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A Case Series Of Rare Variants Of Epidermolysis Bullosa In A Tertiary Care Centre In Tamil Nadu.

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ABSTRACT

Epidermolysis bullosa (EB) is a heterogeneous group of genetic disorders that present with skin and sometimes with mucosal fragility, predisposing the affected individuals to the development of blisters and/or erosions after minimal trauma or friction[1]. Aim of study: To get a deeper understanding of the various clinical variants of Epidermolysis bullosa. It is a retrospective study based on the case history of 3 patients with rare variants of Epidermolysis bullosa. Our series comprises of 3 cases which came to the dermatology department of a tertiary care centre over a period of 8 months.

Keywords: Epidermolysis bullosa, Kindler syndrome, BART syndrome, Epidermolysis bullosa Ogna

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INTRODUCTION

Epidermolysis bullosa (EB) encompasses a group of rare, clinically and genetically heterogeneous genodermatoses with an estimated 500 000 cases worldwide [2]. There are three classic types of inherited EB (simplex, junctional and dystrophic). They are differentiated by the level of blister cleavage and subdivided according to the pattern of genetic inheritance, morphology/topography of lesions and genetic mutation involved [1]. The clinical presentation of inherited EB varies according to the type of disease, and diagnosis can only be reached by skin biopsy and immunofluorescence or electron microscopy, the latter being considered the gold standard [3]. Epidermolysis bullosa simplex (EBS) is characterized by a disorder of keratinocytes, intraepidermal blistering and little systemic involvement. Junctional epidermolysis bullosa (JEB) is an autosomal recessive disorder characterized by separation of the lamina lucida in the dermo-epidermal junction. Dystrophic epidermolysis bullosa (DEB) is due to mutations in the gene encoding type VII collagen, resulting in defective anchoring fibrils and consequent separation of the sub-basal lamina.

MATERIALS AND METHODS

Our series comprises of 3 cases which came to the dermatology department of a tertiary care centre over a period of 8 months.

It is a retrospective study based on the case history of 3 patients with rare variants of Epidermolysis bullosa.

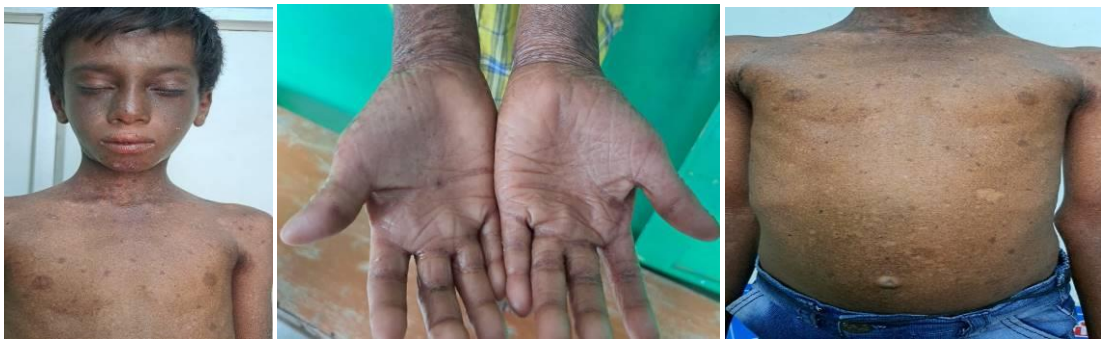
Case 1

A 10-year-old male child came to the op with complaints of history of blisters all over the body since birth. There is history of bleeding after trauma which was more frequent during early life. There is history of burning sensation when exposed to sunlight. He also complaints of anorexia. He is known case of bicuspid aortic valve and circumcision was done 2 years back for phimosis. He is the offspring of a non-consanguineous marriage with normal antenatal history. No history of similar complaints in the family.

On examination, he is moderately built and nourished. No pallor, icterus, clubbing, cyanosis or edema. Dermatological examination showed diffuse reticulate pigmentation with poikiloderma changes over face, trunk, back and extremities. Erosions over dorsal aspect of both foot and left hand. Post inflammatory hyperpigmentation all over the body +. scalp: normal, palms and soles: cigarette paper skin, ear: otitis media, oral mucosa: hemorrhagic gingiva, genital mucosa: normal.

All routine blood investigations were found to be normal except mild anemia. Peripheral smear showed moderate hypochromic anemia with neutrophilic leukocytosis. USG abdomen and pelvis showed no abnormalities. X Ray of long bones showed normal epiphysis. Skin biopsy showed epidermal atrophy and hyperkeratosis with flattened ridges and dermal lymphocytic infiltrate.

The patient was managed conservatively and was asked for regular follow-up.



