Are there any associations between the cardiac septal defects and ROCK2 gene polymorphisms at the childhood? : A case-control study

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Background: Rho/Rho-kinase pathway plays a critical role in the regulation of cellular functions such as proliferation and migration. One of possible theories of the development of ventricular septal defects is cell migration disorder. The aim of this study was to analyze the genotype distributions and allele frequencies for ROCK2 gene Thr431Asn, Asp601Val, and Lys1083Met polymorphisms among the cardiac septal defects patients in a Turkish population. Methods: In this case-control study, 300 patients with cardiac defects (150 patients with ventricular and 150 patients’ atrial septal defects) and control group (150 healthy control subjects) were investigated. A single-nucleotide polymorphism in ROCK2 gene Thr431Asn was analyzed by real-time polymerase chain reaction using a Light-Cycler, and Asp601Val, and Lys1083Met polymorphisms was detected by restriction fragment length polymorphism polymerase chain reaction. Results: Neither genotype distributions nor the allele frequencies for the Thr431Asn, Asp601Val, and Lys1083Met polymorphisms showed a significant difference between the groups. Conclusion: These results suggest that there were no evidence for an association of the ROCK2 gene Thr431Asn, Asp601Val, Lys1083Met polymorphisms with cardiac septal defects in pediatric patients.