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Case Report

Novel and recurrent *COL7A1* mutations in Chinese patients with dystrophic epidermolysis bullosa pruriginosa

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ABSTRACT. Dystrophic epidermolysis bullosa pruriginosa (DEB-Pr) is a rare subtype of dystrophic epidermolysis bullosa (DEB). This disease is characterized by severe itching, lichenoid nodules or prurigo-like lesions, and linear scarring with a predilection for the extensor limbs. Pathogenic mutations in the type VII collagen alpha 1 (*COL7A1*) gene have been identified. We analyzed mutations in the *COL7A1* gene in a Chinese family including 5 affected individuals with typical DEB-Pr and in a patient previously reported with sporadic DEB-Pr. The entire coding region and exon-intron boundaries of *COL7A1* were detected by polymerase chain reaction and direct sequencing. We identified one novel heterozygote mutation (c.6842G>T, p.G2281V) and a second mutation (c.5443G>A, p.G1815R) reported previously in patients with DEB. Our findings contribute to the *COL7A1* mutation database and further reveal the genetic and phenotypic heterogeneity of DEB-Pr.

Key words: Dystrophic epidermolysis bullosa pruriginosa; *COL7A1* gene; Mutation analysis

