GENETICS AND GENODERMATOSES

SIXTEEN NOVEL COL7A1 MUTATIONS IDENTIFIED IN PATIENTS WITH DYSTROPHIC EPIDERMOLYSIS BULLOSA FROM CHINA: A RETROSPECTIVE STUDY FROM A SINGLE CENTER IN SHANGHAI, CHINA (2013-2018)

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Background: Dystrophic epidermolysis bullosa is a group of genetic conditions that cause the skin to be fragile and to blister easily. COL7A1 mutations alter the structure or disrupt the production of type VII collagen.

Objective: The aim of this study was to explore the potential of mutation spectrum in dystrophic epidermolysis bullosa (DEB) in China.

Materials and Methods: Whole exome sequencing was performed using genomic DNA from each case of dystrophic epidermolysis bullosa, followed by massively parallel sequencing. Resulting reads were mapped to the human reference genome hg19. Sanger sequencing confirmed the potentially pathogenic mutations subsequently.

Results: In this study, A total of 32 distinct mutations in COL7A1 were identified in 17 Chinese families who were diagnosed based on clinical presentation, 16 of them being previously unreported, 5 of novel mutation being gross indels. None recurrent mutation was identified. Approximately 93.7% of unique variants appeared only once in the population assessed, with the frequent mutation c.7474C>T appearing twice in unrelated individuals (2/32, 15%).

Conclusion: Frequent mutation c.7474C>T is recurrent in populations among our center, most families carry unique mutations in China.