BRITISH MEDICAL JOURNAL

British Medical Journal

Volume 5, No.2, April 2025

Internet address: http://ejournals.id/index.php/bmj
E-mail: info@ejournals.id
Published by British Medical Journal
Issued Bimonthly
3 knoll drive. London. N14 5LU United Kingdom
+44 7542 987055

Requirements for the authors.

The manuscript authors must provide reliable results of the work done, as well as anobjective judgment on the significance of the study. The data underlying the work shouldbe presented accurately, without errors. The work should contain enough details andbibliographic references for possible reproduction. False or knowingly erroneous statementsare perceived as unethical behavior and unacceptable.

Authors should make sure that the original work is submitted and, if other authors'works or claims are used, provide appropriate bibliographic references or citations. Plagiarismcan exist in many forms - from representing someone else's work as copyright to copying orparaphrasing significant parts of another's work without attribution, as well as claimingone's rights to the results of another's research. Plagiarism in all forms constitutes unethicalacts and is unacceptable. Responsibility for plagiarism is entirely on the shoulders of theauthors.

Significant errors in published works. If the author detects significant errors or inaccuraciesin the publication, the author must inform the editor of the journal or the publisher aboutthis and interact with them in order to remove the publication as soon as possible or correcterrors. If the editor or publisher has received information from a third party that thepublication contains significant errors, the author must withdraw the work or correct theerrors as soon as possible.

UDC 612.13:796015.52+57517

GENETIC DETERMINANTS OF SPORTS QUALITIES: INFLUENCE OF ALLELIC-GENOTYPE VARIATIONS ON ENDURANCE AND STRENGTH

Muminov Aziz Alisherovich
Sadikov Abdushukur Abdujamilevich
Republican Scientific and Practical Center for Sports Medicine, Tashkent.National Anti-Doping Agency of Uzbekistan, Tashkent.

Introduction Genetic research is increasingly gaining attention in the fields of science and sports medicine [3]. Identifying genetic predispositions can help predict athletes' performance and develop personalized training programs [1,5]. Cardiovascular-related genes play a key role in endurance and physical fitness, which is especially important for sports with high aerobic loads. Below is an overview of key genes and their polymorphisms that are important for athletic performance [2,7,12].

ACE gene (Angiotensin - Converting Enzyme is one of the most studied in the context of athletic performance [8]. It encodes an enzyme that is involved in the regulation of blood pressure and electrolyte balance via the renin-angiotensin system [17]. The I/D (insertion/deletion) polymorphism of ACE has been studied in various sports disciplines: the I allele is associated with increased endurance, as it improves aerobic capacity and oxygen efficiency [21]. Studies show that this allele is more common in athletes involved in endurance sports (marathon runners, triathletes) [10]. The D allele is more is common among athletes involved in strength or speed sports such as sprinting or weightlifting [3,19]. Research supports a link between I/D polymorphism and physiological adaptations to training, such as increased cardiac output and resistance to hypoxia. This polymorphism has been studied in relation to physical performance in athletes, especially in terms of endurance and strength, making it an important marker in sports genetics [4].

Numerous studies have shown that the I/I genotype is associated with increased aerobic endurance. Carriers of the I allele have lower levels of angiotensin II, which may contribute to improved cardiovascular performance and more efficient use of oxygen, which is important for endurance athletes [15]. In 1999, Montgomery H. E. published one of the first studies linking the I/I genotype with success in endurance sports. The study was conducted among British climbers and found that carriers of the I allele have better results at high altitude, where aerobic capacity is especially important. In the same year, Myerson S. confirmed that athletes with the I/I genotype demonstrate better results in marathon running and triathlon, which indicates a significant role of this polymorphism in aerobic performance. Studies were also conducted among the military, so in 2001, a team of researchers (Woods DR et al.) showed that carriers of the I/I genotype are superior in endurance and cardiorespiratory adaptation to those with a predominant D allele. Today, it is known that the D allele, on the contrary, is associated with an increase in ACE activity and angiotensin II levels, which can contribute to increased muscle strength and muscle contraction speed [11]. This makes the D/D genotype preferable for strength sports that require explosive strength and power[13]. This is confirmed by the scientific works of such authors as Rigat B. (1990), Woods DR (2000). These authors showed that carriers of the D allele have advantages in strength sports such as sprint and weightlifting, where high power and intensity of muscle contractions are important. Research by Taylor RR in 2004 also confirmed that athletes participating in strength sports more often have the D/D genotype and this is associated with their advantage in maximum power and explosive strength.

