GENETICS AND GENODERMATOSES

FINDING 3 VARIANTS NOT REPORTED IN GENE COL7A1 IN PATIENTS WITH EPIDERMOLYSIS BULLOSA RECESSIVE DYSTROPHIC.

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Dystrophic epidermolysis Bullosa(EBD) is a genodermatosis characterized by skin fragility that is evidenced by the appearance of erosions and blisters on the skin and mucous membranes at the slightest touch or trauma. Transmission is autosomal recessive or dominant. More than 700 mutationsa ssociated with this disease have been described, which are found within the COL7A1 gene that encodes protein collagen VII. In this work, we present 8 patients belonging to 6 unrelated families. In each case, DNA was extracted from peripheral blood, and variants in the COL7A1 gene were determined by Sanger sequencing. These variants were then analyzed in healthy relatives. We found 3 variants not reported in literature (c.7243_7244insT, C.7760delG, C.7104 + 3A> T), The 5 patients in which the variant c.7243_7244insT was found were from Paraguay Thanks to the analysis of the COL7A1 gene it was It is possible to confirm the clinical diagnosis and also allow the genetic counseling of patients and their families.