International Journal Dental and Medical Sciences Research
Volume 5, Issue 3, May - June 2023 pp 480-481 www.ijdmsrjournal.com ISSN: 2582-6018

A Story of Rare Genetic Disease in a Poor Family

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Date of Submission: 01-06-2023 Date of Acceptance: 10-06-2023

INTRODUCTION

Dystrophic EB (DEB) is transmitted in either an autosomal dominant or autosomal recessive manner and is caused by mutations in the type VII collagen gene. Dominant DEB (DDEB) results from dominant-negative mutations, typically a missense mutation that leads to substitution ofanother amino acid for a glycine within the triple-helical domain of this collagen. Although the resultant protein is structurally abnormal, immunohistochemical staining of the dermal–epidermal junction is usually indistinguishable from that of normal skin.

CASE PRESENTATION

A 6 years old male child brought by the parents to the hospital with chief compliants of blistering all over body since birth ,raw areas all over body , blistering near the areas of friction , during birth loss of skin over both ankles is seen which later seen over both legs and abdomen ,both hands ,back of the chest

Patient attended pvt hospital during birth where the child was told as genetic disease ,no evidence of treatment and biopsy present near the patient attender.h/o consaguinous marriage present h/0 similar complaints in childssibling who died at 4 years of age ,no h/o of similar complaints in parents and other family members

0/e child has erythematous rash all over body both upper limbs and lower limbs flexures are more involved ,scarring present over both legs.mitten leg deformity is seen, oral mucosal erosions present.blister over abdomen present ,skin over both hands is fragile , skin eroded if pressure is applied

Scalp is normal

Hair is normal

Nails of both legs are lost.syndactaly is seen

Hypoplastic nails of hands are seen

Genital area is normal

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