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Genetic effects of *PPARG1C* and *TNF1* gene variants related to cardiovascular disorders

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Introduction: Study was aimed to investigate the associations between the SNPs in *PPARG1C* (Gly482Ser), *TNF* (-308G-A) genes and coronary heart disease (CHD) and hypertension and their relations to leukocyte profile and plasma lipid composition in Russian population. **Materials and Methods:** 124 participants older than 55 years were divided into groups according to the anamnesis data. All participants were genotyped for SNPs *PPARG1C* (Gly482Ser) and *TNF* (-308G-A) by allele-specific real-time polymerase chain reaction method using commercial kits. Lipid profiling was measured by homogeneous enzymatic colorimetric test. **Results:** The results of this study suggested the presence of the mutation *PPARG1C* (Gly482Ser) increases the risk of coronary heart disease by 2.4 times (OR 1.17–4.97), but is not associated with arterial hypertension. *TNF* -308A t allele has demonstrated a protective effect, reducing the risk of developing hypertension (OR 0.01–0.84), but is not associated with CHD. The relationship between genotypes and the average values of leukocyte and lipid profiles remained non-significant in our population. However the heterozygous genotype of *TNF* was related to an increased level of drumstick neutrophil, compared to the control group. **Conclusion:** Thus, the investigated polymorphic variant (Gly482Ser) of *PPARG1C* gene is a reliable marker of CHD, whereas in the case of hypertension the association with (-308G-A) of *TNF* gene was identified. This research supported by the grant of the Russian Science Foundation №. 15-15-10022.

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Polymorphisms of TNFA in hypertensive patients

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Introduction: Hypertension is a complex, multifactorial and polygenic disease. This descriptive study analyzed whether there are differences in TNFA -238 G>A and TNFA -308 G>A polymorphisms that encode molecules involved in inflammation among hypertensive patients, refractory hypertensive patients and controls. **Materials and methods:** We performed a case-control study with 444 subjects: 234 hypertensive patients (HTA), 50 refractory hypertensive patients (HTA-R) and 160 controls. The DNA was amplified by PCR and TaqMan trials allowed allelic discrimination. There were no statistically significant differences in either age or sex between all 3 study groups. **Results:** In the analysis of the polymorphisms of TNFA -308 G>A, the distribution of the allele AA for HTA and HTA-R groups was 0% and 2.4%, respectively ($p = 0.02$). No other significant differences were found in the study groups or in the analysis of TNFA -238 G>A genotypes. **Conclusion:** Variations in the polymorphism TNFA -238 G>A do not affect the chances of having refractory hypertension. For the first time, we describe the association between the AA genotype of TNFA -308 G>A and refractory HTA. Individuals carrying the allele A will have an increase in the levels of TNF α which causes endothelial damage. This endothelial alteration could justify the severity of hypertension and the poor response to drugs. This could lead to future therapeutic opportunities in these patients.

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SERPINE1 gene polymorphic variant in the predisposition to cardiovascular diseases

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Genes of predisposition to cardiovascular diseases are actively studied worldwide, including the plasminogen activator inhibitor gene (*SERPINE1*). However, its role in the development of hypertension, atherosclerosis, coronary artery disease, myocardial infarction is poorly understood, and the results of the studies tend to be contradictory. In this regard, the aim of this work was to analyze the association of *SERPINE1* gene polymorphism (-675 5 G/4 G, rs587776796) with the risk of hypertension, atherosclerosis, coronary heart disease (CHD), myocardial infarction development in Russian population. The study involved 121