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A novel insertion mutation in the *ADAR1* gene of a Chinese family with dyschromatosis symmetrica hereditaria

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ABSTRACT. Dyschromatosis symmetrica hereditaria (DSH) is an autosomal dominant pigmentary genodermatosis, characterized by a mixture of hyperpigmented and hypopigmented macules that are mainly present on the dorsal portions of the extremities. The DSH locus was mapped to chromosome 1q11-q12 and, subsequently, pathogenic mutations in the double-stranded RNA-specific adenosine deaminase (*ADAR1*) gene were identified. We performed a mutational analysis of the *ADAR1* gene in a Chinese family that included three individuals affected with typical DSH phenotypes. Mutations within the entire coding region and the exon-intron boundaries of *ADAR1* were detected and confirmed by polymerase chain reaction and direct sequencing, respectively. An insertion mutation within exon 12, c.3035_3036insC (p.P1012fsX1017), was identified in all family members affected by DSH, but not in the healthy members or 100 unrelated controls. This finding improves our understanding of the role of *ADAR1* in DSH.

Key words: Dyschromatosis symmetrica hereditaria; *ADAR1*; Mutation

