GENETICS AND GENODERMATOSES

ANALYSIS OF COL7A1 GENE MUTATIONS IN 6 CHINESE FAMILIES WITH DYSTROPHIC EPIDERMOLYSIS BULLOSA

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Background: Dystrophic epidermolysis bullosa (DEB) is a heterogeneous hereditary skin disorder caused by mutations in the COL7A1 gene encoding type VII collagen. Molecular diagnostics and genotype-phenotype correlations in DEB remain complex owing to the large variety of mutations, high rate of novel mutations, complex protein structure and heterogeneity of phenotypes.

Observation: We detected mutations of the COL7A1 gene in 6 Chinese families with dystrophic epidermolysis bullosa, including two DDEB families (One has eighteen and the other has five affected members), three RDEB families and one epidermolysis bullosa pruriginosa family. There were two recurrent missense mutations (p. Ala2054Val and p. Gly2009Ala) from two DDEB families, which are located very close. The patients in both families had mild localized symptoms and only presented with small milia and mild erythema on the fingers and toes, or only nails were involved. Although partial patients had a transient history of blisters in newborn, there was no blisters later. It suggested that mutations in this site may have a relatively small effect on gene function leading to mild phenotype. In RDEB families, compound heterozygous mutations (p. Gly150Arg and p. Arg2610Ter) inherited from the parents in one patient and novel mutations (p. Thr206HisfsTer19 and c. 7381-9G>A) in the other patient both presented with localized clinical symptoms. The other patient carried truncation mutations (p. Val168GlyfsTer13 and novel p. Lys1733ArgfsTer108) with generalized intermediate symptoms and right eye was blind. Compound heterozygous mutations (p. Arg2685Ter and novel c.1241-6G>A) was found in the EBP family, and the patient presented with severe itching, prurigo-like or lichenoid nodules, milias on the shins and auricle.

Key message: Our study report 6 families with DEB, who had four novel and six previously reported mutations in COL7A1 and helps to disclose the underlying correlations between the phenotype and genotype of DEB.