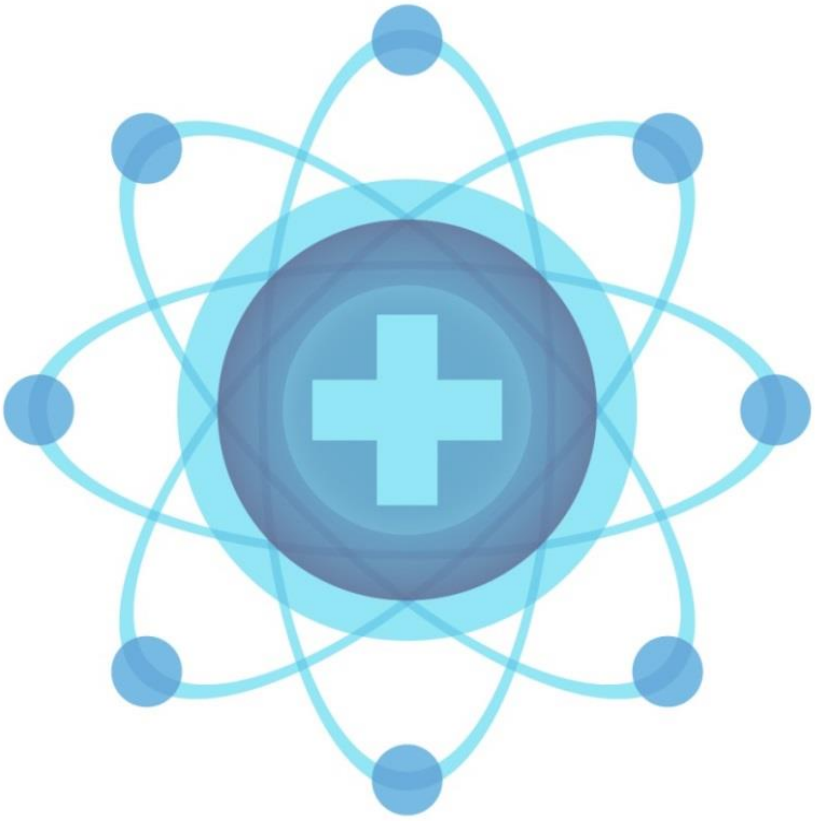

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INFLUENCE OF VARIOUS ALLELIC AND GENOTYPE VARIATIONS OF ACE, ACTN3 AND PPARGC1A GENES ON LABORATORY-BIOCHEMICAL AND MORPHOFUNCTIONAL INDICATORS IN ATHLETES (LITERATURE REVIEW)

[1]Mominov Aziz Alisherovich , [2]Sadikov Abdushukur Abduzhamilevich

[1] Republican Scientific and Practical Center of Sports Medicine, Tashkent.

[2]National Anti-Doping Agency of Uzbekistan, Tashkent.

Abstract. The article examines the influence of allelic and genotypic variations in the ACE, ACTN3 and PPARGC1A genes on laboratory-biochemical, anthropometric and morphofunctional parameters in athletes. The analysis of data on the relationship of polymorphisms of these genes with key parameters such as muscle mass, body composition, aerobic and anaerobic endurance, strength indicators and biochemical markers is carried out. Particular attention is paid to the differentiation of sports results depending on the genetic profile of athletes, which allows us to study the relationship between genetic characteristics and the functional capabilities of the body. Research Results This review may be useful for researchers working on the creation of genetically oriented programs for training athletes in order to improve the effectiveness of the training process.

Key words: Sports genetics, athletes, alleles and genotypes, endurance, strength, ACE, ACTN3, PPARGC1A, athletic performance, polymorphism, training adaptation, lactate, maximal oxygen consumption

Introduction Allelic variations in the ACE, ACTN3, and PPARGC1A genes are key targets of research in sports genetics. These genes have significant effects on physical fitness, including strength, endurance, recovery rate, and the ability to adapt to physical activity.

The ACE gene regulates the functions of the renin -angiotensin system, affecting blood pressure, vascular tone and cardiorespiratory endurance. The I/D (insertion /deletion) polymorphism in this gene is associated with various athletic qualities: the

I allele is associated with improved aerobic performance and more efficient oxygen transport, as confirmed by studies conducted on elite marathon runners (Puthucheary et al ., 2011). The D allele, on the contrary, is more common in athletes of speed-strength disciplines, such as sprinting or powerlifting (Eynon et al ., 2013).