The I/D genotype has also been studied by numerous scientists. It is an intermediate variant between I/I and D/D. Athletes with this genotype can demonstrate both moderate endurance and strength, which makes them versatile in various sports. As studies have shown, athletes with the I/D genotype can be successful in both aerobic and strength disciplines, but without an obvious predominance in one of them (Bouchard C. et al., 1999; Jones A. et al., 2002). In their studies, they drew attention to the fact that carriers of the I/D genotype demonstrate moderate results in endurance and strength, and their reactions to training can be more flexible compared to carriers of other genotypes [21]. In order to identify successful athletes, it is necessary to study the frequency of occurrence of I and D alleles. It can vary depending on ethnicity, which can affect sports results in different populations. The distribution of the I/D polymorphism in different populations is heterogeneous, with the D allele being more common in African and European populations, while the I allele is more prevalent in Asian populations [9]. This may explain differences in dominance in certain sports at the international competition level (Wang G. et al., 2013). Also, in her study, Hruskovicova H. (2006) noted that ethnic differences in allele frequency may reflect adaptation to certain climatic and geographical conditions. Studies show that ACE gene polymorphism may affect adaptation to training loads and recovery after physical activity: Thus, carriers of the I/I genotype recover faster after long-term aerobic exercise, which allows them to train more intensively without the risk of overfatigue (Puthucheary ZA et al., 2011); athletes with the D/D genotype respond better to strength training, with increases in muscle mass and strength after short-term intense training (Gomez - Gallego F . 2009).

The ACTN3 (α-actinin-3) gene is one of the most studied genes in sports genetics, affecting human physical abilities, especially muscle strength, speed and endurance. Polymorphism at position R577X of the ACTN3 gene causes significant variations in the expression of the α-actinin-3 protein, which is present in fast muscle fibers responsible for fast and powerful movements [6]. People with R alleles exhibit better performance in sprint and strength disciplines, since the α-actinin-3 protein contributes to the efficient work of fast muscle fibers [3]. The R allele encodes a functional α-actinin-3 protein, while the X allele is associated with a mutation leading to the absence of this protein [1,17]. This creates three possible genotypes: a combination of R/R, when both alleles are functional (characterized by advantages in strength sports); the RX combination, where one of the alleles is functional (implies a mixed phenotype); X/X, where there is a lack of a functional protein (associated with endurance adaptations), making it more common among marathon runners and skiers. Studies have shown that ACTN3 variations can affect the rate of muscle recovery and the risk of injury [16].

In 1999, North KN, Yang N. and co-authors were the first to establish the R577X polymorphism in the ACTN3 gene, associating it with physical activity. Their study showed that professional sprinters have a higher frequency of the R allele, associated with higher expression of the α-actinin-3 protein. Many scientists have studied this gene from the standpoint of ethnic and geographic differences. For example, Mills M. and co-authors in 2001 found that the frequency of the X allele varies significantly in different populations. For example, in Asian populations, the X/X genotype is more common, while in African populations, the R allele is more common. This may explain cultural and ethnic differences in sports preferences and the dominance of certain sports in different regions of the world. And already in 2013, Wang G. and his team confirmed the presence of ethnic differences in the allele frequency and suggested that the frequency of the X allele is higher in populations leading a traditional lifestyle associated with endurance, which could have been evolutionarily fixed as an advantage for survival. The studies of the ACTN3 gene did not end there and in 2003, Yang N. and co-authors conducted a study among elite athletes and found that carriers of the X /X genotype are less common among sprinters and weightlifters, since they lack the α-actinin-3 protein, which is necessary for rapid muscle contraction. And already in 2007, MacArthur DG and North KN confirmed the role of the R / R genotype in strength and speed sports. They found that athletes participating in sprint disciplines and weightlifting more often have the R / R genotype, which is associated with more efficient work of fast muscle fibers. It is also noted that people with the XX genotype are less represented in these disciplines. All these studies were aimed at studying the allelic-genotypic variations of the ACTN3 gene with strength and sprinting abilities. However, in parallel with these studies, other scientists studied the same gene, but associated it with such a sports quality as endurance. So in 2007, Vincent B. and co-authors studied the effect of the ACTN3 genotype on the aerobic abilities of athletes and found that carriers of the X / X genotype more often demonstrate excellent results in sports requiring long-term endurance, such as marathon and triathlon. The absence of α-actinin-3 can stimulate the body's adaptation to endurance loads through the activation of slow muscle fibers. In 2009, Eynon N. and co-authors conducted a meta-analysis that showed that athletes with the X/X genotype excel in sports related to aerobic endurance.

