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Relationship between TBX20 gene polymorphism and congenital heart disease

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ABSTRACT. Congenital heart disease in children is a type of birth defect. Previous studies have suggested that the transcription factor, TBX20, is involved in the occurrence and development of congenital heart disease in children; however, the specific regulatory mechanisms are yet to be evaluated. Hence, this study aimed to evaluate the relationship between the TBX20 polymorphism and the occurrence and development of congenital heart disease. The TBX20 gene sequence was obtained from the NCBI database and the polymorphic locus candidate was predicted. Thereafter, the specific gene primers were designed for the restriction fragment length polymorphism-polymerase chain reaction (RFLP-PCR) of DNA extracted from the blood of 80 patients with congenital heart disease and 80 controls. The results of the PCR were subjected to correlation analysis to identify the differences between the amplicons and to determine the relationship between the TBX20 gene polymorphism and congenital heart disease. One of

the single nucleotide polymorphic locus was found to be rs3999950: c.774T>C (Ala265Ala). The TC genotype frequency in the patients was higher than that in the controls, similar to that for the C locus. The odds ratio of the TC genotypes was above 1, indicating that the presence of the TC genotype increases the incidence of congenital heart diseases. Thus, rs3999950 may be associated with congenital heart disease, and TBX20 may predispose children to the defect.

Key words: TBX20 transcription factor; Gene polymorphism; Allele; Congenital heart disease; Single nucleotide polymorphism loci; Genotype frequency

