Mutations in the ADAR1 gene in Chinese families with dyschromatosis symmetrica hereditaria

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ABSTRACT. We investigated 2 Chinese families with dyschromatosis symmetrica hereditaria (DSH) and search for mutations in the adenosine deaminase acting on RNA1 (ADAR1) gene in these 2 pedigrees. We performed a mutation analysis of the ADAR1 gene in 2 Chinese families with DSH and reviewed all articles published regarding ADAR1 mutations reported since 2003 by using PubMed. By direct sequencing, a 2-nucleotide AG deletion, 2099-2100delAG, was found in family 1, and a C→T mutation was identified at nucleotide 1420 that changed codon 474 from arginine to a translational termination codon in family 2. Two different pathogenic mutations were identified, c.2099-2100delAG and c.1420C>T, the former being a novel mutation, and the latter previously reported in 3 other families with DSH. To date, a total of 110 mutations in the ADAR1 gene have been reported, and 10
of them were recurrent; the mutations R474X, R1083C, R1096X, and R1155W might be the DSH-related hotspots.

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