The study included many elite athletes and found that the frequency of the X/X genotype was higher in marathon runners than in the control population. In addition to the athletic performance of the athlete, scientists studied the effect of ACTN3 polymorphism on adaptation to training. In 2010, Zempo H. et al. found in their studies how differences in ACTN3 genotypes affect muscle adaptation to training in older people. It was found that R/R carriers build muscle mass faster in response to strength training, while X/X carriers showed a better response to endurance exercises. In 2014, a group of researchers (Petr M. et al.) found that the lack of α-actinin-3 in people with the X/X genotype is compensated for by increased activity of other structural muscle proteins. Such a substitution improves recovery and adaptation to long-term physical exertion, which is especially important for endurance athletes.

The world literature contains numerous studies devoted to the study of allelic-genotypic variations of PPARGC1A. This gene is key in sports genetics, since it affects aerobic capacity, lipid and carbohydrate metabolism, as well as endurance. Gene PPARGC 1 A (Peroxisome Proliferator - Activated Receptor Gamma Coactivator 1 Alpha) plays an important role in mitochondrial biogenesis and metabolic regulation [18]. Its Gly482Ser polymorphism has been associated with aerobic capacity and adaptation to endurance training: the 482Ser allele is associated with better endurance and oxygen metabolism efficiency [20]. This gene also plays a role in cardiovascular adaptation to training, as it regulates the development of new mitochondria and an increase in cardiac output. One of the key variations in the PPARGC1A gene is the Gly482Ser polymorphism (rs8192678), where a glycine is replaced by a serine at position 482. This polymorphism has been studied for its effects on endurance and adaptation to exercise [2,6,22]

Studies show that the Gly482Ser polymorphism affects aerobic endurance in athletes, fat oxidation capacity, and training adaptation [1,3,11]. G/G genotype carriers demonstrate improved aerobic endurance and mitochondrial biogenesis capacity, which contributes to more efficient fat and carbohydrate oxidation [23]. Scientists have found that athletes with the G/G genotype are more often represented in endurance sports, such as marathons and triathlons. These athletes showed better mitochondrial adaptation capacity and improved energy utilization, which positively affects their performance in aerobic exercise (Lucia A., 2005; Eynon N. et al ., 2011). Carriers of the S/S genotype have a reduced ability to oxidize fats and mitochondrial activity, which may negatively affect the results in sports requiring endurance. In 2009, the results of studies by a group of scientists were published, they found that carriers of the S/S genotype demonstrate worse performance in endurance tests compared to carriers of the Gly allele . They suggested that this genotype is associated with reduced expression of PGC-1α and a lower capacity for metabolic adaptation (Yvoine E. et al., 2009)

Several studies have found that G/S heterozygotes exhibit intermediate performance in aerobic tests and training adaptations. A study of long-distance runners found that G/S carriers were intermediate between G/G and S/S in endurance performance. This suggests that heterozygotes have a moderate capacity for mitochondrial biogenesis and fat oxidation (Stepto NK et al., 2007).

Research by other authors shows that the polymorphism of the PPARGC1A gene can influence the specialization of athletes in certain sports. Thus, the Russian scientist Akhmedov I.I. (2010) conducted a study among Russian athletes, showing that carriers of the Gly allele are more often represented in endurance sports, such as long-distance running and cycling, while carriers of the Ser allele are less common among elite athletes.

The frequency of the Gly and Ser alleles varies by ethnicity, which may influence the physical performance of athletes in different populations. Studies have shown that the Gly allele is more common in European populations, which may explain the dominance of Europeans in endurance sports. In contrast, the Ser allele is more common in Asian populations, although its role in sports genetics in this context requires further study.

