ORIGINAL RESEARCH PAPER

Dermatology

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EPIDERMOLYSIS BULLOSA DYSTROPHICA WITH RENAL FAILURE

A. Bhavana* Postgraduate, Department of dermatology, Narayana medical college and hospital, Nellore, Andhra Pradesh, India. *Corresponding Author

N. Chakravarthy Assistant professor, Department of dermatology, Narayana medical college and hospital, Nellore, Andhra Pradesh, India.

K. Uday kiran raja Assistant professor, Department of dermatology, Narayana medical college and hospital, Nellore, Andhra Pradesh, India.

V. Sushitha Postgraduate, Department of dermatology, Narayana medical college and hospital, Nellore, Andhra Pradesh, India.

A. Vijaya Mohan Rao Professor and HOD, Department of dermatology, Narayana medical college and hospital, Nellore, Andhra Pradesh, India.

ABSTRACT

Inherited epidermolysis bullosa (EB) is a family of diseases with blistering of skin and mucous membrane even with mild trauma. It has different subtypes based on the affected protein. Epidermolysis bullosa dystrophica (EBD) is associated with mutations in the COL7A1 gene encoding type VII collagen leading to the fragility of skin and mucosal membranes. EBD may be dominant or recessive. They should be distinguished because increased prevalence of SCC is associated with the recessive form. Here we are reporting a case at our hospital presented with various clinical features, history, clinical spectrum, renal biopsy and gene analysis giving appropriate diagnosis for the disorder. Treatment remains challenge. A multidisciplinary approach is needed for the effective management of EBD.

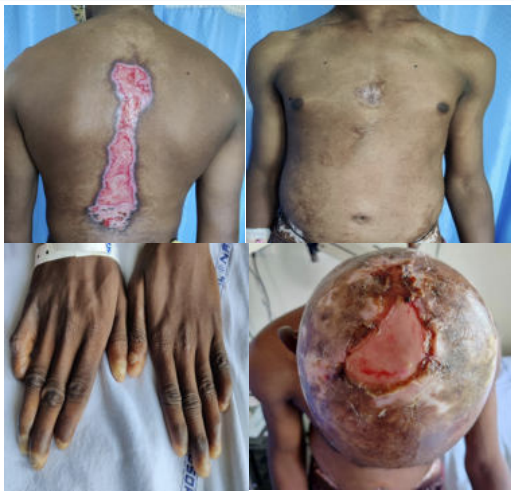
INTRODUCTION:

Epidermolysis bullosa(EB) is heterogenous group of inherited mechanobullous disorders, clinically characterized by development of blisters over skin and mucous membranes following minor frictional trauma. There are four major forms of epidermolysis bullosa – simplex(EBS), junctional(JEB), dystrophic(DEB) and mixed type based on level of cleavage at dermo-epidermal basement zone. In epidermolysis bullosa dystrophica (EBD), blistering occurs in the sublamina densa.

Case Report:

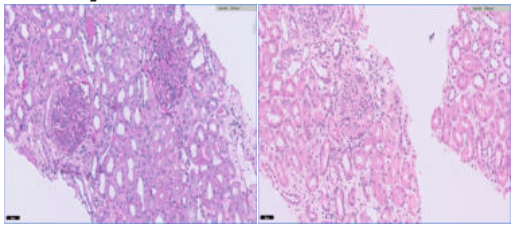
A 22-year-old male patient born of second degree consanguinous marriage with no significant family history born through uncomplicated normal vaginal delivery presented with complaint of wounds over scalp and back since 1 month. History of fluid filled lesions all over the body since birth, healing with scarring. History of loss of nails, body hair and scalp hair since childhood. On examination: Ulcers of sizes 15x6cm to 40x10 cm seen over scalp and posterior trunk with granulation tissue, peripheral atrophic scarring; Depigmented areas with mottled pigmentation seen over scalp, anterior chest, groins, axillae and wrists. Scalp shows cicatricial alopecia, palmar keratoderma present. Nails- Total loss of finger and toe nails. Oral examination shows reduced mouth opening and dental caries present (molars and premolars).

Investigations revealed anemia, deranged renal parameters (serum urea-128mg/dl, serum creatinine-1.84mg/dl), and complete urine examination showed plenty of RBCs, albumin 3+ and nephrology opinion was taken in view of these deranged parameters.



Renal Biopsy:

It showed infection related diffuse proliferative exudative glomerulonephritis.



Gene Analysis:

Homozygous missense variant and homozygous 87bp deletion variant in the COL7A1 gene were identifies by exome sequencing analysis.

DISCUSSION:

- Dystrophic Epidermolysis Bullosa (DEB) is inherited either dominantly or recessively having various subtypes in each.
- Dominant DEB (DDEB) - generalized, acral, pretibial, pruriginosa, nails only and bullous dermolysis of

newborn
- Recessive DEB (RDEB) - severe generalized, generalized other, inversa, pretibial, pruriginosa, centripetalis, bullous dermolysis of newborn.
- Dominant is milder than the recessive form.

Clinically DDEB presents with bullae that heal with scarring and milia, nail dystrophy. Nails can be thickened with discoloration of nail plate or may be totally absent. RDEB- this may be severe generalised or milder subtype.

Blistering is very severe including mucosa and nails, associated with scarring alopecia, flexion contracture of the fingers and toes resulting in syndactyly.

Extracutaneous manifestations- anemia, growth retardation, widespread involvement of GIT, Genitourinary tract, respiratory tract and ocular. The diagnosis of EB is mainly clinical. Management is multidisciplinary, prevention of new blister, treating and preventing infections, wound care, management of extra cutaneous complications and preserving function, nutritional support and psychological support to the patient and family members. As these patients present with chronic recurrent ulcers, complications like secondary bacterial infection is common leading to systemic complications like post streptococcal glomerulonephritis. Other complications include skin dysfunction that leads to nutritional deficiencies like vitamin D3, protein loss; secondary amyloidosis. Bone marrow transplantation after myeloablation can be done.

CONCLUSION:

DEB is caused by collagen VII defect, clinical findings and gene analysis support dominant form of dystrophic epidermolysis bullosa (DDEB). Protective bandaging, prevention of mechanical trauma, loose fitting clothes, regular dressings can be done. Genetic counselling plays a preventive role.

Conflicts Of Interest: NONE.

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