STUDY OF THE ACTN3 GENE POLYMORPHISM TO ASSESS THE PHYSICAL PERFORMANCE OF ATHLETES

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ABSTRACT

In the study of the distribution of genotypes of the ACTN3 gene associated with both speed-strength capabilities and endurance, 397 people took part, of which 296 were athletes of various specializations and 101 control groups to determine the frequency of occurrence of genotypes of the R557X polymorphism of the ACTN3 gene in Uzbek athletes. People involved in cycling, boxing, rugby, football, rowing and control groups - people who are not involved in sports. Thus, the results of the distribution of genotypes with R577X polymorphism of the ACTN3 gene in rowing athletes indicate a favorable effect of the presence of the R allele (genotypes RR and RX) on cycling and rowing

Key words: ACTN3, gene polymorphism, physical performance of athletes

All over the world, in connection with the decoding of the structure of the human genome, much attention is paid to the determination of genetic markers associated with the development and manifestation of physical qualities, as well as with biochemical, anthropometric and physiological indicators that are significant in the conditions of sports activity.

The main advantage of the molecular genetic method for identifying a hereditary predisposition of a person to motor activity is high information content in assessing the potential for the development of physical qualities and the possibility of early diagnosis.

In Uzbekistan, from the first years of Independence, the upbringing of a harmoniously developed younger generation has been identified as one of the priority directions of state policy.

The first gene studied in athletes was the a-actinin-3 gene (ACTN3, actinin alpha 3), which is located in the long arm of chromosome 11 (11q13-q14). In skeletal muscle, there are two isoforms of the α-actinin protein: α-actinin-2 isoform (ACTN2) and α-actinin-3 isoform (ACTN3), which differ in their localization in muscle fibers. All muscle fibers contain α-actinin-2, and the α-actinin-3 protein is expressed only in fast twitch fibers of skeletal muscle. The deficiency of α-actinin-3 in them reduces the speed-strength indicators of a person's physical performance. The reason for this deficiency in the ACTN3 protein is the single nucleotide substitution of cytosine for thymine at nucleotide 577.

The ACTN3 gene encodes the alpha-actinin-3 protein, which stabilizes the skeletal muscle contractile apparatus. Literature data indicate that the absence of alpha-actinin-3 (in the presence of two mutant (XX) copies of the ACTN3 gene) in fast muscle fibers, caused by a nonsense mutation in the coding sequence of the ACTN3 gene, may be a limiting factor in the development of speed and strength [2, 5, 6]. The presence of the R-allele in the genotype contributes to the more rapid development of speed-strength qualities, since the presence of the contractile protein alpha-actinin-3 promotes the synthesis of white, rapidly twitching fibers. The presence of the R allele is associated with an increased degree of muscle fiber hypertrophy [1, 3]. Genotypes containing two X alleles will build muscle more slowly.

In this regard, the purpose of our study was to study the molecular genetic foundations of predisposition to various types of sports activity, as well as to develop and test a method for diagnosing molecular genetic status, which determines the development of human physical qualities, and the following tasks were set:

developing the principles of molecular genetic diagnosis of hereditary a person's predisposition to sports activity, which allows assessing the genetic potential in the development and manifestation of physical qualities and optimization of the training process of athletes.

In the study studying the distribution of the genotypes of the ACTN3 gene associated with both speed-strength capabilities and endurance, 397 people took part, of which 296 were athletes of various specializations and 101 were the control groups.

DNA isolation was carried out according to the method of R. Boom et al. [4]. DNA genotyping for the ACTN3 gene was performed by real-time PCR using oligonucleotide primers and allele-specific fluorescent probes.

PCR amplification was carried out in a volume of 15 µl. The reaction mixture consisted of: 6.8 µl ddH2O, 1.5 µl 10xPCR buffer, 1.5 µl 25 mM MgCl2, 1.5 µl 10 mM dNTP mixture, 0.6 (10 pmol / µl) each oligonucleotide primer, 0.6 (10 pmol / µl) of each probe and 0.1 µl (0.5 units) “hot-star” Taq-polymerase and 1.2 µl of genomic DNA of athletes. Real-time PCR (“real-time PCR”) was performed on a Real Time PCR 7500 device (Appilied Biosystems, USA). The temperature-time regime of real-time PCR was as follows: 96 ° C - 5 min, then 44 cycles: 92 ° C - 15 sec, 60 ° C - 1 min.

Consequently, the carriage of the R allele of the ACTN3 gene, and hence the presence of the alpha-actinin-3 protein in skeletal muscles, gives an advantage when performing speed-power loads, the energy supply of which is carried out due to the anaerobic mechanisms of ATP resynthesis. The presence of the R allele (genotypes RR and RX) is beneficial for rowing athletes where explosive strength and speed are required.

Analysis of the frequency distribution of alleles and genotypes with R577X polymorphism of the ACTN3 gene in Uzbek athletes revealed statistically significant increase the frequency of occurrence of the R allele, as well as the R / R + R / X genotypes at rowing cyclists compared to the control group... It should also be noted that none of the athletes was a carrier of the XX mutant genotype for the ACTN3 gene.

It should also be noted a tendency towards a decrease in the frequency of occurrence of the XX genotype among rugby players (21.4%), among football players (18.2%), among athletes involved in cycling (12.5%) when compared with the control group.

Table 1

Results of the frequency distribution of the genotypes of the ACTN3 gene in the group of athletes and the control group

	RR		RX		XX		
	qty	%	qty	%	qty	%	Total
Cycling	28	43.8	28	43.8	eight	12.5	64
Boxing	twenty	33.3	25	41,7	15	25.0	60
Rugby	eight	14.3	36	64.3	12	21.4	56
Football	22	33.3	32	48.5	12	18.2	66
Rowing	24	42.9	32	57.1	0	0	56
Control	28	28	46	45.5	27	27	101

In rowers, a significant increase in the frequency of the R allele, as well as the RR + RX genotypes of the ACTN3 gene, was found. It should also be noted that none of the rowers were carriers of the mutant XX genotype.

It should be noted that during rowing, almost all the muscles of the body are involved in the advancement of the paddle blade in the water, which imposes great functional requirements on the muscular system.

Thus, the results of the distribution of genotypes with R577X polymorphism of the ACTN3 gene in rowing athletes indicate a favorable effect of the presence of the R allele (genotypes RR and RX) on cycling and rowing and are consistent with the above literature data.

When analyzing the distribution of variant genotypes of the R577X polymorphism of the ACTN3 gene, it was found that the control and cases corresponded to the theoretically expected equilibrium distribution of Hardy-Weinberg, except for the group of athletes engaged in rowing ($p = 0.003$). The frequency distribution of alleles and genotypes for ACTN3 among athletes involved in rowing deviated from the Hardy-Weinberg equilibrium, which indicates that sports selection took place based on a person's genetic predisposition.

Carriage of the R allele of the ACTN3 gene, and hence the presence of the alpha-actinin-3 protein in skeletal muscles, gives an advantage when performing speed-power loads, the energy supply of which is carried out due to the anaerobic mechanisms of ATP resynthesis. The presence of the R allele (genotypes RR and RX) is beneficial for athletes involved in cycling and rowing, where explosive strength and speed are required.

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