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ANALYSIS OF POLYMORPHISM OF GENES ASSOCIATED WITH ENDURANCE IN YOUNG ATHLETES-FOOTBALL PLAYERS


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ЁШ СПОРТЧИЛАР -ФУТБОЛЧИЛАР ЧИДАМЛИЛИГИ БИЛАН БОҒЛИҚ ГЕНЛАРНИНГ ПОЛИМОРФИЗМИНИ ТАҲЛИЛ ҚИЛИШ

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АНАЛИЗ ПОЛИМОРФИЗМА ГЕНОВ АССОЦИИРОВАННЫХ С ВЫНОСЛИВОСТЬЮ У ЮНЫХ СПОРТСМЕНОВ-ФУТБОЛИСТОВ

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Резюме. *ДНК полиморфизмлари ДНК кетма-кетлигидаги ўзгарувчан минтақалар бўлиб, улар камида 1% частотали популяцияда учрайди ва аксарият ҳолларда нейтрал таъсир кўрсатади. Шунингдек, ген экспрессиони даражасига, функционал маҳсулотларнинг (оқсиллар, РНК) фаоллигига ва оқсилларнинг тузилишига таъсир қилиши мумкин бўлган полиморфизмлар мавжуд. Ушбу полиморфизмларнинг функционал аҳамияти шундаки, улар ДНКнинг кодлаш (эксонлар, микроРНК генлари ва баъзи микроРНК интронлар генларини ўз ичига олган) ва тартибга солувчи (тарғиботчилар, кучайтирувчилар, изоляторлар) минтақаларида жойлашган. Айнан шу полиморфизмларнинг энг кам вакили бўлган турлари спорт генетиклари томонидан ассоциатив тадқиқотлар мавзуси деб топилган. Битта нуклеотидли полиморфизмлар битта геннинг бир нечта вариантлари (аллеллар) мавжудлигининг энг кенг тарқалган сабаби бўлиб, улар инсон геномидаги ўзгаришларнинг катта қисмини ташкил қилади. Полиморфизмларга, шунингдек, бир неча жуфт нуклеотидларнинг қўшилиши/ўчирилиши (қўшимчалари/томчилари), сегментар такрорланиши ва такрорланиши киради.*

Калит сўзлар: *генлар полиморфизми, чидамлилик билан боғлиқ генлар, ёш спортчилар-футболчилар.*

Abstract. *DNA polymorphisms are variable regions in the DNA sequence that occur in a population with a frequency of at least 1%, and in the vast majority of cases have a neutral effect. There are also polymorphisms that can affect the degree of gene expression, the activity of functional products (proteins, RNA) and the structure of proteins. The functional significance of these polymorphisms is due to the fact that they are located in coding (exons, microRNA genes and some introns containing microRNA genes) and regulatory (promoters, enhancers, insulators) regions of DNA. It is these, the least represented types of polymorphisms, that are the subject of associative research by sports geneticists. Single–nucleotide polymorphisms are the most common reason for the existence of several variants of one gene (alleles), they account for the vast majority of variations in the human genome. Polymorphisms also include insertions/deletions (inserts/drops) of several pairs of nucleotides, segmental duplications and repeats.*

Keywords: *polymorphism of genes, genes associated with endurance, young athletes-football players.*

According to modern concepts of molecular genetics of sports, it is believed that individual differences in the degree of development of certain physical and mental qualities of a person are largely due to DNA polymorphisms, of which there are at least 12 million. DNA polymorphisms are variable regions in

the DNA sequence that occur in a population with a frequency of at least 1%, and in the vast majority of cases have a neutral effect. There are also polymorphisms that can affect the degree of gene expression, the activity of functional products (proteins, RNA) and the structure of proteins. The functional significance of these polymorphisms is due to the fact that they are located in coding (exons, microRNA genes and some introns containing microRNA genes) and regulatory (promoters, enhancers, insulators) regions of DNA. It is these, the least represented types of polymorphisms, that are the subject of associative research by sports geneticists. Single–nucleotide polymorphisms are the most common reason for the existence of several variants of one gene (alleles), they account for the vast majority of variations in the human genome. Polymorphisms also include insertions/deletions (inserts/drops) of several pairs of nucleotides, segmental duplications and repeats [1].

To date, about 40 genes are known, the polymorphisms of which are associated with the development and manifestation of such a physical quality of a person as endurance, as well as functional signs and biochemical parameters that change under the influence of physical exertion of various directions. In addition to "sports" genetic markers of endurance, there are also genetic markers of "trainable endurance" identified as a result of dynamic (longitudinal) studies, when the effect of training and its relationship with genotypes are analyzed.

Among other genes involved in the implementation of genetic determination of physical endurance, genes of primary (ACE, PPARGC1A) and secondary importance (AT2R1) can be distinguished)[[2, 4, 6]. ACE is a gene of angiotensin converting enzyme (ACE), which is an important physiological regulator of blood pressure and water-salt metabolism. ACE converts inactive angiotensin I, which is circulating in the blood, into angiotensin II, which has a powerful hypertensive effect. This peptide not only regulates the state of human hemodynamics, but also as a growth factor enhances the synthesis of structural proteins in myocardial cells, which leads to hypertrophy of the heart muscle. PPARGC1A is a 1-alpha coactivator gene of the gamma receptor activated by peroxisome proliferators. It makes a significant contribution to the intensity of metabolic processes in skeletal muscles and myocardium. Through appropriate transcription factors, it affects the activity of the processes of adoptive thermogenesis, the formation of mitochondria and the intensification of oxidative processes, the relative content of "slow" muscle fibers, insulin secretion, gluconeogenesis, lipogenesis and chondrogenesis.

