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Genetic variants of the endothelial NO synthase gene (*eNOS*) may confer increased risk of sporadic congenital heart disease

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ABSTRACT. The endothelial NO synthase (eNOS) enzyme is expressed during the early stages of cardiogenesis and plays an important role in normal heart development. Genetic variations of *eNOS* G894T have been shown to influence individual susceptibility to some phenotypes of congenital heart disease (CHD) in different populations. We conducted a case-control study comprised of 945 CHD patients and 972 non-CHD individuals in a Chinese population. Two functional single nucleotide polymorphisms (SNPs) (T-786C: rs2070744 and G894T: rs1799983) and one tagging SNP (rs7830) were evaluated in our study, and we assessed their association with the risk of CHD. Compared with the rs7830 CC/AC genotypes, the *eNOS* rs7830 AA genotype showed a significantly increased risk of

CHD (adjusted odds radio (OR) = 1.45, 95% confidence interval (CI = 1.13-1.85). A stratified analysis was performed and showed that the association between the rs7830 AA genotype and CHD risk was stronger in patients with perimembranous ventricular septal defects (adjusted OR = 1.62, 95%CI = 1.20-2.20). Our results suggest that the *eNOS* rs7830 polymorphism may contribute to the susceptibility of sporadic CHD in a Chinese population.

Key words: *eNOS*; Congenital heart disease; Polymorphism

