Abstract

Retinitis pigmentosa is a genetic disorder affecting the retina. The progression of the disease leads to vision loss. This thesis concentrates on the causation of autosomal dominant retinitis pigmentosa. More specifically, the second biggest responsible mutation group is outlined. The above mentioned gene-mutations group is responsible for the formation of mutant variants of their corresponding splicing proteins. These proteins and consequences of their mutations are reviewed and presented in the thesis. The outline of mutation impact on the retina is presented for each mutated protein. The proteins in question are: PRPF8, PRPF31, PRPF3, PRPF4, PRPF6, SNRNP200, DHX38 (an exemption causing an autosomal recessive retinitis pigmentosa), PAP-1, CWC27 (an exemption causing an autosomal recessive retinitis pigmentosa). The literature review allowed the thesis to conclude that splicing proteins are highly likely to play a critical role in retina's health. In addition, some other noteworthy findings are briefly presented. For example, findings regarding lack of data about some of the mutations. Another example of such finding is that it still remains unknown why these mutations cause such a tissue-specific phenotype.

Key words: splicing, retinitis pigmentosa, snRNP, retina, autosomal dominant retinitis pigmentosa, splicing proteins