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Case Report

Blistering in a newborn: a rare case report

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ABSTRACT

Epidermolysis bullosa is a heterogeneous group of inherited mechanobullous disorders that present with skin and mucosal fragility, leading to blister formation after minimal trauma. 18 days old female baby presented with fluid-filled blisters of variable sizes over the lower lip, bilateral heels, hands, buttocks, chest, legs and arms, which ruptured spontaneously leaving raw areas. The history and physical examination suggested epidermolysis bullosa simplex and so, skin biopsy was done to confirm the diagnosis. On confirmation, patient's parents were counselled about the disease and its management.

Keywords: Epidermolysis bullosa simplex, Trauma, Erosions

INTRODUCTION

Epidermolysis bullosa (EB) is a heterogeneous group of inherited mechanobullous disorders that present with skin and, in some cases, mucosal fragility, predisposing patients to the development of blisters and/or erosions after minimal trauma or friction. Children with a recurrent history of these kinds of lesions or neonates that present them in the absence of another reasonable explanation should be investigated. Diagnosis must be based on clinical and histopathological findings. To date, management of inherited EB basically consists of avoiding traumas that trigger lesions, as well as preventing infection and facilitating healing of the wounds with the systematic use of bandages.[1] Epidermolysis bullosa simplex is one of the major forms of EB, the exact prevalence of which is unknown, but this condition is estimated to affect 1 in 30,000 to 50,000 people.[2] We are reporting this case to create awareness regarding this rare disease and its management.

CASE REPORT

18 days old female child born from non-consanguineous marriage presented with fluid-filled blisters, of variable sizes, over the lower lip, bilateral heels, hands, buttocks, chest, legs, and arms, present since birth (Figures 1-5). Bullae ruptured spontaneously within a few hours leaving raw areas. Mother of the child was diagnosed with positive IgG for herpes in TORCH panel during pregnancy, so the child was treated for herpes simplex by the local practitioner with no relief. The child was referred to a dermatologist for further evaluation. There was no history of fever, seizures, or any other mucosal involvement.

On examination, the sites involved were oral mucosa, bilateral arms, legs, chest, hands and feet. Multiple bullae, of size varying from 0.5-2.5 cm in diameter, covered the sole of the right foot. Erythematous raw areas over the sole of the left foot, bilateral palms and chest were seen. Multiple crusted plaques were present over the chest. Oral mucosa had 1 large erythematous raw area

over the inner side of lower lip. Nails had erythematous to black hyperpigmentation with mottled appearance. Based on history and clinical examination, provisional diagnosis of epidermolysis bullosa simplex was made and skin biopsy was sent, which showed extensive vacuolization of basal layer with a cleft at the dermo epidermal junction consistent with the diagnosis of epidermolysis bullosa simplex (Figure 6-7). Parents were counselled regarding the disease and advised to visit the hospital at regular intervals.

Figure 1: Erosion on right forearm.

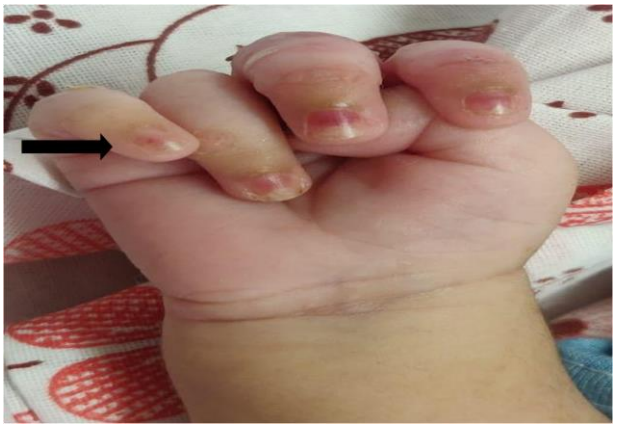

Figure 2: Blistering on the hand around the nails.

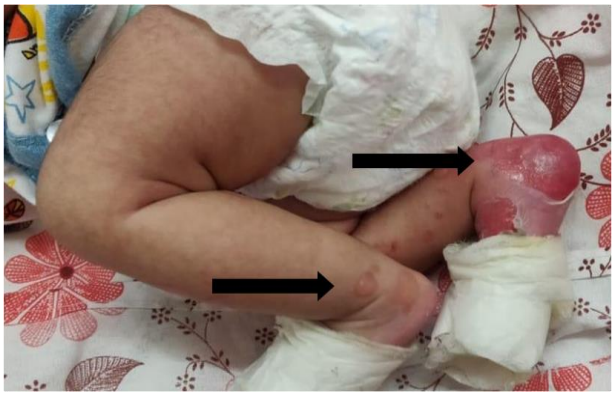

Figure 3: Blistering and erosions on soles of the feet and lower leg.

