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Opinion Article

Molecular Genetics of Primary Congenital Glaucoma

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DESCRIPTION

Recent molecular genetic studies have shed light on the molecular defects underlying Primary Congenital Glaucoma (PCG) and the clinical and genetic manifestations of this childhood eye disease. The existence of an inherited form of PCG segregation as a highly penetrant autosomal recessive trait has been identified. Mutations in the cytochrome *CYP1B1* gene have been discovered as the major molecular defect underlying most PCG cases. This gene is expressed in tissue of the anterior chamber angle of the eye. Molecular modeling experiments suggest that the mutations observed in a PCG patient affect the integrity of *CYP1B1* molecule, as well as its ability to adopt a normal conformation and bind heme. *CYP1B1* is involved in normal eye development and function by metabolizing essential molecules that may be used in signalling pathways. Glaucoma is a disorder of progressive death of the retinal optic nerve, resulting in the characteristic appearance of the optic nerve head and loss of vision. Glaucoma affects an estimated 66 million people worldwide, and glaucoma patient's account for nearly 50% of all cases in Asia. A total of 1.2% of children in the UK has glaucoma, and 3-7% of her children in India suffer from the disease. Based on the modern classification system, primary glaucoma can be divided into three different subtypes.

Primary congenital glaucoma is the leading cause of blindness in children and is characterized by congenital trabecular meshwork and anterior chamber angular dysplasia. Despite being a rare disease, PCG has a significant impact on patients' quality of life. However, the pathogenesis of PCG has not yet been fully elucidated. Although PCG patients display significant genetic heterogeneity, it was previously suggested that genetic factors play an important role in PCG pathogenesis. Mutations in the gene cytochrome P450 family 1 subfamily B member 1 are associated with PCG, and other factors reported to be involved in PCG are myocilin, type I collagen α1 chain and latent transforming. PCG is a rare form, accounting for 1-5% of all glaucoma cases. Additionally, PCG is highly prevalent, with a prevalence of over 32% in children with glaucoma. The overall incidence of PCG in Denmark is 4.8 per 100,000 live births.

Vision is one of the five senses that enable an autonomous and quality life; changes in ocular components are associated with multiple clinical phenotypes, from conjunctivitis to severe visual loss and irreversible blindness. Most of the clinical phenotypes are significantly associated with mutations in genes that regulate normal formation and maturation of the anterior segment of the eye. Among the diseases of the anterior segment, special attention is paid to glaucoma. Glaucoma is one of the leading causes of bilateral blindness worldwide and its development is due to Mendelian or multifactorial genetic features.

The clinical manifestations that characterize anterior segment disease reflect both phenotypic and genotypic heterogeneity and include multiple overlapping manifestations and associated genes. First, it is important to distinguish between anterior segment diseases characterized by Mendelian or simple genetics and diseases that develop as a result of complex or multifactorial traits. Mendelian diseases, in particular, can be traced to rare mutations responsible for specific genes (causative genes) and are inherited according to Mendelian laws, whereas multifactorial diseases can be traced to several genetic and environmental factors caused by the interaction of various factors. Early detection and treatment are essential to maximize visual potential. In the future, prenatal genetic screening may emerge as a preventative measure. It can be offered to parents in at-risk populations, such as those with family history or in consanguineous relationships in areas with higher PCG prevalence.

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