The ACTN3 gene encodes a protein expressed in fast muscle fibers, which are involved in performing intense, short-term loads [1,4]. The R577X polymorphism (replacement of arginine with a stop codon) leads to differences in muscle function. Carriers of the R allele have an advantage in speed-strength sports, such as sprinting (Yang et al ., 2003). At the same time, carriers of the X allele demonstrate better resistance to fatigue, which may be an advantage in aerobic sports (MacArthur et al ., 2007).

The PPARGC1A gene plays an important role in energy metabolism, regulation of mitochondrial biogenesis, and adaptation to exercise. The Gly482Ser polymorphism affects the efficiency of oxygen use and the rate of recovery from exercise. For example, the Gly allele has been identified in athletes involved in endurance sports such as triathlon (Lucia et al ., 2006), whereas the Ser allele is associated with better adaptive capacity under high-intensity training conditions (Eynon et al ., 2011).

These findings highlight the importance of genetic profile in athletic performance. This article will examine the impact of allelic variations in these genes on athletic performance based on different athlete phenotypes and their ability to perform specific physical activities. Genetic variations such as ACE, ACTN3, and PPARGC1A polymorphisms may determine how efficiently the body utilizes oxygen, recovers from exercise, and is able to sustain prolonged effort, which is critical for success in endurance and strength sports (Eynon et al ., 2011; Puthucheary et al ., 2011).

Main part. The ACE gene encodes an enzyme involved in the regulation of blood pressure and electrolyte balance. Polymorphism of the ACE gene (insertion /deletion - I/D) divides people into three genotypes: II, I/D and D/D (Danser et al ., 2007). The II genotype is associated with increased aerobic endurance, which is

useful in cyclic sports such as long-distance running and swimming [8]. People with the II genotype often show better adaptation to endurance loads and use oxygen more efficiently. The D/D genotype is associated with strength indicators and a tendency to rapid muscle mass development, which is especially useful in sprint disciplines, weightlifting and other sports requiring explosive power [11, 25,36]. The ID genotype is considered intermediate and may provide advantages in mixed disciplines.

\Analyses of samples of athletes from different sports confirmed the associations between the II genotype and endurance, and the D/D genotype and strength. For example, a high frequency of the II genotype was found in marathon runners, while the D/D genotype predominated in sprinters (Ma et al ., 2013).

The study of the relationships between the ACE gene and its allelic variations (insertion /deletion - I/D) with laboratory parameters of athletes is an important topic in sports medicine and genetics. ACE polymorphism (I/D) affects the activity of angiotensin-converting enzyme and, therefore, various physiological processes, including blood pressure, muscle blood flow, endurance and recovery [13]. ACE gene polymorphism is associated with a number of laboratory parameters, such as lactate and lactate threshold level. The ability to maintain low lactate levels is associated with the II genotype of this gene, which provides better aerobic endurance. Low lactate levels help athletes maintain intense loads longer and recover faster [17, 34].

VO2max levels are often higher in individuals with the II genotype, which improves oxygen exchange efficiency and reduces cardiovascular stress during prolonged exercise [3]. Individuals with the D/D genotype have significantly higher ACE activity, which is associated with higher muscle blood filling, increased blood pressure, and enhanced strength performance [15].

Genotypic variations of the ACE gene also have different effects on physiological indicators depending on the type of sport and the nature of the load. Thus, genotype II (insertion) is considered favorable for athletes involved in cyclic sports, such as marathon, swimming and cross-country skiing [5,12]. Studies show

that athletes with genotype II can demonstrate higher aerobic endurance and low levels of lactate accumulation , improved VO2max, and resistance to prolonged loads. Genotype D/D (deletion), in turn, is associated with strength indicators, improved muscle blood supply and rapid activation of muscle fibers, which is useful for speed-strength sports (Woods et al ., 2001). Laboratory parameters in athletes with the D/D genotype include higher lactate levels during intense exercise, which reflects their ability to withstand short-term strength exercises, as well as increased activity of angiotensin converting enzyme, which affects muscle growth and strength [2,10,21]. The I/D genotype is an intermediate variant and is suitable for mixed sports. Athletes with the I/D genotype show parameters similar to both strength and aerobic characteristics, which allows them to combine strength and endurance loads [1].

Athletes with the II genotype tend to have better recovery processes, as their blood supply system supplies muscles with oxygen more efficiently, which reduces muscle tension and promotes the removal of waste products such as lactate [28]. The D/D genotype, although promoting rapid muscular adaptation and strength gains, may also be associated with higher levels of inflammatory markers after intense training. This may indicate the need for additional recovery measures for such athletes [9].