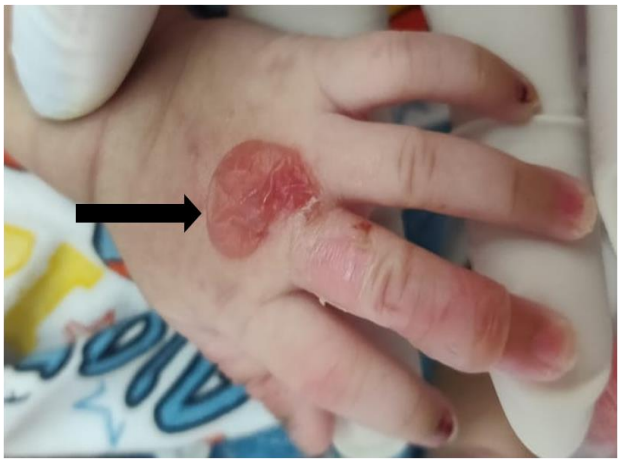

Figure 4: Flaccid bulla on hand.

Figure 5: Erosions on the lateral aspect of right side of chest.

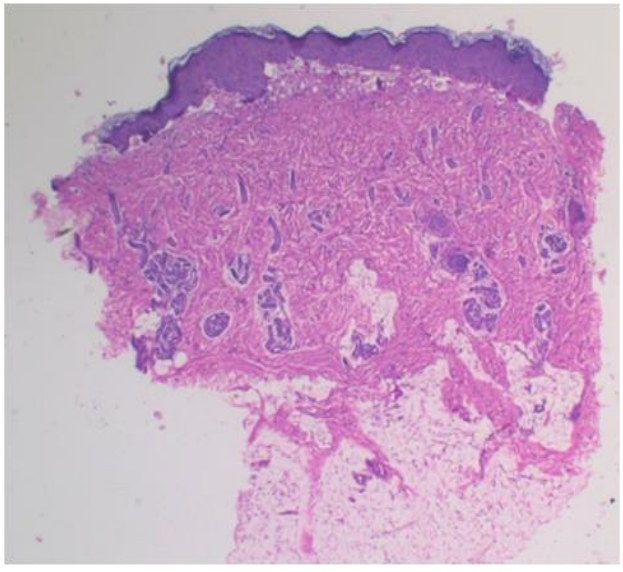

Figure 6: Extensive vacuolization of basal layer with a cleft at dermo epidermal junction.

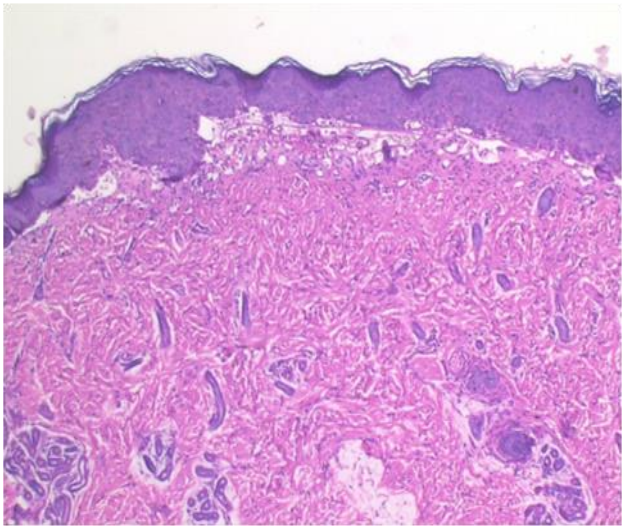

Figure 7: Close-up view of extensive vacuolization of basal layer with a cleft at dermo epidermal junction.

DISCUSSION

Epidermolysis bullosa (EB) simplex is a rare genetic condition typified by superficial bullous lesions that result from frictional trauma to the skin. Most cases are due to dominantly acting mutations in either keratin 14 (K14) or K5, the type I and II intermediate filament (IF) proteins tasked with forming a pancytoplasmic network of 10 nm filaments in basal keratinocytes of the epidermis and in other stratified epithelia. Defects in K5/K14 filament network architecture cause basal keratinocytes to become fragile and account for their trauma-induced rupture.[3]

Koebner was the first author that used the term "epidermolysis bullosa" to describe the condition in 1886. Children suffering from this condition are also called "butterfly children" – as their skin is as fragile as the wings of a butterfly- or "cotton wool babies", or "crystal skin children" (in south America).[4]