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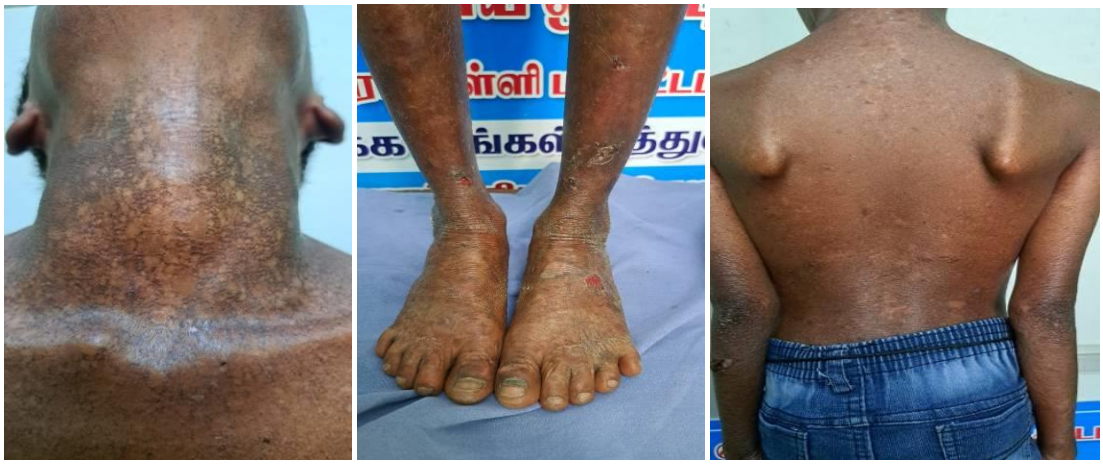
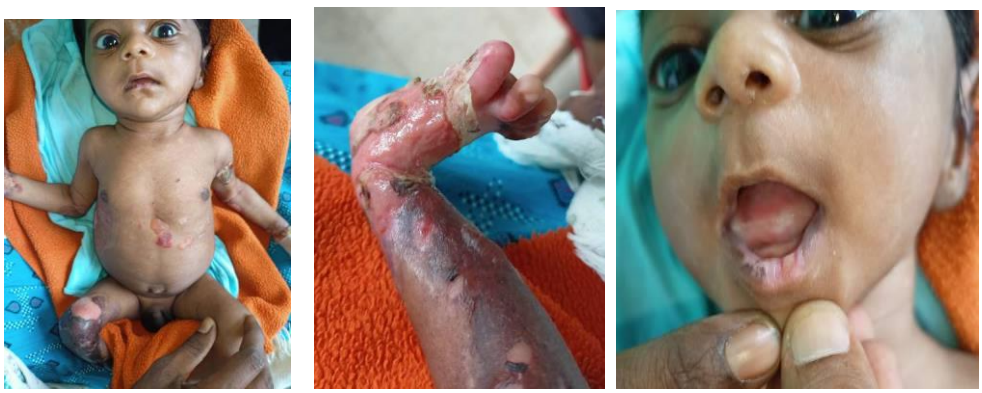


Case 2

A 2 month old male child was brought by parents with complaints of absence of skin over right great toe and dorsal aspect of both foot. On the sixth day of his life, he developed blisters over trunk, extremities and lastly over the lips which later formed erosions. There is history of blistering following handling of the baby. He is a baby of a non-consanguineous marriage with normal antenatal history. No history of similar complaints in the family.

Dermatological examination revealed multiple tense fluid filled and hemorrhagic bullae present over the chest and upper limbs. Absence of epidermis over right great toe and dorsal aspect of both foot. Diffuse erosions with crusting present over neck, trunk , bilateral upper and lower extremities. Post hypo and hyper pigmentation present over lips, neck, trunk, upper and lower extremities. Scalp : normal, palms: normal, soles: erosions + ; hair and nails : normal; oral cavity : erosions over lips +, buccal mucosa normal; genital mucosa : normal.



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Routine blood investigations were found to be within normal limits. An USG abdomen and pelvis was taken to rule out pyloric stenosis and an ECHO was taken to rule out cardiomyopathy and both were found to be normal.

The baby was managed conservatively. Wound care was given with topical antibiotics. Parents were educated about avoiding any form of trauma and regular followup.

