ORIGINAL RESEARCH PAPER

Dermatology

KEY WORDS: Blisters, Dystrophic Epidermolysis bullosa, multidisciplinary approach, gene therapy.

DYSTROPHIC EPIDERMOLYSIS BULLOSA - CASE SERIES OF CRYSTAL SKIN CHILDREN

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ABSTRACT

Epidermolysis bullosa (EB) refers to a group of inherited disorders that involve the formation of blisters following trivial trauma. Here we report a case series of Dystrophic Epidermolysis bullosa(DEB) at our hospital presented with various clinical features. History ,clinical spectrum and histopathological examination gives appropriate diagnosis for this disorder. Treatment remains challange. A multidisciplinary approach is needed for the effective management of EB. Good nursing care, and gene therapy could possibly significantly alleviate the suffering of the patients in the future.

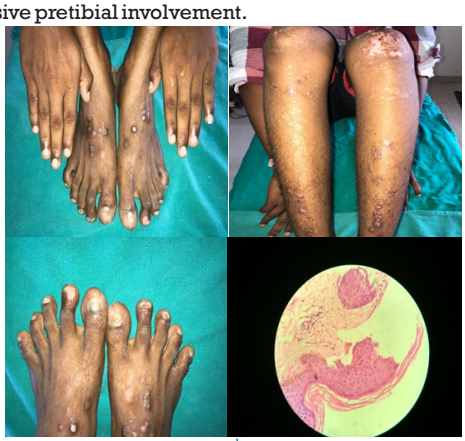
1.INTRODUCTION:

Epidermolysis bullosa (EB) 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. Based on level of cleavage they are classified into EB simplex (EBS), junctional EB (JEB), and dystrophic EB (DEB)[1].

2.CASE SERIES

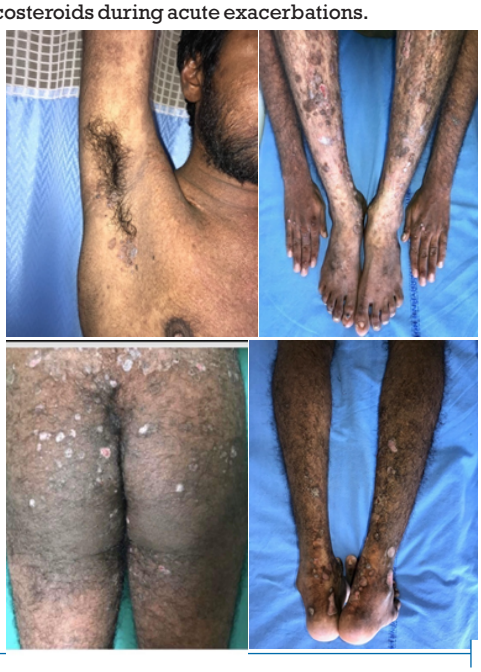
CASE 1:

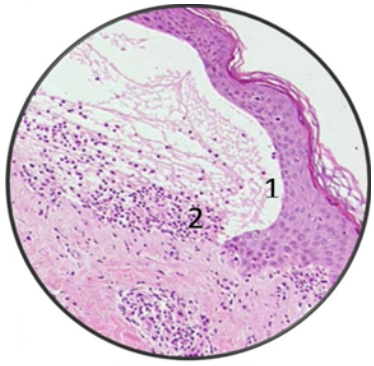
A 16-year-old born to non- consanguineously married couple presented with intensely itchy papules on bilateral upper and lower limbs from four months of age. Initially he had occasional pruritic fragile blisters on trivial trauma confined to shins which healed spontaneously with scarring. By the time he attained 10 years, he started developing milia. No history of atopy, food allergy, drug allergy, summer exacerbations and there were no similar complaints in the family. On cutaneous examination there were multiple lichenoid papules forming plaques at few areas, milia, scars, erosions, crusting and albopapuloid lesions present over bilateral upper and lowerlimbs more predominantly on shin region. Bullae were seen over the knee, toe nails were dystrophic. Mucosa, hair and teeth were normal. Nikolsky sign negative. Routine investigations and review of other systems were within normal limits. Direct immunofluorescence(DIF) showed negative result. On histopathological examination revealed subepidermal split and dermal inflammatory infiltrate. Hence according to history, clinical features, histopathology diagnosed as pretibial DEB as he had exclusive pretibial involvement.



