ABSTRACT

The anterior segment of the eye includes the eyelids, eyelashes, tear film, cornea, conjunctiva, iris, pupil, ciliary body, chamber angle, and lens. Cataracts and anterior segment dysgenesis are a highly heterogeneous group of diseases exhibiting all types of Mendelian inheritance.

The aim of this thesis was molecular genetic analysis of 51 patients from 23 families with congenital and early onset cataracts and/or anterior segment dysgenesis, using a modern as well as conventional methods, such as next-generation sequencing, direct sequencing, bioinformatic and functional analyses including the use of artificial splicing vectors.

We identified 24 causal variants in the coding, non-coding and regulatory regions of the *PAX6*, *FTL*, *FOXC1*, *PITX2*, *FOXE3,* and *CHRDL1* genes. We confirmed the role of the n.37C>T variant in *MIR204* in the development of chorioretinal dystrophy variably associated with iris coloboma, early-onset cataracts and congenital glaucoma. Splicing assay confirmed pathogenic effect of two mutations on pre-mRNA splicing of the *PAX6* gene.

Establishing molecular genetics diagnosis improves patient counselling and their relatives in terms of the prognosis and risk of developing secondary glaucoma

Key words: molecular-genetic diagnosis, anterior segment dysgenesis, next-generation sequencing, novel variants, cataract