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Review article

STRUCTURE OF GENES AND RELATION TO THE PHYSICAL ACTIVITY

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Abstract

Genetic features (genome) to be passed down from generation to generation are encoded in DNA sequences. The construction of DNA and the placement of sequences differ from individual to individual. The differences between individuals cause different levels of responses to internal and external stimuli (training loads, training adaptation, environmental adaptation, diet type, composition of muscle fibers, blood pressure level, etc.). To date, only 20 of the more than 200 genes predicted to be associated with athletic performance are thought to affect high-level athletic performance. It has been observed that genetic characteristics are significantly affected by factors arising from environmental interactions and lifestyle, and that physical performance differences between individuals are directly related to exercise or lifestyle. So many foreign factors come together on a sports event, even if one competitor is considered superior to another, the result is almost always doubtful. It is obvious that ideal genes probably drive an athlete to greatness, and at the same time, these good genes do not guarantee a result.

Keywords: genes, lifestyle, physical activity.

1. Introduction

Genetics is the investigation of the source of functional behaviors that are shaped by the change of the organism, lifestyle, and environmental factors, or the internal causes of variation of living organisms, by considering physiological parameters. These small changes, transmitted from generation to generation between people, create differences between individuals (1). The genes in which the codes of genetics are located are a structuring consisting of a physical and chemical inheritance unit consisting of deoxyribonucleic acid (DNA), localized on the chromosome, of different lengths of mutually matched base-pair sequences (sequence), carrying the codes of the hereditary characters of cells and therefore of living things, proteins, enzymes, and other macro and micro molecules necessary for their lives, and also examining the control and progress of genetic, biochemical, physiological and similar events. This configuration also leads to the regulation of thousands of genes on chromosomes and thirty-seven genes of mitochondrial DNA (mtDNA) located in cell cytoplasm.

Genetic features (genome) to be passed down from generation to generation are encoded in DNA sequences. Sequences consist of four different elements (nitrogen base pairs) adenine (A), thymine (T), cytosine (S), and guanine (G). A, T, C, and G nucleotides link with each other to make a single DNA sequence (AACGGT). Each nitrogen base binds to another essential base (thymine binds to adenine and cytosine to guanine) to form a double-stranded DNA helix (2, 3, 4, 5, 6). The construction of DNA and the placement of sequences differ from individual to individual. The aforementioned differences between individuals cause different levels of responses to internal and external stimuli (training loads, training adaptation, environmental adaptation, diet type, ratio or composition of muscle fibers, blood pressure level, etc.), (7).

DNA molecules are genetic determinants of biological properties. There are about 3 billion nitrogen base pairs in the human genome that allow them to form their characters. The emergence of these features is due to the different sequencing of the sequences or sequences are located in peptide chains and encode a specific amino

acid called a codon. The variations that occur as a result of this small difference are seen as single base pair change, single base pair deletion, single base-pair insertion-addition, or insertion. The most common human genome change occurs in a single base pair. This type of change is called a single nucleotide polymorphism (SNP) or point mutation. The differences or changes that occur in this context depend on the prevalence in the population and the effect on the phenotype (appearance), (7, 8, 9).

2. Characteristic Reflections of DNA Sequences

The base sequences and numbers that make up the genes are stable within the species and have very important roles in the protection of the species in question.
However, due to the determination of each amino acid with one or more specific codons, the decrease in the sensitivity of enzymes during the replication and polymerization (the combination of small molecules to form large molecular weight components), the wrong bases to line up, or the presence of factors that will trigger mutations, and so on, although rare, lead to some changes between the base sequences and cause the development of mutations. At the end of such changes, new generations (mutants) with different genotypes and phenotypes are formed from cells that divide and multiply (3, 4). While gene expression is influenced by multiple environmental factors, genetic predisposition is a critical factor in the development of athletic performance and is characterized by multiple gene polymorphisms (7, 9, 10). Although all the sequences in the DNA strands are encoded (encrypted), approximately 95% of the genome plays a fundamental physiological role. Especially in the regulation of gene expiration, it is very important whether genes are transported to RNA to produce proteins. There are two copies of each gene in the human genome, with one gene located in the specific region of the specific chromosome, while another gene (the same gene not in the same sequence) can be found in the same location as the homologous chromosome. One or two copies of the most common variable in individuals are normal copies, the least frequent variable with one or two copies is the mutation copy. Genotype changes occurring in this framework are homozygous normal (two alleles, a pair of genes one maternal and one paternal, the normal copy gene), heterozygous carrier (one allele normal copy, one allele changed), and homozygous mutant (two alleles are polymorphic or mutated, with both the maternal and paternal copy altered). Created within the framework of the human genome mapping program initiated by an international group formed with the participation of different countries in 1990, the "Human Genome Project" has provided significant impetus to developments related to health and exercise. Most of the project in question was completed in 2000 and the chemical codes of DNA were decoded. However, considering the multi-gene and multi-factor variations, it is certain that a little more time is needed to identify the sources of the disease, athletic performance development, and similar causes by determining the locations of DNA sequences and the functions they are associated with (11, 12, 13, 14, 15, 16, 17, 18, 19, 20).

