Research Paper Medical Science

Epidermolysis Bullosa Dystrophica-A Case Report

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ABSTRACT

A 5-month-old male child presented with superficial blisters and erosions over pressure sites, healed areas of hyper and hypo pigmentation over face, trunk and extremities and deformities of hands and feet. The child was born of non-consanguineous marriage and had no family history of blistering disorder. The case diagnosed as epidermolysis bullosa emphasizes the significance of its clinical consideration in neonatal blistering disorders for early diagnosis and treatment.

Keywords : Epidermolysis bullosa,atrophy,dyspigmentation,Onychodystrophy,onycholysis

Introduction

Epidermolysis bullosa comprises of heterogeneous group of inherited mechano-bullous disorders usually encountered in infancy or early childhood and are characterized by spontaneous blister formation in response to trivial trauma.1These relatively rare disorders (incidence: 1/50000 live births to 1/500000) live births) have been broadly classified into three groups; simplex, junctional & dystrophic types2 based on level of split in the basement membrane zone.

Case report

A 5-month-old male child was referred to dermatology OPD from paediatric department with complaints of spontaneous blister formation at pressure sites (knees, back, elbows, ankle, soles) ruptured blisters forming ulcers and healed atrophic hypopigmented scars. No consanguinity was reported in parents. There was no history of affection of siblings or any family members.

Examination revealed superficial flaccid blister on dorsum of left hand (Figure 1). Ruptured blisters with superficial peeling and erosions were seen on medial side of flexor aspect of left forearm (Figure 2) and over both soles. Face, trunk, back and extremities revealed multiple, discrete, focal areas of atrophy and mottled dyspigmentation (hyper and hypo pigmentation) (Figure 3).Oral lesions were absent.

Hands and feet revealed mitten deformities (fused digits) of left hand (Figure 2) and almost all toes of both feet (Figure 4 & 5).Onychodystrophy and onycholysis of almost all the nails was seen. There was no systemic involvement. Patient was clinically diagnosed as epidermolysis bullosa (dystrophic type).

Blister fluid culture sensitivity was done to rule out secondary infection. Patient was screened by ultrasound to rule out presence of pyloric atresia or other anomalies of gastrointestinal tract.

The patient was treated with Vaseline gauze and antibiotic gauze dressings, topical mupirocin at sites of erosion and oral antibiotics. Barrier protection and avoidance of trauma was advised. Regular follow up with dentist and ophthalmologist was advised to rule out oral and eye complications. Patient is regularly screened for the development of pre-malignant skin lesions and squamous cell carcinoma.

Discussion

Different clinical and histological types of epidermolysis bullosa have underlying defects in genes coding for structural proteins involved in constitution of hemidesmosomes of dermoepidermal junction e.g proteins K5/K14 in simplex type; laminin5 and α6β4 integrins in junctional type and COL7A1 in dystrophic type.2,3.Loss of adhesions between basal cells,basement membrane and underlying dermis results in non-inflammatory split in or below the epidermis.Based on the level of split, these dermatoses have been grouped into 3 major categories; simplex,junctional and dystrophic types.2,4.

Epidermolysis bullosa simplex usually presents with milder forms of disease while junctional and dystrophic types may present with severe forms with multiorgan involvement.4,5.

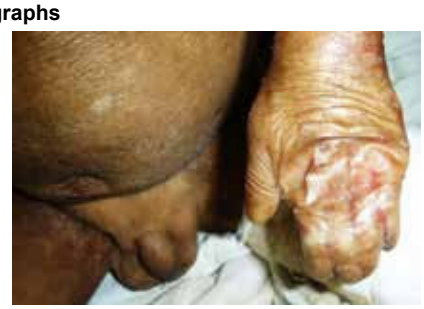
Skin biopsy, immunofluorescent microscopy and electron microscopy are the investigation usually employed for the diagnosis various clinico-histopathological variants (simplex, junctional, dystrophic) of epidermolysis bullosa. Prenatal diagnosis can be done reliably if genetic mutations are identified in family pedigree by DNA obtained through chorionic villus sampling around 9th week or by amniocentesis around 11th week of gestation.6,7.

Irrespective of clinical types, the management is directed towards avoiding precipitating events like trivial trauma; maintenance of barrier function, treatment of infections, promoting conditions for healing of erosions and ulcers and treatment of ocular, oral and gastrointestinal complications.

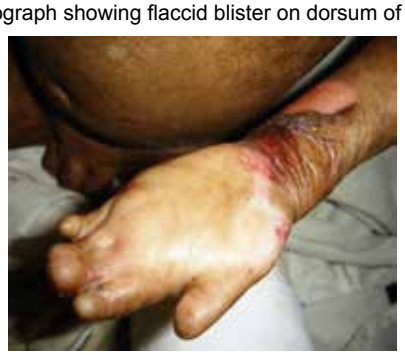
Conclusion

Epidermolysis bullosa encompasses a subset of bullous disorders encountered in infancy and early childhood with or without multisystem anomalies.Except for certain lethal variants(herlitz type of juctional variant and recessive dystrophic epidermolysis bullosa), most of the other variants are milder with minimal affection of life expectancy.Early diagnosis and treatment of the complications is warranted to prevent development of deformities.But it still is a grey area in which therapeutic alternatives are needed to be explored.

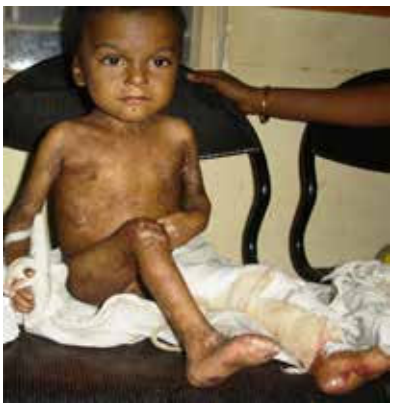
Photographs



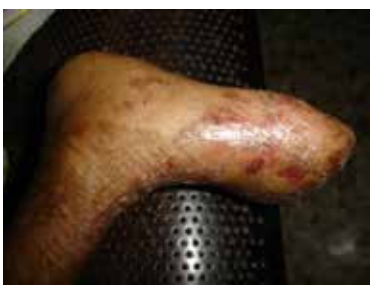
1. Photograph showing flaccid blister on dorsum of left hand

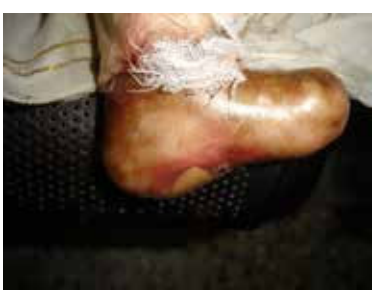


2. Photograph showing ruptured blister on medial side of flexor aspect of both forearms along with fused 3rd and 4th digits



3. Photograph showing areas of mottled dyspigmentation and atrophy of face, trunk and extremities





4 and 5.Photographs showing blisters of soles of both feet and loss of digits of both feet

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