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Common ABCB1 polymorphisms associated with susceptibility to infantile spasms in the Chinese Han population

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ABSTRACT. Infantile spasms are a severe epileptic encephalopathy with a variety of etiologies that occur in infancy and early childhood. Subjects with infantile spasms are at a higher risk for evolving into intractable epileptic spasms, tending to be refractory to conventional antiepileptic drugs. Genetic polymorphisms of the P-glycoprotein-encoding gene ABCB1 are suspected to be associated with pharmacoresistance phenotypes in epilepsy patients. Conflicting findings have been reported in different populations; few studies have explored whether this apparent association is affected by other host factors, such as specific epilepsy syndrome. We performed a case-control study to determine whether the risk of infantile spasms is influenced by common ABCB1 polymorphisms in a Han Chinese children's population consisting of 91 patients and 368 healthy individuals. DNA was isolated from whole blood, and three genetic polymorphisms (C1236T, G2677T/A,

and C3435T) were assayed by PCR-RFLP. There were significant differences in the distributions of 3435TT [P = 0.001; odds ratio = 2.47; 95% confidence interval (CI) = 1.44-4.27] and 3435CT [P < 0.001; odds ratio = 0.28; 95%CI = 0.15-0.54] genotypes between infantile spasm cases and controls. No significant differences were observed in allelic and haplotypic frequencies of ABCB1 polymorphisms between the two groups. This study demonstrated that variations in the C3435T gene play an important role in the pathogenesis of infantile spasms in the Han Chinese population; 3435TT is associated with increased risk of having this epilepsy syndrome.

Key words: Infantile spasms; ABCB1 gene; Genetic polymorphisms

