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EPIDERMOLYSIS BULLOSA DYSTROPHICA - A CASE REPORT

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ABSTRACT

A series of hereditary illnesses known as epidermolysis bullosa (EB) are characterised by deteriorated dermo-epidermal integrity, blistering, and skin erosions caused by gene COL7A1 mutations. EBD-causing mutations can either be recessive or dominant. Mutations of type VII collagen, the main component of the anchoring fibrils, have been reported in recessive variants of epidermolysis bullosa dystrophica. mucous membranes following minor injury. In decreasing order of occurrence, EBD is distinguished by flat, pink, scar-producing bullae on the ankles, knees, hands, elbows, and feet. Only a few case reports of EBD have been documented in the literature. In this case report, the use of a symptoms healing treatment was evaluated in a 56 year old male patient. These disease relatively accounting as a rare disorders (incidence:1/50000 live births to 1/500000 live births).There is currently no effective treatment available; instead, operations are used to manage side effects and symptomatic relief.

Keywords: Epidermolysis bullosa dystrophica, Skin lesion, Erosions, Hyperpigmentation, Dominant, Resessive

INTRODUCTION:

EBD is a heterogenous is an inherited disease characterized by blister formation and erosions with hyperpigmentation all over the skin and mucous membrane. The signs and symptoms of dystrophic epidermolysis bullosa vary widely among

affected individuals. Currently EB is classified into four major types according to the location of the lesions: epidermolysis bullosa simplex in the epidermis, junctional EB within lamina lucida of the skin base membrane, dystrophic EB in the uppermost dermis, and Klinder syndrome in multiple layers of the skin **[1]**. A Uncommon hereditary skin condition called EPIDERMOLYSIS BULLOSA DYSTROPHICA (EBD) is brought on by mutations in the gene COL7A1, which codes for collagen type VII. Clinical signs of COL7A1 mutation include nail dystrophy, minor localised blistering, and extensive skin blistering **[2]**. Mutations in the COL7A1 gene, which is found in the short arm of the third chromosome and codes for a chain of VII type collagen, a component of the anchoring fibrils at the dermal-epidermal junction, are the primary cause of epidermolysis bullosa dystrophica. Scar tissue after wound healing is frequently visible since the lesion lies below the cutaneous basement membrane **[3]**.

EBD subdivided into mainly other two type dominant dystrophic epidermolysis bullosa (DDEB)and recessive dystrophic epidermolysis bullosa (RDEB) which also known as the "Cockayne-Touraine disease"and "Hallopeau–Siemens disease The current course of treatment for recessive dystrophic epidermolysis bullosa includes supportive care for the skin and other organ systems, including wound management, infection support for chronic wounds, surgical management as necessary, nutritional support, and preventative screening for squamous cell carcinoma. In the literature, there have been 21 recorded cases; we are adding additional cases and providing more background on one of those cases.

Case Report:

56 year of male patient reported complaint of the skin lesion over B/L lower limb, upper limb, since last one month he also present with multiple designed erosions over chest, neck, back. Clinical examination revealed on elaboration of origin, duration and progression patient was relatively alright before one month then patient begin to developed skin lesions characterized by thickening of skin, later increasing in a size in over a limb F/B peeling of skin over upper and lower limb seeing behind raw areas that showed later tendency to heal. B/L upper limb and lower limb lesion developed bleeding point.as per the cutaneous examination showing erosions with hyperpigmented rim present in all over neck, chest, B/L LL, B/L UL, back, buttocks. Development of itching of these lesion has mild to moderate and informetent. By increasing severity in night these lesion were associated with mild to moderate pain. The patient was diagnosed

with epidermolysis bullosa dystrophica. inversa due to his clinical and histological findings, absence of specific immune accumulation. He had no any family history with similar compalaints.his past medical history showed similar complaint of B/L upper and lower limb skin lesion developed severe bleeding with untolerable pain. due to blood loss PCV blood transfusion was done. His finger and toenails were absent.

