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Title: Single nucleotide polymorphism of PSMA6 gene in Lithuanian patients with asthma

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Body: Introduction: Asthma is a chronic airway inflammatory disorder which morbidity is increasing worldwide, but the predisposing factors are not well known. Recently much attention has been given to the association between polymorphism of proteasome encoding genes and various diseases. Aim: The aim of this study was to evaluate single nucleotide polymorphism (SNP) of PSMA6 gene in Lithuanian subjects with asthma and healthy subjects. Methods: Patients with asthma (n=145; mean age 46.4±15.3yrs) and healthy subjects (n=150; mean age 41.0±14.4yrs) as a control group were involved in this study. Asthma was diagnosed according to GINA criteria. DNA from peripheral blood was extracted using Qiagen (Germany) DNA blood mini kit standardized protocol. SNPs were evaluated using allele specific amplification and cleaved amplified polymorphic sequence methods. These SNPs of PSMA6 gene were evaluated: rs2277460 in promoter region (-110 from the translation start, -110C>A) and rs1048990 in 5'UTR region (-8 from the translation start, -8C>G). Results: The frequency of A allele (-110C>A) polymorphism in patients with asthma was 12.8% and 9.3% in healthy subjects. CA genotype was also found more frequently in asthmatics (25.0% vs 18.7%), but there was no association between asthma and this genotype ($\chi^2=1.94$; p=0.16). The G allele (-8C>G) was found in 18.1% of asthmatics and in 15.3% of healthy subjects. The frequency of genotypes CG, GG did not significantly differ between studied groups (in patients with asthma: CG – 28.0%, GG – 4.0%; in healthy subjects – 28.0%, 1.3%, respectively). Conclusions: The study showed that some SNPs of PSMA6 gene are present more frequently in patients with asthma than in healthy subjects.