Thus, the literature review showed that polymorphisms of the ACE, ACTN3, PPARGC1A, VEGF and NOS3 genes play an important role in regulating the cardiovascular system in athletes, influencing endurance, adaptation to physical activity, recovery from training and overall performance. Let us consider in more detail the role of these polymorphisms:

Conclusion Genetic studies in sports science allow us to more accurately predict the physical capabilities of athletes and their adaptation to training. Polymorphisms of genes such as ACE, ACTN3, PPARGC1A play an important role in the functioning of the cardiovascular system and affect endurance, strength and recovery. These genes affect factors such as blood supply, cardiac performance, oxygen capacity and the rate of recovery after exercise. Their variations can help determine the predisposition to success in various sports disciplines and facilitate the development of personalized training programs. These data can be used to individualize training programs to optimize athletic performance depending on the genetic predisposition of athletes, select promising athletes, and develop genetically based recommendations in the field of sports and health. In the future, understanding these genetic variations can help create personalized training programs to maximize athletic performance and prevent injuries.

Used literature.

1. Akhmetov I.I. Molecular genetics of sport. Monograph. – M.: Sovetsky Sport, 2009. – 268 p.

2. Akhmetov I.I., Popov D.V., Mozhayskaya I.A., Missina S.S., Astratenkova I.V., Vinogradova O.L., Rogozkin V.A. Association of polymorphisms of regulatory genes with aerobic and anaerobic performance of athletes // Russian Physiological Journal named after I.M. Sechenov. - 2007. - Vol.93. - No.8. - P.837-843.

3. Akhmetov I.I. Genetic testing in the system of medical and biological support of physical culture and sports. Methodological manual. – Kazan: KSMA. – 2012. – 72 p.

4. Voroshin I.N., Akhmetov I.I., Astratenkova I.V. Association of gene polymorphisms with the level of development of special endurance in 400-meter runners // Scientific notes of P.F. Lesgaft University. - 2007. - No. 3. - P. 9-15.

5. Akhmetov II, Donnikov AE, Trofimov DY ACTN3 genotype is associated with testosterone levels of athletes // Biology of Sport. – 2014. – V.31(2). - P.105-108.

6. Baranova TI, Berlov DN, Glotov OS, Korf EA, Minigalin AD, Mitrofanova AV, Ahmetov II, Glotov AS Genetic determination of the vascular reactions in humans in response to the diving reflex // American Journal of Physiology-Heart and Circulatory Physiology. – 2016. DOI: 10.1152/ajpheart.00080.2016

7. Druzhevskaya AM, Akhmetov II, Astratenkova IV, Rogozkin VA Association of the ACTN3 polymorphism with power athlete status in Russians // European Journal of Applied Physiology. – 2008. – V.103(6). – P.631–634.

8. Egorova ES, Borisova AV, Mustafina LJ, Arkhipova AA, Gabbasov RT, Druzhevskaya AM, Astratenkova IV, Akhmetov II The polygenic profile of Russian football players // Journal of Sports Sciences. – 2014. – V.32(13). – P .1286-1293.

9. Foster BP, Morse CI, Onambele GL, Ahmetov II, Williams AG Genetic variation, protein composition and potential influences on tendon properties in humans // The Open Sports Medicine Journal. – 2012. – V.6. – P.8-21.

10. Gineviciene V, Jakaitiene A, Aksenov M O , Aksenova AV, Druzhevskaya AM, Astratenkova IV, Egorova ES, Gabdrakhmanova LJ, Tubelis L, Kucinskas V, Utkus A. Association analysis of ACE, ACTN3 AND PPARGC1A genes polymorphisms in two cohorts of European strength and power athletes. Biol Sport. - 2016. - V.33(3). - P.199-206.

11. Leońska-Duniec A., Ahmetov II, Zmijewski P. Genetic variants influencing the effectiveness of training programs in obesity – an overview of human studies // Biology of Sport. – 2016. – V.33. – P.207-214.

12. Maciejewska A., Sawczuk M., Cięszczyk P., Mozhayskaya IA, Ahmetov II The PPARGC1A gene Gly482Ser polymorphism in Polish and Russian athletes // Journal of Sports Sciences. – 2012. – V.30(1). – P.101-113.

13. Malczewska- Lenczowska J., Orysiak J., Majorczyk E., Zdanowicz R., Szczepańska B., Starczewski M., Kaczmarski J., Dybek T., Pokrywka A., Ahmetov II, Sitkowski D. Total hemoglobin mass, aerobic capacity and the HBB gene in Polish road cyclists // Journal of Strength and Conditioning Research. - 2016. - V.30(12). - P.3512-3519.