AT2R1 is a type 1 angiotensin II receptor gene. Mediates one of the main cardiovascular effects of angiotensin II, whose role is to regulate blood pressure. Through it, not only the constrictor effect of angiotensin II is realized, but also the expression of growth factors and the proliferation of smooth muscles.

I allele of the ACE gene, Gly/Gly allele of the PPARGC1A gene and A/A allele of the gene AT2R1 are markers of endurance [5].

The aim of the study was to identify and analyze the polymorphism of three genes: angiotensin converting enzyme, 1-alpha coactivator of the gamma receptor activated by peroxisome proliferators and angiotensin II type 1 receptor in young athletes engaged in a game sport (football).

Materials and methods of research. Samples of buccal epithelium of 91 youth football team of Tashkent were used as the material for the study. The analysis of gene polymorphism was carried out by polymerase chain reaction (PCR). DNA was isolated by the perchlorate method, which is based on cell lysis with sodium dodecyl sulfate and protein degradation by proteinase K, treatment with a mixture of sodium perchlorate, chloroform, isoamyl alcohol, DNA precipitation with ethanol. The DNA isolated by this method is suitable for long-term storage and makes it possible to use samples containing degraded DNA. The resulting DNA was used as a matrix in a polymerase chain reaction in the presence of primers of the author's design synthesized using the MerMade4 oligonucleotide synthesizer (Bioautomation, USA). Amplification of DNA fragments was carried out on programmable thermal cyclers (Biometra, Germany) using thermophilic DNA polymerase (ODO "Primtech", Belarus). Optimization of PCR conditions was carried out by varying the time and temperature parameters of the reaction, as well as using different pH buffer solutions, concentrations of magnesium chloride to ensure the specificity of the reaction. Amplified DNA fragments containing single nucleotide polymorphisms were treated with suitable restriction endonucleases (NEB, USA) in accordance with the methods recommended by the manufacturer. Restriction products, as well as PCR polymorphism products containing insertions/deletions, were separated using 2-3% agarose gel electrophoresis followed by visualization in the gel documentation system (Vilber Lourmat, France) [5]. In their works, Williams A.G. & Folland J.P. (2008) for the first time applied the approach of determining the optimal polygenic profile for the physical quality "endurance". This method seems to us simple, accessible and therefore probably found its development in subsequent publications [3].

To assess the genetic prospects of the tested football players on the basis of the polygenic profiles obtained by us, individual profiles of the studied polymorphisms were used in the method of calculating the "total genetic score" (OGB) with the assignment of points to their variants (0, 1, 2):

1. ACE I/D polymorphism: I/I = 2, I/D = 1, D/D = 0.

2. PPARGC1A polymorphism Gly482Ser: Gly/Gly = 2, Gly/Ser = 1, Ser/Ser = 0.

3. AT2R1 polymorphism A1166C: A/A = 2, A/C = 1, C/C = 0.

$$\text{OGB endurance} = \frac{100}{6} \cdot (\text{OGB } ACE + \text{OGB } PPARGC1A + \text{OGB } AT2R1)$$

Results. Polygenic profiles of the BGB range associated with the "endurance" quality in 91 tested young football players ranged from 16.7 to 100 with an average value of 58.3.

Thus, the analysis of the polygenic profiles of the examined young football players allowed us to identify genetically predisposed individuals to display the quality of "endurance" in them. Information about the genotype can be used by coaches to select promising athletes, choose an individual approach to training, correctly build the process of wellness classes, prevent the negative effect of excessive training effects that can lead to hypertrophy of the heart muscle.

Conclusion. It should be noted that all the students examined by us are genetically predisposed to engage in various sports, including football. A smaller number of individuals are likely to achieve a certain success and sportsmanship, since, in addition to genetic predisposition, favorable environmental factors are still necessary for this.

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АНАЛИЗ ПОЛИМОРФИЗМА ГЕНОВ АССОЦИИРОВАННЫХ С ВЫНОСЛИВОСТЬЮ У ЮНЫХ СПОРТСМЕНОВ-ФУТБОЛИСТОВ

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Резюме. *ДНК- полиморфизмы – это вариабельные участки в последовательности ДНК, которые встречаются в популяции с частотой не менее 1%, и в подавляющем большинстве случаев обладают нейтральным эффектом. Существуют также полиморфизмы, способные повлиять на степень экспрессии генов, активность функциональных продуктов (белков, РНК) и структуру белков. Функциональная значимость данных полиморфизмов связана с тем, что они расположены в кодирующих (экзоны, гены микроРНК и некоторые интроны, содержащие в себе гены микроРНК) и регуляторных (промоторы, энхансеры, инсуляторы) регионах ДНК. Именно эти, наименее представленные типы полиморфизмов, являются предметом ассоциативных исследований спортивных генетиков. Однонуклеотидные полиморфизмы – наиболее частая причина существования нескольких вариантов одного гена (аллелей), на их долю приходится подавляющее большинство вариаций в геноме человека. К полиморфизмам также относятся инсерции/делеции (вставки/выпадения) нескольких пар нуклеотидов, сегментальные дупликации и повторы.*

Ключевые слова: *полиморфизм генов, гены ассоциированные с выносливостью, юные спортсмены-футболисты.*