Studies have shown that the D/D genotype may be associated with increased blood pressure and tachycardia during exercise, especially anaerobic exercise. Genotype II, on the contrary, is associated with a more stable heart rate and cardiovascular resistance to prolonged exercise [19,31].

Blood filling parameters: High ACE activity in the D/D genotype promotes better blood supply to muscles, which increases the rate of delivery of nutrients and oxygen in the short term. However, this benefit may be less relevant for endurance activities that require stable cardiovascular function [6].

Based on the literature data presented, ACE gene polymorphism has a significant impact on physiological and laboratory parameters that may be important

when choosing a sports specialization, developing training programs and recovery methods.

The product of the ACTN3 gene, the protein α-actinin-3, is found in fast muscle fibers and is involved in maintaining their structure and strength [1,4,8]. The ACTN3 gene has a polymorphism R577X, in which the R allele is associated with the performance of muscle fibers, and the X allele leads to the absence of α-actinin-3 (North et al ., 1999). The RR genotype is often associated with high explosive strength and speed, useful in power and speed sports such as sprinting and weightlifting. The R/X genotype is also associated with good performance in power sports, but with a smaller effect than RR (Yang et al ., 2003). The X/X genotype is generally associated with endurance performance, as the absence of the α-actinin-3 protein may improve metabolism during aerobic exercise.

Extensive studies have confirmed the high prevalence of the RR genotype among sprinters and strength athletes, while the X/X genotype is more common in marathon runners and skiers. The ACTN3 gene encoding the α-actinin-3 protein has been widely studied in sports genetics. The R577X polymorphism of the ACTN3 gene (variants of the R and X alleles) has a significant effect on muscle properties, as well as laboratory parameters such as lactate levels , muscle strength, muscle contraction velocity, and recovery. We will consider these relationships in a review of the literature [20,32,38].

The R577X polymorphism of the ACTN3 gene is associated with various laboratory parameters that affect physical performance. Thus, the R/R and R/X genotypes provide improved strength and power characteristics. Athletes with these genotypes usually have higher maximal strength and muscle contraction velocity. The X/X genotype is associated with lower speed and power, but it can promote metabolic efficiency during aerobic exercise. These athletes demonstrate lower peak power, but can maintain stable performance over long distances [7,14]. The R/R genotype promotes a higher rate of lactate accumulation during intense physical activity (Vincent et al ., 2007). This is due to the fact that muscle fibers fatigue faster and require recovery. The X/X genotype typically exhibits lower lactate levels and a

higher anaerobic threshold, making it more suitable for sports requiring prolonged endurance, such as long-distance running and swimming [4, 23,35]. The X/X genotype is better adapted to prolonged exercise due to its improved ability to utilize energy in aerobic metabolism. This is due to the fact that the absence of α-actinin-3 makes muscles more metabolically efficient during prolonged exercise. The R/R and R/X genotypes are less resistant to prolonged exercise, since their fibers expend energy faster, but can develop greater power during short-term exercise [16,22].

The R/R genotype is associated with higher muscle fatigue and longer recovery after intense exercise. Athletes with this genotype have been shown to have higher levels of creatine kinase (a marker of muscle damage) after exercise [30]. Athletes with the X/X genotype have faster recovery due to lower intensity of muscle damage and better use of aerobic metabolism to remove waste products such as lactate [18].

Athletes with the X/X genotype may have a lower heart rate during prolonged exercise, which helps them perform more efficiently in aerobic mode [29]. The X/X genotype is often associated with a higher mitochondrial density in muscle, which improves oxygen utilization. This is especially important for endurance athletes, as it promotes improved energy utilization and resistance to fatigue [30].

Literature data confirm that ACTN3 gene polymorphism has a significant impact on laboratory parameters related to muscle strength, power, lactate levels and recovery after exercise. These characteristics play an important role in predisposition to various sports.

Gene PPARGC1A (coactivator peroxisome activated receptor γ subtype 1-alpha) is involved in the regulation of mitochondrial biogenesis and oxidative metabolism, which is important for endurance and aerobic performance (Lucia et al ., 2005). One of the most studied polymorphisms is Gly482Ser.

The Ser allele is associated with improved aerobic endurance and mitochondrial metabolic efficiency, which is important for athletes involved in cyclic sports. The Gly allele is more common in athletes specializing in strength training, although the association is weaker than that of Ser [1,, 26,33]. Research: Research

has shown that the presence of the allele Ser is correlated with higher aerobic performance in marathon runners and cyclists [7,19,22].