3. The relationship between physical activity and gene

Unless specific confounding effects occur within populations, both alleles and genotype frequencies remain constant and are passed from generation to generation (Hardy-Weinberg Equilibrium), (4). In the case of body composition, heart rate, and similar genetic-based (multi-gene and multifactorial) traits, it is generally accepted that many genes, each gene separately, affect these traits cumulatively or simultaneously. Random variations in nucleotide sequences passed down from generation to generation between humans create differences between individuals (height, the color of hair or eyes, athletic performance and level of obesity, hypertension, the likelihood of suffering from metabolic and coronary diseases, etc.), (11).
Physical activity or performance measurement values are directly influenced by the instruments used, making it difficult to understand the relationships between performance and inheritance and different parameters. Although the accuracy of the total energy loss calculations measured considering the short and long-term loads is quite high, it is also known that direct physiological measurements of physical activity are generally

expensive to be used in large epidemiological studies and are not practical enough. The ability to identify genes associated with athletic performance depends on whether the behavior of physical activity is targeted, the state of the environmental conditions, and the level at which the variability revealed by the gene or genes studied affects athletic performance. Genetic structure affects the individuals' fitness level (4) and adaptation to exercise. Individuals with the same genotype structure show similar reactions to the same type of exercise load (21). In high-level sports disciplines, alternative routes for the talent required for future athletes to specialize depend largely on objective physical, technical and tactical characteristics. Genetic markers have been associated with a general increase in improved aerobic capacity, strength and athletic abilities, including several single nucleotide polymorphisms. While some of the candidate genes that reveal athletic ability show inconsistencies in individual performance development, it is known that both ACE and ACTN3 polymorphisms are accepted as directly related candidate genes in strength and endurance parameters. Recently, many candidate genes that have been reported to be associated with exercise effort have been identified, but inhomogeneous studies and activities where different energy systems are predominantly used make it difficult to identify the relationship between gene and athletic performance (4).

It is obvious that genes that affect physical performance interact with similar variables such as the external environment and lifestyle (4, 15). The reality that allows some to be perfect in 10,000 hours, others in 3000 hours is genetics. Many internal and external demographic factors have been studied in relation to exercise. Among these variables, the most prominent reflection of the mobility behavior is the wealth level, social lifestyle and gender, while the chronological age, body composition, racial characteristics arising from genealogy and similar parameters are ineffective criteria for exercise participation and continuity (4, 22). Not only metabolism but also anthropometric and biomechanical factors are factors that increase elite performance. In some studies, to date, the role of gene variants that play a dominant role in both aerobic and anaerobic activities in endurance running performance has not been fully proven. Studies have not found sufficient evidence regarding the responses of endurance ability to aerobic capacity development as a result of extensive training. It is also stated that epigenetic modifications may play a role in inconsistent findings revealed by candidate genes (23, 24, 25).

4. Candidate genes affecting athletic performance

To date, only 20 of the more than 200 genes predicted to be associated with athletic performance are thought to affect high-level athletic performance. Among these, the genes whose relationship with performance has been studied most are the Angiotensin Converting Enzyme (ACE) gene and the alpha-actinin-3 (ACTN3) gene (26). Angiotensin-converting enzyme (ACE) and alpha-actinin-3 (ACTN3) performance genes, which determine blood pressure and muscle contraction rate, are also highly effective genes in determining muscle strength (athletic, muscular, and large bodies), contraction speed, and endurance (slim). The I allele of the ACE genotype was associated with improved endurance performance, while the D allele was associated with a higher ACE activity, increased strength, and sprint performance. People with high ACE enzyme activity (D allele) cannot provide sufficient blood flow to the muscle tissue due to increased vasoconstriction, so they can be quite successful in endurance sports, on the contrary, they can be quite successful in short distance activities. Studies revealing the relationship between ACE genotype and training show that DD genotype provides an advantage in short-term aerobic performance improvement (26). The second most widely studied candidate gene within the context of athletic performance improvement is the ACTN3 gene, which encodes the α-actinin-3 protein. There are two variations of the ACTN3 gene, one of which carries more alpha-actinin-3 protein to fast-twitch muscle fibers (R allele). The R allele is generally advantageous in sports with a predominant force-oriented explosive force, whereas the X allele in which actin is not producing protein is associated with low sprinting ability and muscle strength. It is known that the ACTN3 genotype is an important genetic variant that affects metabolic function in muscle contraction. The production of alpha-actinin-3 enables strong contraction of skeletal muscles. Genetic factors play an important role in parameters related to athletic performance such

as power, strength, and aerobic capacity (26, 27).

