

Combination of single nucleotide polymorphism of hemostatic system genes in patients with single ventricle

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Aim: To study the combination of single nucleotide polymorphism (SNP) of hemostatic system genes associated with thrombosis appearance in patients with single ventricle (SV).

Methods and materials: Molecular genetic study with diagnostics of SNP of hemostatic system genes was performed. The average age was 3,3 (0,6; 5,0) years. Out of 102 examined children thrombosis was recorded in 13 patients with SV during follow up. Signs of thrombosis were not discovered in 89 patients during defect surgical correction. Study material was whole blood. DNA samples were tested for single nucleotide polymorphism in hemostatic genes: F2:20210 G>A (Factor II), F5:1691 G>A (Factor V, Leiden Mutation), FGB: -455 G>A (Factor I), ITGA2:807 C>T (platelets collagen receptor GP Ia-IIa), ITGB3:1565T>C (platelets fibrinogen receptor GP IIb-IIIa), PAI-1:-675 5G>4G (plasminogen activator inhibitor I). Genotype was detected by polymerase chain reaction method using market reagent kit (DNA-Technology, Russia).

Results: Carriership of one polymorphism was discovered in 9 (10,1%) examined children without thrombosis, and children with thrombosis had more than one polymorphism. The combination of two polymorphisms was disclosed in 16 (18,0%) children with SV without thrombosis, and in 6 (46,2%) children with thrombosis. Three polymorphisms were discovered in 35 (39,3%) children without thrombosis and in 3 (23,1%) patients with thrombosis. Polymorphism of four genes was detected in 20 (22,5%) children with SV without thrombosis, and in 3 (23,1%) children with thrombosis. Five polymorphisms were discovered in 7 (7,9%) children without thrombosis, in 1 (7,7%) patient with thrombosis. Six polymorphisms were marked in 1 (1,0%) child without thrombosis, children with thrombosis had no that combination. Depending on a carriership of polymorphism combination statistically significant differences ($\chi^2=0,34$, $p=0,56$) were not disclosed in comparative analysis of children with and without thrombosis.

Conclusion: As a result of molecular genetic analysis risk of thrombosis appearance depending on combination of single nucleotide polymorphism of hemostatic system genes in patients with single ventricle was not detected.