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MODERN IDEAS OF THE BASIS OF GENETIC PREDISPOSITION IN THE DEVELOPMENT AND COURSE OF MYOPIA

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Relevance of the problem. Over the past three decades, there has been an increase in axial farsightedness among young populations around the world, mainly in West and East-West Asia. This is caused primarily by hereditary factors, pregnancy defects, and with them the level of illumination and the color of various colors, the quality of daily diet, outdoor activities, environmental conditions and a number of other factors. which is explained by stress factors in preschool organizations and secondary schools. The state of health of students studying in schools and the degree of their mastery of subjects, resistance to environmental factors, the functional state of organs and systems, associated mainly with visual activity and its acuity, are presented in a number of scientific publications.

The genetic determinism of myopia was noted in the three-factor theory of Avetisov E.S. (1995), according to which refractive errors are formed with the participation of environmental factors, conditions of visual activity and hereditary factors.

Various studies have shown that myopia can be inherited in both autosomal dominant and autosomal recessive types. In the first case, the manifestation of the disease occurs mainly in adolescence with a milder clinical course. In the autosomal recessive inheritance type, myopia often develops at an early age and tends to progress with the development of complications.

According to existing ideas, the development of myopia involves increased extensibility of the sclera, resulting from mutations in one or more genes of the extracellular matrix of the sclera [Sun Y, Sha Y, 2024]. Also, the development of myopia is associated with a change in the elastic strength characteristics of the sclera and an increase in the anterior-posterior axis of the eyeball, a decrease in the content of glycosaminoglycans in the scleral membrane, a change in the structure of collagen, fibrillin, weakening of cross-links in collagen fibers, etc. [Iomdina E.N., Tarutta E.P., 2014; Troilo D, Smith EL, 2019; Sun Y, Sha Y, 2024].

Numerous scientific works of ophthalmologists have proven that myopia develops in the case of a number of genetic diseases accompanied by connective tissue dysplasia, such as Marfan syndrome, Ehlers-Danlos syndrome, Cohen syndrome, Knobloch syndrome, etc. For example,changes in the *ACTC1* gene involved in the formation of contractile cells – scleral myofibroblasts, the *GJD2* gene associated with the regulation of eyeball growth,

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the *GRIA4* gene associated with the metabolism of retinoic acid, the decrease in *MMP2* expression observed in myopia significantly suppresses the decrease in the accumulation of the Iα1 collagen chain in the sclera [Markosyan G.A., Tarutta E.P., 2016; Zhao F, Zhou Q, 2018; At , present, the search for the decisive genetic factors that determine the hereditary predisposition to myopia continues to be an urgent scientific task.

It is known that the formation of biologically active molecules is controlled by genetic mechanisms that underlie the regulation of the expression of the genes that code for them. Gene expression is carried out in the process of implementing the information encoded in the DNA structure with the formation of an mRNA molecule, then an amino acid sequence of a protein molecule that performs its intended function.

The level of expression of various genes has a high individual variability. A pathological or conditionally pathological process, for example, damage to tissue integrity, can lead to modulation of gene activity. But the level of expression of certain genes is also determined by the presence of genetic variants – single nucleotide polymorphisms (SNPs), when an individual has structurally and functionally alternative alleles of a certain gene.

A single nucleotide polymorphism occurs as a result of a point mutation, in which the DNA sequence of one of two identical regions of homologous chromosomes changes by one nucleotide. The leading approach to identifying genetic variants associated with polymorphisms is the study of candidate gene associations in case-control studies.

To assess the contribution of genetic predisposition to the development and course of myopia, it is necessary to study polymorphic variants of the transforming growth factor beta 1 (*TGFB1*) gene: *TGFB1* (C509T) – rs1800469, *TGFB1* (T869C) – rs1800470, *TGFB1* (G915C) – rs1800471, *TGFB1* (G800A) – rs1800468, as well as a polymorphism of the angiotensin-converting enzyme *ACE* – rs4646994 (I/D).

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