STRESZCZENIE PRACY W JĘZYKU ANGIELSKIM

High myopia (HM) is a disorder in which the light entering the eye is not focused correctly on a retina. It is characterized by a refractive error greater than -6.0 dioptres and axial length exceeding 26.0 mm. HM etiology is caused by environmental and genetic factors. Numerous loci, candidate genes and sequence variants related to HM have been identified in specific populations and families, however, little is known about genetic and epigenetic causes of HM in Polish population.

The aim of the PhD thesis was identification of novel genetic and epigenetic factors in HM etiology in Polish patients. Experimental procedures with the use of molecular biology techniques were conducted on DNA extracted from peripheral blood samples from members of Polish HM families, Polish children with HM, and children without HM as control group.

As a part of PhD thesis the statistical assessment of clinical data of Polish HM families' members was performed. Sequence variants in *ABCC6* and *FLRT3*, *SLC35E2B* genes segregating with HM phenotype in specific families were identified by Sanger sequencing and segregation analyses verification of chosen exome sequencing results conducted on DNA from HM families. The analyses of selected sequence variants did not reveal the segregation with axial length, intraocular pressure, corneal curvature, and HM in studied four families. Based on the genome-wide methylation results on DNA of Polish HM children and children without HM hyper- and hypomethylated CpG dinucleotides in promoter regions of *GSTM1*, *LCE3C*, *FARP2,* and *SORBS2*, *TANC1*, *ATXN1*, *ADAM20* genes and several enriched molecular pathways/processes were identified.

Statistical analyses of clinical data confirmed the correctness of dividing the families' members into subgroups according to refractive error and axial length criteria. Variants in *ABCC6*, *FLRT3*, and *SLC35E2B* genes could cause HM in the assessed families, but other than chosen for evaluation sequence variants could contribute to the ophthalmological parameters values. DNA hyper- and hypomethylation could contribute to gene silencing/overexpression and together with the enriched molecular pathways/processes impact the HM etiology in Polish children. Presented results confirm the complexity of genetic and epigenetic background of HM in families and children from Polish population.