EBD (epidermolysis bullosa dystrophica) is incurable and the treatment is palliative and non-specific. PRP (platlate rich plasma) two sitting has been done using injection of patient on plasma at the injuries site of the skin lesion. Symptomatic treatment of intertriginous lesons included local application of 0.01% betamethasone cream and white soft paraffin which can be reliefable. Oral treatment was started with capsule thalidomide 100 mg BD and tab prednisolone 5mg with milk in the morning.

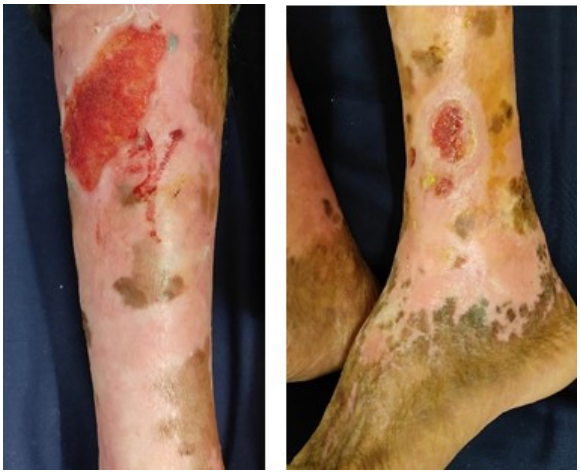

Figure 1: Skin lesions on foot

DISCUSSION:

We are present the case of patient with EBD characterized by blister formation and erosions with hyperpigmentesion all over the skin and mucous membrane. Dystrophic epidermolysis bullosa which was identified by Gedde and Dahl for the first time in 1971 **[4]**. This group is classified in to four types of EB: EB simplex, junctional EB, DEB, and Klinder syndrome. The gene COL7A1, which codes for α chain of VII type collagen, a component of the anchoring fibrils at the dermal-epidermal junction, and is situated in the short arm of the third chromosome, is the primary genetic cause of EBD-epidermolysis bullosa dystrophica. While junctional and dystrophic varieties of epidermolysis bullosa can present with severe forms with multiorgan involvement, the illness often manifests in milder forms in epidermolysis bullosa simplex **[5-6]**.

EBD further subdivided in mainy two type Dominant dystrophic epidermolysis bullosa (DDEB)Also known as "Cockayne-Touraine disease", this variant is characterized by vesicles and bulle on the extensor surfaces of the extremities and *Recessive* dystrophic epidermolysis bullosa (RDEB) **[7]**. Also known as "Hallopeau–Siemens variant of epidermolysis bullosa" and "Hallopeau–Siemens disease", this variant results from mutations in the gene encoding type VII collagen, COL7A1, characterized by debilitating oral lesions that produce pain, scarring, and microstomia **[8]**. In comparison to other EB forms, Dominant Dystrophic Epidermolysis Bullosa (DDEB) exhibits less expression of type VII collagen. In RDEB, blistering can start spontaneously and develop soon after delivery. The most common areas affected include the eyes, teeth, mouth, and oesophagus, as well as skin blistering **[9]**. The RDEB HS subtype has the highest risk (26%). As was already established, there is currently no effective treatment or cure for EB. However, over the past ten years, enormous strides have been made in the development of novel treatments, including gene therapy, protein replacement therapy, cell therapies, the culture of revertant mosaic keratinocytes, gene engineering, and the use of inducible pluripotent stem cells in clinical settings **[10-11]**. Different phenotypes of hereditary EBD exist depending on which layer of the skin is affected by the pathological process. It is significant to determine the site of the mutation since, even for the same type of EBD, the clinical characteristics might range widely. This makes it possible to predict how the disease will progress and carry out sensible precautions before problems are identify.

CONCLUSION:

EBD causes depend on which genes are involved in the process .there is no any particular cure available for EBD.however certain therapy such as protein replacement therapy,gene therapy ,cell therapy has been developed for symptomatic treatment.

CONFLICT OF INTEREST:

The author declares no conflict of interest.

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