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A66G and C524T polymorphisms of the methionine synthase reductase gene are associated with congenital heart defects in the Chinese Han population

W. Zeng[1], L. Liu[1], Y. Tong[1], H.M. Liu[1,2], L. Dai[3] and M. Mao[1]

[1]The Laboratory of Early Development and Injuries,
Department of Pediatrics, West China Second University Hospital,
Sichuan University, Chengdu, China
[2]Department of Pediatrics, The Pulmonary Vascular Remodeling Research Unit,
West China Second University Hospital, Sichuan University, Chengdu, China
[3]The Laboratory of Molecular Epidemiology of Birth Defects,
West China Second University Hospital, Sichuan University, Chengdu, China

Corresponding author: M. Mao
E-mail: dffmmao@126.com

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ABSTRACT. Congenital heart defects (CHDs) are the most common birth defects; genes involved in homocysteine/folate metabolism may play important roles in CHDs. Methionine synthase reductase (*MTRR*) is one of the key regulatory enzymes involved in the metabolic pathway of homocysteine. We investigated whether two polymorphisms (A66G and C524T) of the *MTRR* gene are associated with CHDs. A total of 599 children with CHDs and 672 healthy children were included; the polymorphisms were detected by PCR and RFLP analysis. Significant differences in the distributions of A66G and C524T alleles were observed between CHD cases and controls, and slightly increased risks of CHD were associated with 66GG and 524CT genotypes (odds ratios = 1.545 and 1.419, respectively). The genotype frequencies of 524CT

in the VSD subgroup, 66GG and 524CT in the PDA subgroup were significantly different from those of controls. In addition, the combined 66AA/524CT, 66AG/524CT and 66GG/524CT in CHDs had odds ratios = 1.589, 1.422 and 1.934, respectively. Increased risks were also observed in 66AA/524CT and 66GG/524CT for ASD, 66AG/524CT for VSD, as well as 66GG/524CT for PDA. In conclusion, *MTRR* A66G and C524T polymorphisms are associated with increased risk of CHDs.

Key words: Congenital heart defect; Polymorphisms; Homocysteine; Methionine synthase reductase; Folic acid