Three major forms of EB have been defined using clinical and histological criteria. The dystrophic, junctional, and simplex forms of EB are characterized by loss of tissue integrity in the upper dermis, at the dermo-epidermal interface, and within the epidermis, respectively. With rare exceptions, EB simplex is inherited in an autosomal dominant fashion. Although EB simplex is the most frequently occurring form of EB (approximately 1 case per 25,000 live births), it also is the least severe.[5]

In EB simplex, trauma-induced loss of tissue integrity consistently occurs within the basal layer of epidermal keratinocytes. The inherited defect renders basal keratinocytes fragile, causing them to rupture when the epidermis (and, in some cases, other stratified epithelia) is subjected to mechanical stress. Associated skin pigmentation anomalies can occur, but terminal epithelial cell differentiation and epidermal barrier function appear normal.[6]

Several clinical variants of EB simplex have been described. The most frequent and widely known variants - EB simplex-generalized (EBS-generalized; in which the distribution of blistering is "generalized" over the body), EB simplex-localized (in which the distribution of blistering is "localized," e.g., primarily restricted to hands and feet), and EB simplex Dowling-Meara (EBS-DM; in which blisters are also generalized but show a distinct "herpetiform" or clustered pattern) - differ primarily according to the distribution, frequency, and severity of skin blistering over the body.[7]

Other forms of EB simplex are less frequent (e.g., EB simplex–autosomal recessive [EBS-AR], which resembles EBS-generalized but is recessively inherited) and/or exhibit additional clinical features. EB simplex with mottled pigmentation (EBS-MP) is characterized by anomalies in skin pigmentation, while EB simplex with muscular dystrophy (EBS-MD) is accompanied by a progressive, limb-girdle type of muscular dystrophy.[5] Despite the degree to which clinical presentation varies, all variants of EB simplex are caused by genetically determined defects in intracellular proteins whose function is to provide essential structural support in keratinocytes of the epidermis and related tissues.[8]

After Koebner, multiple cases of EB simplex have been reported. Passarge observed 21 affected persons in 4 generations of a family with generalized EB simple described a large Arab family originating from Jerusalem in which 38 affected individuals spanning 4 generations had EB simplex. Livingston et al reported a patient who presented at 3 to 4 days of age with widespread generalized blistering, leading to painful hyperkeratosis of the palms and soles in his teen years.[9]

Diagnostically, EB remains a challenge. The definitive diagnosis of inherited EB is made with transmission electron microscopy, immunofluorescence antigen mapping (IF), and EB related monoclonal antibody testing as well as mutational analysis. In order to make a correct diagnosis, it is most important that a skin biopsy should be performed.[10]

In spite of extensive research on the molecular mechanisms and clinical manifestations of various forms of EB, a definitive treatment for the disease is still far from reality. Avoidance of provoking factors for blistering remains the mainstay of management. Heat and humidity lower the threshold for blistering in patients with EB simplex, and therefore measures to reduce both these factors are important.[10] The skin should be evaluated at least every six months for documentation and treatment of new lesions, even though most patients are reluctant to this exposure.[1]

Infants require greater care and control of the environment around them to prevent trauma. This includes gentle manipulation techniques by their caregivers, use of foam to cover bony prominences and zinc oxide anti-adherent diapers. In older children, the use of special shoes and foam in the knee to prevent blistering is recommended. Treatment decision should consider the location of the lesions, need for extra cushioning and protection, use of special dressings and clothing. Lesions should be cleaned with solutions of low toxicity, such as saline solution and water.[1] Gene therapy is the ultimate goal which is likely to become a reality in the near future.[10]

EB simplex patients can suffer from malnutrition due to a combination of decreased food intake and increased nutrient demand, which can lead to failure to thrive, delayed puberty, anemia and a cascade of clinical and biological events that, together, culminate with the interruption or retardation of wound healing.[11] Hence, this aspect of the disease should also be taken care of, with adequate diet plans, and intake-output charts.

CONCLUSION

EB simplex is a rare disease with difficult management. Caregiver counselling is the most important aspect here as multiple doctor visits and gentle skin care (at all costs) can take a toll on the caregivers. Skin biopsy is one of the simpler most accurate diagnostic techniques and should be done in every newborn with a suspicion of EB simplex. Gene therapy is the ultimate goal, which is not available yet. Till then, avoidance of provoking factors like heat and humidity, gentle manipulation techniques by the caregivers and cleaning of the areas with low toxicity solutions can be done to manage this disease. Nutritional aspect of this disease should not be avoided as malnutrition can lead to delayed wound healing.

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