GENETICS AND GENODERMATOSES

EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED INTERMEDIATE: A FAMILY REPORT

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Background: Epidermolysis Bullosa Simplex (EBS) generalized intermediate (EBS-gen intermed; previously known as Koebner type), inherited in an autosomal dominant manner, is one of the four most common subtypes of EBS. The molecular basis of this genetic condition resides in defects of cytokeratines 5 (KRT5 gene) and 14 (KRT14 gene) with alterations in the assembly, structure and / or function of the intermediate skeleton of keratin filaments of basal keratinocytes. The result of these genetic defects is blistering above the dermal-epidermal junction, especially on hands and feet.

Observation: The family reported in this article presented one affected individual in three subsequent generations, demonstrating the autosomal dominant inheritance of the pathology; as matter of fact, each child of an affected individual has a 50% chance of inheriting the pathogenic variant and having EBS. Genetic molecular test and a skin biopsy with histological examination and immunofluorescence antigen mapping (IFM) were performed in all affected individuals of the family. Genetic analysis identified a heterozygous mutation in the KRT5 gene (12q12-q13) which presented a substitution from Valine (V) to Alanine (A) at position 323 (p. Val323Ala), responsible for the disease.

Key message: This is a rare disease and no case has been reported from southern Italy till date. The skin biopsy and the genetic analysis with IFM are two fundamental steps to diagnose Epidermolysis Bullosa Simplex and the specific subtype, as demonstrated by the cases reported herein.