HLA class II genes are known to be highly polymorphic, even in the regulatory non-coding gene regions. Polymorphism in the promoter region potentially forms a strong basis for an uneven allele-specific expression. Even though it is known that the amount of HLA class II molecules on the cell surface has a significant role in shaping immune response, HLA class II expression polymorphism has not yet been thoroughly measured.

The thesis aims to shed light into allele-specific mRNA expression and promoter DNA methylation of HLA class II genes. Two studies, each addressing different aspects of the HLA class II allele expression regulation, were conducted.

Study A examines the DNA methylation of 10 *DQA1* promoter and its effect on the *DQA1* mRNA expression. DNA methylation in whole blood cells was determined with bisulfite sequencing and mRNA expression was measured using RT-qPCR. Even though inter-allelic differences in overall methylation were observed (the most methylated alleles were *DQA1*02:01* and **04:01*), the expected negative correlation between the *DQA1* promoter DNA methylation density and the allele expression was not observed. We suggest that the genetic polymorphism in the region (especially region upstream of position -400, which is almost completely methylated in all alleles) may lead to different interpretation of the same 5meCpG mark in different allelic contexts.

Study B analyses *DQA1* and *DQB1* mRNA expression in whole blood cells, B lymphocytes and monocytes using RT-qPCR. Class II transcription level is higher (with the exception of *DQB1*06* alleles) and shows lower interallelic variation in B cells compared to monocytes. The *DQB1* expression hierarchy can be generalized into *DQB1*06 > *03,*05 > *02* pattern in monocytes and *DQB1*06 > *02, *03, *05* in B cells. The *DQA1* expression hierarchy is *DQA1*03 > *01 ≥ *02 > *05* in monocytes and *DQA1*03, *05 > *01, *02* in B cells. Because of the low number of samples for certain allele-cell type combinations, only some of these results were statistically significant. CIITA isoforms are discussed as the factors that could drive the cell-type specific expression of class II alleles. Finally, we tried to relate DQ dimer expression level to the risk it carries for the development of the autoimmune diabetes.

In conclusion, this thesis confirms and describes mRNA expression polymorphism of the *HLA-DQA1* and *-DQB1* genes, and provides the expression hierarchy of these alleles in the B cells, monocytes and whole blood cells. It presents a DNA methylation profile of the *DQA1* gene promoter and shows that DNA methylation by itself is not able to explain the inter-allelic expression differences. In our simplified model of DQ dimer expression, the expression level by itself is not able to explain dimer association with T1D.