ANALYSIS OF TMCO1 GENE POLYMORPHISMS IN LATVIAN PATIENTS WITH PRIMARY OPEN ANGLE GLAUCOMA

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Introduction. Glaucoma is a complex, chronic neurodegenerative optic neuropathy and one of the leading causes of adult visual impairment and irreversible blindness, affecting over 60 million people worldwide. Primary open angle glaucoma (POAG) is the most common form of the disease. It is thought that the crucial risk factor for POAG pathogenesis is an elevated level of intraocular pressure (IOP) but other coexisting risk factors that plays an important role are age, race, sex, diabetes mellitus type 2, vascular alteration and oxidative DNA damage. At the present time, glaucoma is clinically defined, but the exact etiology is unknown. Genetic studies are one approach to identify the molecules and pathways involved in disease pathogenesis.

Aim. The purpose of this study was to determine the association of the single nucleotide polymorphism (SNP) rs 4656461 near the TMCO1 gene with POAG in the Latvian population.

Material and Methods. The study included 65 patients with POAG and 43 control persons (with cataract). Genomic DNA was isolated from peripheral blood leukocytes. A real-time PCR approach was used for the genotyping of the SNP rs4656461 (TaqMan® SNP Genotyping Assay, Life Technologies). The real time-PCR reactions were carried out in 10μl volume in accordance to manufacturer`s protocol.

Results. All samples were successfully genotyped. The observed results were in accordance to minor allele frequencies for this SNP. The genotype "A/A" was predominant among cases (78%) as well as among the controls (84%). The distribution of mutant allele was similar in both study groups.

Conclusions. There were no statistically significant differences between POAG and cataract patients in terms of the different genotype distribution. Additional studies in a larger cohort are required to investigate the associations of different genetic loci with the POAG in Latvian population.