Abstract

Circadian rhythm refers to wide scale of biological mechanisms which change periodically, for example changes in hormonal secretion, changes in body temperature and the sleep cycle. It is a self-sustaining system which repeats roughly every 24hours. Circadian rhythm is designed to response and synchronize with external environment to effectively react to external changes. Yet, it is able to maintain its rhythm despite the absence of any outer signals. This applies to humans as well, although the timing of „the inner clock" is not the same for every individual and can vary a lot. Chronotype describes an individual setting of physiological, biochemical and psychological variables, which closely depend on the sleep cycle. The timing of the sleep cycle along with the preferred time of activity reflects the chronotype of the individual. The genetic effect account for about one-half of the chronotype, yet to now days the mechanisms or precise locuses are still unclear. The aim of this diploma thesis is to select relevant polymorphisms of clock genes and other relevant genes and test their association with chronotype. Based on the results obtained from MEQ questionnaire, we received saliva samples from participants with extreme chronotypes. We isolated and sequenced this DNA samples using classical genetic methods (PCR, RFLP). The samples were tested for nine polymorphisms located in the following genes: *BMAL1, PER1, PER3, SIRT1* and *PGC-1α.* Afterwards, we statistically analysed each polymorphism individually and then we analysed the effect of polymorphisms pairing with one another. The results indicate that allele A in variant rs3736265 associates with evening chronotype. This polymorphism is located in *PGC-1α* gene which is mostly known for its crucial role in several metabolic pathways, but it is also involved in regulation of molecular clocks. In conclusion results of this diploma thesis points out the complexity of the circadian rhythm on molecular level and supports the approach which extends the focus on non-clock genes involved in molecular clocks.

Key words: BMAL1, circadian rhythm, CLOCK, chronotype, clock genes, metabolic genes, PER1, PER2, PER3, PGC-1α, SIRT1