



## HEREDITY AND GLAUCOMA FORMATION

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**Abstract:** This scientific paper describes the role of heredity in the development of glaucoma, in a constant or periodic increase in intraocular pressure above the tolerance level of the optic nerve, in a decrease in visual functions, in disorders of the hydrodynamics and hydromechanics of the eye. The article summarizes the available data on the genetics of angle-closure glaucoma.

**Key words:** BYOBG, classification, genetics, polymorphism, acute attack of glaucoma.

### Relevance of the Topic

Glaucoma is considered a large group of diseases characterized by disruption of the eye's hydrodynamics and hydro-mechanics, persistent or periodic elevation of intraocular pressure above the tolerance level for the optic nerve, characteristic optic neuropathy, and a gradual decline in visual functions, primarily changes in the visual field. At present, glaucoma is one of the most challenging problems from both a medical and socio-economic perspective: today, 14% of all blind individuals have lost their vision due to this disease. Diagnoses such as ocular hypertension and suspected glaucoma cannot be overlooked. Genetic research plays a key role in understanding the mechanisms underlying glaucoma. Identifying genetic risk factors allows for the detection of individuals at high risk of the disease and the implementation of preventive measures. The MYOC and OPTN genes are among the most studied genes associated with glaucoma, with their mutations often linked to the development of primary open-angle glaucoma. This gene encodes myocilin, which plays a crucial role in maintaining the structure and function of the eye's trabecular meshwork. Abnormalities in this gene can lead to increased intraocular pressure, which in turn damages the optic nerve. The OPTN gene, associated with encoding optineurin protein, has also been identified as a genetic risk factor for glaucoma. Mutations in this gene can disrupt the activity of retinal ganglion cells and contribute to the development of glaucoma, especially in individuals with a family history of the disease.

Glaucoma can be suspected without the presence of ocular hypertension. The Shields Glaucoma Manual (2005) identifies a group for suspected glaucoma with a very high risk of disease development:

- IOP persistently greater than 21 mmHg;
- Central corneal thickness less than 555 microns;
- Advanced age;
- Glaucoma in blood relatives.

Let us examine family inheritance in more detail. In our country, the Russian Federation Ministry of Health adopted Order No. 869 dated October 26, 2017, "On Approval of the Procedure for Medical Examination of Certain Groups of the Adult Population," according to which intraocular pressure should be measured every 3 years (for citizens aged 60 and older), and for relatives of patients with glaucoma, the age limit is reduced without specification. All



educational activities with patients' relatives consist of teaching them the basics of assisting individuals with impaired vision. The goal is to prove the high likelihood of hereditary transmission of this disease and the necessity of taking measures in this regard.

### **Materials and Methods**

Retrospective analysis of patients' medical histories and clinical examination results at the Department of Eye Diseases Clinic of Andijan State Medical Institute. One clinical case was taken as an example.

### **Clinical Case No. 1**

On December 4, 2023, a 22-year-old young man complained of decreased vision in his right eye and periodic dizziness, noted approximately 3 years prior. He denies concomitant diseases. Objective: VIS = OD = 0.2 concav (-)1.5 D = 1.0; OS = 1.0. IOP = 26/21. Diagnosis = Mild degree myopia, ocular hypertension. Recommendations: Consultation with a neurologist, monitor IOP in 2-3 weeks. On December 14, 2023, IOP 22/22 under control. Monitoring without antihypertensive agents prescribed.

### **Conclusion**

I would like to draw the attention of the scientific community to the chronological order in which the clinical case is presented. The patient is mother and son, and later it became known that the mother and grandmother, i.e., in this family, there is direct inheritance of glaucoma. It should be remembered that the hereditary nature of this disease has been a confirmed scientific fact for a long time; however, despite the high frequency of family cases (often one or two generations after direct inheritance), there are no notes or additions for examining patients' blood relatives. I emphasize the need to lower the age for relatives of patients with glaucoma and increase the frequency of examinations to at least once a year until the age of 45. Additionally, educational activities with relatives of diagnosed patients should not only aim at improving the quality of life of victims and their visual adaptation, but also at independent and regular examinations, as the disease is often asymptomatic in many cases.

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