Case 3

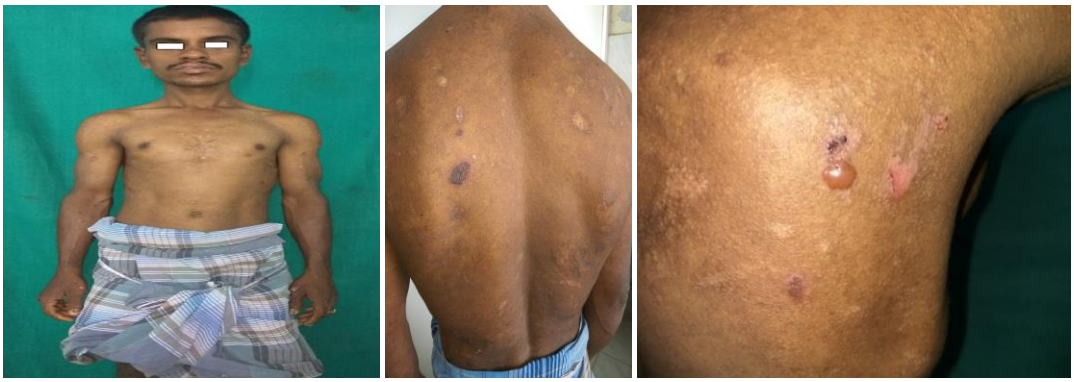
A 27-year-old male came to the OPD with complaints of history of recurrent blisters following trivial trauma since birth. He had history of spontaneous improvement of skin lesions. There is no history of pain , pruritis, oral lesions, photosensitivity, hoarseness of voice or any muscle weakness. He has no other known comorbidity. He is a child of second-degree consanguineous marriage with an uneventful antenatal and postnatal history. There is history of similar complaints in his mother, elder sister and twin brother.

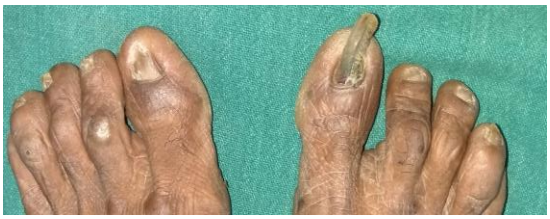
On examination, he is moderately built and nourished, short stature.

Dermatological examination revealed multiple tense clear fluid filled blisters of size 0.5 *0.5 cm to 1.5 * 1.5cms seen in the dorsum of both hands, upper back, medial aspect of arm. Newer blisters are tense filled with clear fluid and older blisters – flaccid contain hemorrhagic fluid. Erosions of various size seen in the face, upper chest & dorsum of hand. Multiple hyper & hypopigmented skin lesions. No scarring & milia. Onychogryphotic great toe nail.

Histopathology showed subepidermal blisters.

On immunofluorescence antigen mapping Collagen IV mapped in the floor of the blisters. There was a normal intensity of collagen VII, laminin 332 and cytokeratin 14. He was managed conservatively.





DISCUSSION

Kindler syndrome is a rare hereditary disorder, which was first described in a 14-year-old girl by Kindler in 1954 [4]. It presents with blistering in the early years of life, poikiloderma and photosensitivity. The gene responsible is KIND I gene, which encodes for kindlin-1 protein, found on the chromosome number 20. Kindlin regulates the over secretion of basement membrane components by basal keratinocytes associated with disruption of the basement membrane and abnormal deposition of type VII collagen both in regions with active lesions and in lesion-free areas [1]. Apart from the skin changes,

changes in the oral and conjunctival mucosa, phimosis and radiological changes, namely a dome-shaped skull (turricephaly), rib and mandibular abnormalities have been reported [5]. Treatment is mainly conservative with proper sun protection which could delay the onset of poikiloderma. The patient will have a normal lifespan but complications like secondary infections can occur.

Bart syndrome is a rare inherited disorder characterized by the localized absence of the skin, blister formation, and nail deformity and is considered a variant of aplasia cutis congenita (ACC) with epidermolysis bullosa (EB) [6]. The most common site to be involved in Aplasia cutis congenita is the scalp.

Group	Associations
I	Scalp ACC without multiple abnormalities
II	Scalp ACC with limb abnormalities, hypospastic or absent distal phalanges, syndactyly, club foot, others
III	Scalp ACC with epidermal and sebaceous nevi
IV	ACC overlying embryologic malformations such as gastroshisis, omphalocele, meningomyelocele, and others
V	ACC with fetus papyraceus or placental infarction
VI	ACC associated with epidermolysis bullosa (extremities and torso)
VII	ACC limited to extremities without epidermolysis bullosa
VIII	ACC due to teratogens such as methimazole (scalp), varicella and herpes simplex infections (any area)
IX	ACC associated with syndromes of malformations such as Goltz Syndrome, trisomy 13, ectodermal dysplasia, and others

Table 1: Friedens classification of ACC [7].

Epidermolysis bullosa simplex ogna is an autosomal dominant blistering disorder due to the defect in plectin protein present in the basal keratinocytes which is coded by PLEC1 gene on chromosome 8q24 . Most commonly it will be a misn sense mutation. It is characterized by generalized bruising tendency, hemorrhagic bullae and onychogryphotic great toe nail. The clinical picture will be mild with lesions healing with post inflammatory pigmentation.

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