3 Conclusions

The aims of PhD thesis were to contribute to knowledge of variability of human mitochondrial DNA in Czech population and to evaluate possible associations of variability with type 2 diabetes.

Sequence analysis of HVRII in D-loop shows high variability of this region, which is competently used for forensic or evolutionary studies. HVRII together with HVRI and polymorphic sites in encoding region of mtDNA form geographically characteristic haplotypes.

Prevalence of a whole range of disorders, including type 2 diabetes, varies in different populations. Hypothesis, whether genetic background of mitochondrial DNA could influence common type 2 diabetes, was tested in many studies. However, the results have been controversial.

Low incidence of A3243G tRNA\textsubscript{Lee(UUR)} mutation, which is the most common cause of mitochondrial diabetes, cannot entirely explain the fact that type 2 diabetes is more frequently inherited from a mother.

In this PhD thesis is shown that in Czech type 2 diabetic population the A3243G tRNA\textsubscript{Lee(UUR)} mutation have not key role to perform a common form of this disease. The patients screening for A3243G tRNA\textsubscript{Lee(UUR)} mutation is not well-founded.

Type 2 diabetes belong to complex diseases therefore study of polymorphism influence on develop and disease process. Candidate polymorphism for diabetes is the T16189C in the hypervariable region I of D-loop. In this PhD thesis was discovered influence of presence variant C to anthropometric and biochemical parameters.
In comparison to other studies which describe influence of variant C to parameters associated with diabetes or metabolic syndrome there is not any evidence of similar results. This part of thesis is pilote study for selection of variant C carriers which could be used for consequential study. Detection of possible length polymorphism which results due to variant C and association variant C with tested parameters are necessary for exact evaluation of variant C influence to diabetes.

Defined aims of the PhD thesis were carried out and as well as shown that this interesting questions of mitochondrial DNA variability is still actual for consequestion studies.