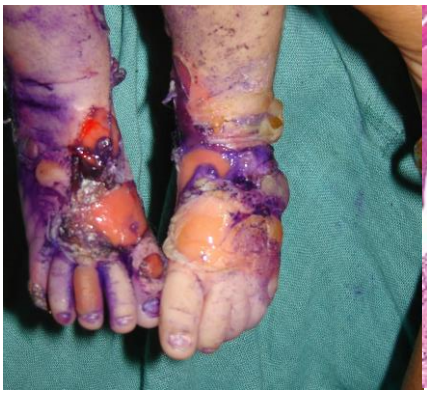

Figure 2 10 days old infant showing erosions on the feet.

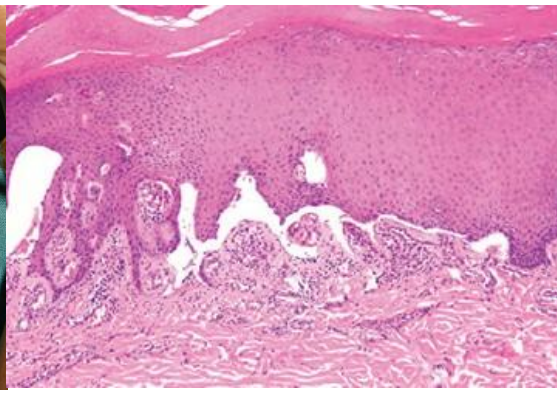

Figure 3 Histopathological image of EB Simplex showing cleft in the lamina lucida.

and feeding. The parents were advised to go in for genetic counselling.

Discussion

Epidermolysis bullosa (EB) is a group of genetic blistering disorder in which there is marked fragility of the skin and mucous membranes.[3] There is appearance of blisters and erosions over the trauma prone areas appearing at or just after birth. There are various types of EB with variable morbidity and mortality.[4] In the milder forms There is continuous blistering in the milder variant but it does not really affect the survival of an individual. But in the severe forms, the mortality rate of infants is high due to dehydration, metabolic disturbances and sepsis.

There are four types of EB depending on the level of split: EB Simplex (epidermolytic), Junctional EB (lucidolytic), Dystrophic EB (dermolytic) and kindler's syndrome (mixed cleavage plane). The laboratory investigations are important to confirm the diagnosis and classification of EB. The important investigations are immunofluorescence antigen mapping, transmission electron microscopy and mutation analysis. Immunofluorescence antigen mapping is relatively easy to perform and results are also rapid.[6]

Various treatment options include optimal skin and wound care, avoiding trauma and basics of topical treatment of EB include avoiding trauma, good skin care, disinfection and wound management. The management of EB represents a multidisciplinary approach involving dermatologists, paediatricians, dentists, ophthalmologists, surgeons, nutritionists, psychologists and social workers, since it has high personal and socioeconomic impact on the patients and their family members.

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