14. Nabatov AA, Troegubova NA, Gilmutdinov RR, Sereda AP, Samoilov AS, Rylova NV. Sport- and sample-specific features of trace elements in adolescent female field hockey players and fencers // Journal of Trace Elements in Medicine and Biology. - 5 November 2016. - DOI: 10.1016/j.jtemb.2016.11.002 .

15. Papadimitriou ID, Lucia A., Pitsiladis YP, Pushkarev VP, Dyatlov DA, Orekhov EF, Artioli GG, Guilherme JP, Lancha Jr AH, Ginevičienė V., Cieszczyk P., Maciejewska-Karlowska A., Sawczuk M., Muniesa CA, Kouvatsi A., Massidda M., Calò CM, Garton F., Houweling PJ, Wang G., Austin K., Charlton V., Irving R., Morrison E., Druzhevskaya AM, Astratenkova IV, Ahmetov II, Bishop DJ, North KN, Eynon N ACTN3 R577X and ACE I/D gene variants influence performance in elite sprinters: A multi-cohort study // BMC Genomics. – 2016. - V.17 - 285.

16. Pitsiladis Y., Wang G., Padmanabhan S., Wolfarth B., Fuku N., Lucia A., Ahmetov II, Cieszczyk P., Collins M., Eynon N., Williams A. Genetic testing in sport and exercise // Sport and Exercise Scientist. – 2013. – V.38. – P.14-15.

17. Wang G., Padmanabhan S., Wolfarth B., Fuku N., Lucia A., Ahmetov II, Cieszczyk P., Collins M., Eynon N., Klissouras V., Williams A., Pitsiladis Y., Sawczuk M. Are genetic testing results trustworthy in sports? // Sport wyczynowy . – 2013. – V.3-4(547-548). – P.68-83.

18. Wang G., Mikami E., Chiu LL., de Perini A., Deason M., Fuku N., Miyachi M., Kaneokas K., Murakami H., Tanaka M., Hsieh LL., Hsieh SS, Caporossi D ., Pigozzi F., Hilley A., Lee R., Galloway SDR, Gulbin J., Rogozkin VA, Ahmetov II, Yang N., North KN, Ploutarhos S., Montgomery H., Bailey MES, Pitsiladis YP Association analysis of ACE and ACTN3 in Elite Caucasian and East Asian Swimmers // Medicine and Science in Sports and Exercise. – 2013. - V.45(5). – P.892–900.

19. Wang G., Tanaka M., Eynon N., North KN, Williams AG, Collins M., Moran CN, Britton SL, Fuku N., Ashley EA, Klissouras V., Lucia A., Ahmetov II, de Geus E. , Alsayrafi M., Pitsiladis YP The future of genomic research in athletic performance and adaptation to training // Medicine and Sport Science. – 2016. – V.61. – P.55-67.

20. Webborn N., Williams A., McNamee M., Bouchard C., Pitsiladis Y., Ahmetov I., Ashley E., Byrne N., Camporesi S., Collins M., Dijkstra P., Eynon N., Fuku N ., Garton F.C., Hoppe N., Holm S., Kaye J., Klissouras V., Lucia A., Maase K., Moran C., North KN, Pigozzi F., Wang G. Direct-to-consumer genetic testing for predicting sports performance and talent identification // British Journal of Sports Medicine. - 2015. - V.49(23). - P.1486-1491.

21. Zarębska A., Jastrzębski Z., Moska W., Leońska-Duniec A., Kaczmarczyk M., Sawczuk M., Maciejewska- Skrendo A., Żmijewski P., Ficek K., Trybek G., Lulińska-Kuklik E., Semenova EA, Ahmetov II, Cięszczyk P. The AGT Gene M235T Polymorphism and Response of Power-Related Variables to Aerobic Training // Journal of Sports Science and Medicine – 2016. – V.15(4). – P.616-624.

22. Znazen H., Touhami I., Chtara M., Le Gallais D., Ahmetov II, Chamari K., Soussi N. Genetic advantageous predisposition of angiotensin converting enzyme ID polymorphism in Tunisian athletes // The Journal of Sports Medicine and Physical Fitness. – 2016. – V.56(6). – P.724-730.

23.Zmijewski P., Grenda A., Leońska-Duniec A., Ahmetov II, Orysiak J., Cięszczyk P. Does BDKRB2 gene -9/+9 polymorphism affect training improvements in competitive swimmers? // Journal of Strength and Conditioning Research. – 2016. – V.30(3). – P.665-671.