Clinical Genetics

AB021. An observational study to identify keratin 5 and 14 mutations in patients with epidermolysis bullosa simplex in MacKay Memorial Hospital, Taipei, Taiwan

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Background: Epidermolysis bullosa simplex (EBS) is a rare inherited skin disorder which is characterized by fragility of the skin, non-scarring blisters and erosions due to minor mechanical trauma. EBS is usually inherited as an autosomal dominant condition. EBS is commonly caused by a mutation in either the *KRT5* or *KRT14* genes, and less commonly by mutation(s) in *PLECTIN*. This study is designed to establish a mutation database and phenotype-genotype correlation of patients with EBS for further clinical research and development.

Methods: Patients with a suspected diagnosis of EBS based on clinical findings were enrolled. Skin biopsies were first performed as a screening method before mutation analysis was completed. Genomic DNA will be analyzed by next generation sequencing (NGS) technique.

Results: In 2016, a total of 13 suspected EBS patients were recruited. Nine were consistent with EBS by electron microscopy, while the others were compatible with a dystrophic type of EB. NGS analysis was done on samples from 9 EBS patients. One patient carried a *PLECTIN* mutation (c.194-1G>C; c.6991C>T), one carried a *KRT14* mutation (c.373C>T) Three patient carried a pathogenic mutation (c.504G>C), and four had variants of uncertain significance (VUS) in *KRT5* gene. Six EBS patients belonged to the generalized (Dowling-Meara type), and three belonged to the localized (Weber-Cockayne type).

Conclusions: EBS is a heterogeneous group of disorders with blister formation. In our study, skin biopsies were obtained as a screening method and then mutational analysis was performed. However, skin biopsies are invasive and time-consuming, and may not be ideal or feasible in some emergency cases. The EBS-targeted NGS sequencing is a fast, minimally invasive, and promising approach for these conditions. Our study also highlights the need to consider VUSs in patients undergoing EBS-targeted NGS to ensure that the needs of patients as well as their families are met.

Keywords: Epidermolysis bullosa simplex (EBS); autosomal dominant; mutation analysis; next generation sequencing (NGS); variants of uncertain significant

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