

**CLOSED ANGULAR WHAT IS GLAUCOMA ? IN PATHOGENESIS
MOLECULAR GENETICS FACTORS TO STUDY**

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Abstract. This scientific article discusses molecular genetic aspects in the pathogenesis of angle-closure glaucoma, as well as the etiology and pathogenesis of various types of this disease. The contribution of some genetic determinants to the development of the primary closed-angle form of glaucoma is analyzed. Pharmacogenomic studies have been shown to identify genetic factors that determine the side effects of pharmacotherapy.

Key words : BOBG, BYoBG, secondary glaucoma, genetics, trabeculae, neuropathy.

Primary angle-closure glaucoma (PACG) is one of the most unfavorable forms of glaucoma in terms of visual impairment. Tham YC et al. (2014) indicated that by 2040, the incidence of PAG will increase to 32.04 million people worldwide. The problems of primary angle-closure glaucoma have been highlighted in modern works by many authors [2-4]. However, the problems of PAG are no less significant, since the risk of blindness is at least three times higher than in open-angle glaucoma [5]. The relevance of the PAG problem is also confirmed by the fact that Varma D. K. et al. (2017) showed that 1 in 11 patients presenting to specialized glaucoma centers with a diagnosis of closed-angle glaucoma actually has a closed anterior chamber angle [6]. Epidemiology and risk factors Epidemiological studies have shown that the majority of cases of OCD occur in the Asian region due to the anatomical features of the structure of the eyeball. Representatives of the Mongoloid race have a short axial length of the eyeball and a smaller anterior chamber [7, 8]. The incidence of OCD is particularly high in China, with Chinese patients accounting for 47.5% of the total number of OCD cases worldwide [9]. The ratio of open-angle to closed-angle forms among people of Asian ethnicity and the Chinese population is 1:3 [11]. It is considered a deterministic disease resulting from the interaction of genetic and epigenetic factors. Consideration of genetic polymorphisms and other biomarkers significantly increases the effectiveness of diagnosis and treatment of the disease.

Glaucoma is a complex inherited disease, often associated with increased intraocular pressure (IOP), defects in the trabecular meshwork and anterior chamber, leading to impaired outflow of aqueous humor and progressive degeneration. However, increased IOP is neither necessary nor sufficient for the onset or progression of the disease. Mechanisms independent of the IOP are also involved in glaucomatous degeneration [25]. Glaucoma is classified according to etiology (primary and secondary), anterior chamber anatomy (open-angle and closed-angle), and time of onset (infantile-juvenile and adult). The different forms of primary glaucoma are divided into 3 main groups: open-angle glaucoma, the most common, angle-closure glaucoma, and congenital (hereditary) glaucoma [17]. According to large epidemiological studies, the prevalence of blindness due to angle-closure glaucoma is approximately 2 million people worldwide [30]. Primary angle-closure glaucoma (PACG) is one of the most unfavorable prognostic forms of glaucoma, accounting for 20-30% of primary glaucoma cases. There is no literature available. Studies have shown that ethnic origin also affects the risk of glaucoma, with Asian people being most susceptible to this

type of glaucoma. Some studies suggest that PACG is associated with mutations in 7 genes and other genetic markers. Disease-modifying genes and pharmacogenetic factors - the main method used in pharmacogenetic studies is association, which consists in comparing the allele frequencies of a candidate gene in patients and healthy people and determining the association of alleles with a particular phenotype. Association studies are based on a working hypothesis, which in turn is based on the known or suspected effect of a polymorphism of a candidate gene on the pathogenetic mechanism. Pharmacogenomic studies identify genetic factors that determine the side effects of pharmacotherapy. Singaporean researchers have identified three new genetic variants associated with primary angle-closure glaucoma, a disease that affects 15 million people worldwide (80% of whom live in Asia) and is a leading cause of blindness among Chinese people. Research institutions led by Singaporean Professor Tin Aung conducted a genome-wide association DNA analysis of 1,854 patients with primary angle-closure glaucoma and 9,608 controls. To do this, samples were taken from 5 libraries collected in Asia. They then conducted validation experiments using samples from another 1917 patients with primary angle-closure glaucoma and 8943 healthy individuals from six other sample collections collected around the world. The analysis identified three previously unknown loci associated with an increased risk of developing primary angle-closure glaucoma: the PC7 locus in the PLEKA11024102 gene, and the PC8 chromosome located between the PC1015213 and ST18 genes in the COL3753841A11 gene. The data obtained confirm the assumption made many years ago based on clinical observations about the hereditary nature of the disease. The researchers believe that their discovery will expand the possibilities of early diagnosis and prevention of primary angle-closure glaucoma, as well as help to understand the mechanisms of pathogenesis of this disease.

LITERATURE / REFERENCES

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