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Association between *FBN1* polymorphisms and TGF-β1 concentration within aneurysms and dissections of ascending thoracic aorta

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Transforming growth factor β1 (TGF-β1) is a cytokine that participates in a broad range of cellular regulatory processes and is associated with various diseases including aortic aneurysm. An increased TGF-β1 level is associated with Marfan syndrome (MFS) caused by fibrillin-1 (*FBN1*) mutations and subsequent defects in signaling system. *FBN1* single nucleotide polymorphisms (SNPs) rs2118181 and rs1059177 do not cause MFS but are associated with dilatative pathology of ascending aorta. A purpose of the investigation was to test hypothesis does an association between *FBN1* SNPs (rs2118181, rs1059177) and TGF-β1 level in human blood plasma exist among sporadic cases of dilatative pathology of ascending aorta. A study group was recruited from 312 patients who had undergone aortic reconstruction surgery due to dilatative pathology of ascending aorta and 741 healthy control subjects of Kaunas population (N=269) without cardiovascular disorders, except hypertension. Genomic DNA was isolated from potassium EDTA blood. Genotyping of *FBN1* SNPs was carried out by using ABI 7900HT Real-time PCR Thermocycler with commercially available kits from Applied Biosystems. TGF-β1 quantitated detection was tested with eBioscience Platinum human TGF- β1 ELISA commercially available kit based on standard sandwich enzyme-linked immune-sorbent assay technology according manufacturers' instructions. Non-parametric Kruskal-Wallis test was used for data analysis. The results showed a quantitative dependence of SNP genotype and TGF-β1 concentration. A presence of a single rs2118181 minor allele (G) increased the median amount of TGF-β1 level. Two copies of *FBN1* rs1059177 minor allele (G) were required to give a significant rise of TGF-β1 level in blood plasma. We also found higher TGF-β1 concentrations in men compared to women (p=0.001). The results are indicating that presence of minor allele of *FBN1* SNPs rs2118181 or presence of homozygous genotype of minor alleles of rs1059177 is associated with the significant increase in TGF-β1 blood plasma level but the mechanism of this association is still unknown.

Biography

Ramune Sepetiene is currently a PhD Student at Lithuanian University of Health Sciences. She has obtained her MD with medical laboratory specialization in 1999 from Lithuanian University of Health Sciences, Medicine Academy. She has more than 15 years of clinical work experience within immunology, hematology and genetics. She is a Junior Researcher in Laboratory of Molecular Cardiology, Institute of Cardiology, LUHS and part time laboratory MD position in patients' clinic. Recently she has published 4 papers within PhD dissertation subject in reputed journals of cardiac surgery and genetics.

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