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Analysis of MTR and MTRR Gene Polymorphisms in Chinese Patients With Ventricular Septal Defect.

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Abstract

BACKGROUND: Congenital heart defects (CHDs) are the most common birth defects and ventricular septal defects (VSDs) are one of the most common types of CHDs. Genes involved in homocysteine/folate metabolism may play important roles in CHDs. Methionine synthase and methionine synthase reductase (MTRR) are key regulatory enzymes involved in the metabolic pathway of homocysteine.

METHODS: We investigated whether a polymorphism (A2756G) of the methionine synthase and 2 polymorphisms (A66G and C524T) of the MTRR gene are associated with VSDs. A total of 183 children with VSDs and 201 healthy children were studied.

RESULTS: The polymorphisms were detected by polymerase chain reaction amplification and sequencing of the amplified product. Significant differences in the distributions of the A66G and C524T alleles were observed between VSD cases and controls, and a slightly increased risk of VSDs was associated with either of the 66AG, 524CT, and 524TT genotypes [odds ratios (OR)=1.796, 1.909, and 2.088, respectively]. The genotype frequency of 66AG in VSDs patients was significantly different from those of controls (ORs=3.147). In addition, the combined 66AG/524CT and 66GG/524TT in VSDs had ORs 2.937 and 5.344, respectively.

CONCLUSIONS: MTRR A66G and C524T polymorphisms are associated with increased risk of VSDs.

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