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Distribution of allelic variants of hemostatic genes in patients with single ventricle

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Aim: To study the frequency of allelic variants of hemostatic genes associated with thrombophilia in patients with single ventricle.
Methods and materials: Molecular genetic testing with diagnostics of allelic variants of hemostatic genes was performed to 102 patients with single ventricle. 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 fibrinogenic 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: 97.1%) of examined children had wild type (GG) of factor II gene, 2.9% - heterozygous (GA), 0% – homozygous (AA). Carriers of wild type factor gene (GG) were 94.1% of children, heterozygous (GA) – 5.9%, homozygous for a mutant allele (AA) – 0%. Polymorphism of factor FGB gene was the following in children with single ventricle: 52.0% – wild type (GG), 39.2% – heterozygous (GA), 8.8% – homozygous genotype (AA). Study of allelic variants of PAI-1 gene disclosed wild type (5G5G) in 5 (14.7%) children with CHD, heterozygous (5G4G) – in 50 (49.0%), homozygous (4G4G) genotype – in 37 (36.3%). Study of thrombocytic glycoproteins GP Ia-IIa gene discovered wild type (CC) in 45 (44.1%) children with single ventricle, heterozygous (CT) – in 41 (40.2%), homozygous (TT) variants – in 16 (15.7%). Polymorphism of GP IIa-IIIb was the following in children with single ventricle: wild type (TT) – in 77 (75.5%) pts, heterozygous (TC) – in 22 (21.6%) pts, homozygous (CC) genotype – in 3 (2.9%) pts.
Conclusion: The results of molecular genetic testing of allelic variants of hemostatic genes together with current diagnostic procedures can be used for risk prediction and early therapy management of thrombophilic state in patients with single ventricle.