CASE 2:

A 22years old male patient, born to a consanguineous couple presented with multiple episodes of fluid filled lesions with severe itching and burning sensation over trunk and limbs since birth .These lesions used to erupt spontaneously or followed by trivial trauma and healed with scarring. History of summer exacerbations, oral ulcerations, loss of nails were present with no history of photosensitivity. No h/o similar complaints in the family. On cutaneous examination ther were multiple skin colored papules, atrophic scars, vesicles and bullae measuring 0.5-2cm in diameter present in axillae, trunk, upper and lower limbs, few oral ulcerations with white lacy pattern on buccal mucosa and dystrophic nails were present. Nikolsky and Bulla spread sign shows negative result. Tzank smear showed inflammatory cells with no acantholytic cells. Routine investigations and review of other systems were with in normal limits. Direct immunofluorescence was negative. Histopathological examination showed a subepidermal blister with minimal inflammatory infiltrate. According to history, clinical features, histopathology diagnosed as Dystrophic Epidermolysis bullosa Inversa. Patient responded to oral Dapsone and systemic corticosteroids during acute exacerbations.

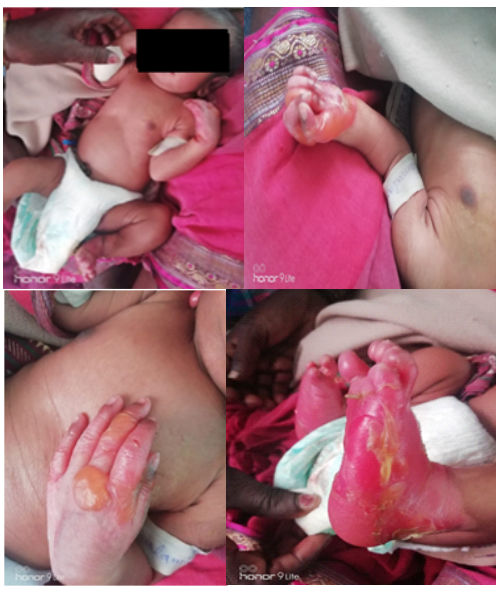


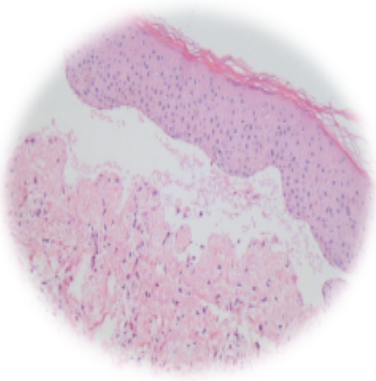


1.Subepidermal split
2.Dermal inflammatory Infiltrate

***CASE* 3:**

A 3month old female baby born by full term vaginal delivery to non- consanguineous couple was brought by parents with complaints of fluid filled lesions over the body since birth. No h/o similar complaints in the family. No h/o atopy, food allergy, drug allergy. On examination few bullae of sizes 4*4cms to 6*4cms with diffuse erythema present over dorsal and palmar aspect of both hands with few erosions present over both hands and plantar aspect of feet. Nails, oral cavity, scalp were normal. Nikolsky sign and DIF were negative. VDRL of mother negative. Histopathological examination showed Subepidermal split with inflammatory infiltrate and dermal edema. According to history, clinical features, histopathology diagnosed as DEB – Acral.





1. Sub epidermal split
2. Dermal inflammatory infiltrate

3.DISCUSSION

EB are classified into three subgroups on the basis of the level at which the skin separates. The three subgroups are - intraepidermal/epidermolytic (EB simplex), intra-lamina lucida/junctional (junctional EB), and sub-lamina densa/dermolytic (DEB).[1] DEB represents a group of inherited disorders that are characterized by defects in Type VII collagen and sub-lamina densa blisters. Type VII collagen is encoded by COL7A1 gene which is present on chromosome 3p21.[2],[3]. Avoidance of exacerbation 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. A key to triumphant management is good nursing care. Nursing the babies on thick foam pads protects them from trivial trauma induced blistering. Distinctive precautions need to be taken for older children while using of adhesive tapes, sphygmomanometer BP cuffs, tourniquets and other instruments that cause shearing of skin or mucous membranes. Further management needs to be commence in a multidisciplinary approach keeping in mind the multisystem involvement of the disease. The erosions should be wiped with sterile normal saline and covered with nonadherent dressings. Topical antibiotics are usually not preferred because of the risk of emergence of antibiotic resistant bacteria. Oral and dental hygiene should be intiated as early as tooth eruption begins[4] .Use of cyclosporine[5], thalidomide[6], dapsone[7] and topical tacrolimus[8] for the treatment of pruritus associated with the DEB-Pr can be beneficial .Advances in the treatment by transplantation of genetically modified epidermal stem cells and successful gene expression repair therap assure to change the treatment of EB.

4.CONCLUSION

Epidermolysis bullosa are a group of genetic blistering disorders associated with appreciable morbidity. Prevention of new blisters and wound management plays a crucial role. These disorders should be diagnosed early by pre-natal diagnostic tests. Genetic counselling helps in understanding the risks.

5.FUNDING

No funding sources

6.CONFLICTS OF INTEREST

None to declare

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