5. Environmental factors and effects of genetic structure on physical activity habits

There is not much information available to suggest that health and wellness, attitudes, normative beliefs, or susceptibility to disease predict raising the mobility threshold. In a recent study, it was stated that the effect of the habitat that is supposed to trigger mobility in adults is not at the expected level (2, 3). The most important reasons for the deviations caused by the variables of gene sequences in the threshold of mobility are intellectual life and the area of activity in which it is located (3, 4, 5). The reflections of social lifestyle and movement areas other than body composition, age, gender, and similar variables that determine the mobility threshold are not taken into consideration (3). It has been observed that children of families with high activity level have higher mobility thresholds than their inactive peers. It is obvious that genetic factors contribute to mobility and activity habits (26). Just like sports and exercise-related features, all of the various performance characteristics including maximum oxygen intake, muscle fiber ratio, various strength measurements, flexibility, speed, and ability to train, inter individual variation or differences also have a strong genetic basis (26). Studies focus on determining inter individual variations in DNA sequencing that affect the aforementioned features. In the following years, when the place occupied by the gene for each character in each homologous chromosome is defined, more detailed information will be obtained about the mechanisms regulating each phenotype (18, 27, 28, 29, 30). Genetic structure, the environment and behaviors that are influenced by the social lifestyle are the expressive reflections of the characters. This also applies to anatomical, physiological, and psychological factors that contribute to physical performance improvement.

6. Talent selection and genetic tests

Research in the field of genetics determines the differences in DNA sequences, or the reflections of amino acids encoded or not produced on behavior. However, genetic research of this nature raises troubling ethical questions, and the danger of misuse is perceived as false or true. Genetic studies can be applied to investigate possible factors that underlie human physical performance and are very strong in this regard. Twin and family studies have shown that variability in many exercise-related parameters is partially inherited. These variables include maximum oxygen uptake, workload capacity, anaerobic power, maximum workload rate, muscle fiber type distributions, muscle enzyme concentrations, and the ability to train several of these factors (5, 26).

Field tests are carried out to measure characters (strength, speed, and endurance) specific to the sports discipline. It is the actual genetic background that determines many of these characters that triggers success at the Olympic level (4, 26). There are two main differences between genetic tests and field measurements and especially effort tests. First, any genetic test can produce unexpected results. Studies have shown that the initially modest differences in lipid profile of the APO E4 variant are associated with familial Alzheimer's disease that will emerge later. Second, DNA tests can be done from the moment an individual'sgenomic DNA is taken, and the same can be done before birth. Since DNA is virtually unchanged throughout life, taking DNA from an embryo, a child, or an adult will not make any difference. Although there is not much difference between candidate gene determination and field tests, important clues that may reveal the relationship with diseases can be revealed in the loci in the DNA sequences. In addition, the fact that genetic tests can be performed from the beginning of formation, will provide a predictive perspective on performance improvement that may occur in the future. Many of the athletes are committed to any discipline when they are young and require long-term training in the following years to be distinguished. Learning about the genetic predisposition before or after may be disappointing for the athletes in question. Therefore, in order to prevent such ethical concerns, parents should be counseled before gene testing, and children or young people should be engaged in the sports disciplines they love without any anxiety (28, 29, 30).

It has been observed that genetic characteristics are significantly affected by factors arising from environmental

interactions and lifestyle, and that physical performance differences between individuals are directly related to exercise or lifestyle. When candidate genes that reveal athletic ability are used in athletes performing at the elite level, they are important clues that can reveal the scope and intensity of training practices, loading, resting and recovery processes, nutrition and injury risks, and their reflection on the field or podium. However, the suspicion that the tests or candidate genes are approaches or practices that are far from revealing possible predictions of later athletic performance markers in children and adolescents is also puzzling. Athletic performance can be positively or negatively affected by the coordinated reflection of parameters such as genetics (muscle fiber type, aerobic and anaerobic power ratio, training adaptation), psychology, social factors, physical condition, the knowledge and experience of the trainer, and the athlete's ability and good planning. Genetic heritage, environmental factors, and the character (volume, and intensity) of exercise loads applied within the framework of lifestyle, neuromuscular development, balanced diet, and cultural differences that trigger athletic success can reveal individual changes or differences. Considering all these variables, monitoring and control of performance improvement and athletic success graphs may become more predictable.

7. Conclusion

So many foreign factors come together on the occasion of a sports event, even if one competitor is considered superior to another, the result is almost always doubtful. It is obvious that ideal genes probably drive an athlete to greatness, and at the same time, these good genes do not guarantee a result. While some claim that many years and thousands of hours of training, determination, and dedicated effort are sufficient to produce elite talent, the backgrounds of hereditary studies and athlete families are clear evidence that innate qualities give certain individuals an advantage for athletic endeavors.

Undoubtedly, athletes with a favorable genetic profile who interact with correct training practices are more likely to achieve higher performance levels. However, it is likely that the possible combinations of genetic and environmental factors that result in elite-level performance will remain enormous and often unpredictable. Although only 1 out of 10,000 people has high-level fitness success, it seems highly optimistic that genetic screening will identify that person better than current talent identification strategies.

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