

Abstract

Introduction: The epigenetic modifications can significantly affect and alter the gene activity by regulating their expression, having direct impact on various processes in human body. Epigenetic processes are involved in etiopathogenesis of many diseases. From this point of view, MHC genes are very important as they were linked to many autoimmune disorders, for example type 1 diabetes mellitus. In general autoimmune diseases appear to be connected to certain MHC class II genes.

Aims: The aim of this thesis is to determine the relationship between expression levels and histone modifications present in the promoter area of MHC class II gene, DQA1. Moreover, we also analyze and compare the DQA1 gene mRNA expression depending on the QAP promoter allele.

Methods: We isolated both nucleic acids (DNA and RNA) and leukocytes from peripheral blood samples collected from voluntary donors. DNA was utilized for genotypization of individuals. RNA was subjected to reverse transcription and the quantitative PCR was performed in order to determine the level of expression. Leukocytes were used for chromatin immunoprecipitation, which was evaluated using quantitative PCR.

Results: The expression level of QAP allele 3.1 was found to be higher than for the rest of the alleles Allele 4.1A showed, on the other hand, expression significantly lower. Histone modifications were measured for DQA1 alleles *01, *02, *03 and *05. No statistically significant relationship between allelic expression and histone modifications present was found.

Keywords: MHC class II, DQA1, epigenetics, histone modification, RNA expression, genotypization