Abstract (EN)

Type 1 diabetes (T1D) belongs among polygenic multifactorial autoimmune diseases. The highest risk is associated with HLA (human leukocyte antigen) class II genes, including *HLA-DQA1* gene. Our aim was to investigate DNA methylation of *HLA-DQA1* promoter alleles (QAP) and correlate methylation status with individual *HLA-DQA1* allele expression of T1D patients and healthy controls. DNA methylation is one of the epigenetic modifications, that regulate gene expression and is known to be shaped by the environment.

61 T1D patients and 39 healthy controls were involved in this study. Isolated DNA was treated with sodium bisulfite and *HLA-DQA1* promoter sequence was amplified using nested PCR. After sequencing, DNA methylation of *HLA-DQA1* promoter alleles was analyzed. Individual mRNA *HLA-DQA1* relative allele expression was assessed using two different endogenous controls (*PPIA, DRA*).

We have found statistically significant differences in *HLA-DQA1* allele 02:01 expression (PPIA normalization, P_{corr}=0.041; DRA normalization, P_{corr}=0.052) between healthy controls and T1D patients. The complete methylation profile of the *HLA-DQA1* promoter was gained with the most methylated allele *DQA1*02:01* and the least methylated *DQA1*05:01* in both studied groups. Methylation profile observed in T1D patients and healthy controls was similar, and no correlation between *HLA-DQA1* allele expression and DNA methylation was found. Although we have not proved significant methylation differences between the two groups, detailed DNA methylation status and its correlation with expression of each *HLA-DQA1* allele in T1D patients have been described for the first time.

Key words: autoimunity, HLA class II genes, *HLA-DQA1*, epigenetics, DNA methylation