Abstract:

The circadian system has evolved in organisms as an adaptation to periodic changes in the environment. Its task is to ensure regular entrainment between the solar cycle and the internal period of the organism, and to generate signals that synchronize behavioral and physiological processes in the body with the solar cycle. The whole mechanism takes place at the cell level, where there are regular oscillations of the transcriptional translation loops of the clock genes occur within 24 hours, thus ensuring a regular rhythm of the organism. However, the circadian system may not generate the same length of period in humans and may differ in the degree of entrainment with the external cycle. Base on that there are developed so-called individual time preferences. These different preferences are referred to as chronotypes, which fall into five categories: extremely evening, moderate evening, intermediate, moderate morning, and extremely morning type. Clock gene polymorphisms are considered to be one of the possible causes of these differences. The association of selected clock gene polymorphisms with extreme chronotypes is the subject of this diploma thesis. We obtained a saliva sample for DNA isolation from volunteers with extreme chronotypes. Using molecular methods of PCR, restriction digest and sequencing, we genotyped selected polymorphisms of the clock genes *Bmal1*, *Clock*, *Per2* and *Per3*. Statistical and correlation analyzes showed that in the *Clock* (T3111C) polymorphism, the C allele associates with the morning chronotype. Our results also suggest that the combination of haplotypes with the *Clock* gene polymorphism is affected by the observed association between the C allele and the morning chronotype. The combination "(AG + AA) / (55 + 45)" of the *Bmal1*/*Per3* haplotype also associates with the morning chronotype. Our work significantly expands the knowledge of other laboratories and confirms the assumption that the circadian phenotype of humans is not given by a simple change in protein structure given by a single polymorphism, but is most likely formed by a specific pattern of linked clock gene polymorphisms.

Keywords: circadian system, chronotypes, polymorphism, clock genes