The Gly482Ser polymorphism of the PPARGC1A gene is associated with various physiological and laboratory parameters that affect endurance, energy utilization, and recovery (Eynon et al ., 2010). The Ser allele is often associated with higher VO2max, as it promotes increased mitochondrial density and oxygen efficiency. This makes the Ser allele particularly beneficial for marathon runners, cyclists, and other endurance athletes [27]. The Gly allele shows a lower correlation with VO2max and is more common in strength athletes, although the difference between these groups is not always significant. The presence of the allele Ser promotes higher mitochondrial density, which increases the capacity of muscle cells for aerobic metabolism and improves resistance to fatigue (Ruiz et al ., 2010). The Gly allele is less associated with mitochondrial density, which may reduce endurance performance in athletes [1,37].

Athletes with the allele Ser often exhibit lower lactate levels and a higher lactate threshold, which helps them sustain intense exercise longer without rapidly accumulating fatigue [4,22] . allele Gly can accumulate lactate more rapidly under high loads, which limits their endurance, but is suitable for short-term, explosive activity [24]. The Ser allele is associated with increased fatty acid oxidation capacity and improved metabolic adaptation, which is beneficial for long-distance athletes [11,21,34].

Gly allele has less effect on fatty acid oxidation, which may reduce endurance, but potentially improves anaerobic performance, making it useful for strength sports [1,36].

Athletes with the allele Ser often recover more quickly from aerobic exercise because improved mitochondrial function facilitates faster removal of waste products [1,12]. In athletes with the allele Gly may exhibit higher levels of inflammatory markers after intense exercise, which is associated with greater anaerobic stress and higher lactate levels [21,39].

Athletes with the allele Ser may exhibit lower heart rates during prolonged aerobic exercise, which helps reduce cardiovascular stress and improve cardiovascular efficiency. Research suggests that athletes with the allele Ser alleles are better able to conserve glycogen stores, allowing them to maintain high aerobic performance for longer [13,29]. The Gly allele may result in faster glycogen depletion, which reduces endurance over long distances [8,31].

A review of the literature confirms that allelic variations in the PPARGC1A gene have significant effects on laboratory parameters related to aerobic endurance, lactate levels , mitochondrial density, and recovery. The Ser allele is often associated with improved aerobic endurance, higher VO2max, and improved recovery characteristics, making it advantageous for endurance athletes. In contrast, the Gly allele is more suitable for athletes requiring anaerobic power, such as power athletes.

Discussion. The relationship between allelic variations of the ACTN3 gene and the morphofunctional status of athletes has been established and well documented in sports genetics. The ACTN3 gene encodes the α-actinin-3 protein, which is present in fast muscle fibers responsible for muscle contraction during powerful and high-speed loads. The R577X polymorphism (variants of the R and X alleles) leads to different genotypes: R /R, R/X and X/X, which differently affect physical performance, including muscle strength, contraction speed, endurance and recovery.

The R577X polymorphism of the ACTN3 gene has a significant impact on the morphofunctional characteristics of athletes. The R/R and R/X genotypes are associated with high muscle strength and power, which makes them advantageous for speed-strength sports. The X/X genotype, on the contrary, promotes aerobic endurance, increased capillarization and improved mitochondrial function, which is suitable for endurance athletes.

The relationship between allelic variations of the PPARGC1A gene and the morphofunctional status of athletes has been confirmed by studies. The PPARGC1A gene (coactivator of the receptor gamma, peroxisome proliferator- activated receptor -1α) plays an important role in the regulation of energy metabolism, mitochondrial biogenesis and muscle adaptation to exercise. Polymorphisms of this gene, especially

Gly482Ser, affect aerobic capacity, recovery, muscle endurance and metabolic parameters that are important for athletes.

Conclusion. The combined effect of the studied genes - ACE, ACTN3 and PPARGC1A - can create complex genotypic profiles that help predict predisposition to success in certain sports disciplines. For example, a combination of II (ACE), X/X (ACTN3) and Ser (PPARGC1A) genotypes can predict success in endurance sports, while D/D (ACE), R/R (ACTN3) and Gly (PPARGC1A) are more suitable for strength and speed disciplines.

Based on the analysis of literature data, it can be stated that genotypic variations in ACE, ACTN3 and PPARGC1A have a significant impact on athletic performance and efficiency. However, it is important to consider that athletic success is determined not only by genetics, but also by environmental and social factors, as well as training methods.

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