Abstract
Background: Transforming growth factor- receptor II (TGFBR2) is a key component of TGF- signaling pathway. TGFBR2 can be detected in the generation of heart. The mouse embryos of TGFBR2 gene knockout exhibited congenital heart defects. Methods: We conducted a case-control study to investigate the association between TGFBR2 gene polymorphisms and congenital heart defects in Han Chinese population. 125 patients with congenital heart defects and 615 unrelated controls were recruited. Two tagging single nucleotide polymorphisms (tagSNPs) in 5’ upstream of TGFBR2 gene (rs6785358, -3779A/G; rs764522, -1444C/ G) were selected and genotyped by polymerase chain reaction (PCR)-restriction fragment length polymorphism (RFLP) assay. Results: A significant difference was seen in the distribution of genotypes between patients with congenital heart defects and controls for SNP rs6785358 (P=0.043). For SNP rs6785358 the carrier of the G allele (AG/GG genotype) showed a significantly higher risk of congenital heart defects compared with AA homozygotes (OR=1.545, 95% CI: 1.013–2.356). Further analysis by sex stratification indicated that individuals carrying G allele (AG/GG genotype) for SNP rs6785358 have a higher susceptibility to congenital heart defects (OR=2.088, 95%CI: 1.123-3.883, P=0.019) in males, but not females (OR=1.195, 95%CI: 0.666-2.146, P=0.55). No statistical significance was detected in the distribution of genotypes and allele frequencies for SNP rs764522 between patients and controls. Conclusion: Our result suggested that SNP rs6785358 of TGFBR2 gene was associated with increased risk of congenital heart defects in Han Chinese men and further research would be warranted.

Keywords
Congenital heart defects, TGFBR2, Gene polymorphisms, Case-control study.