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Genetic factors associated to sports performance

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Abstract

Over the years, sports performance has been associated with several factors that modify the athlete's capacity to adapt to the effort required to perform such activities. Recent studies prove that these factors correspond not only to the training routines that the athletes follow, but also to genetic characteristics that contribute to a better response of the organism through changes that involve processes like protein synthesis to increase the metabolic rate or the production of the components of contractile machinery. Moreover, the genetic traits found in the athletes allow preventing injuries or conditions that could risk the individual's life by identifying characteristic markers of these pathologies.

Keywords: Physical Activity; Sports Performance; Epigenetic Changes; Genetic Variances.

1. Introduction

Physical performance refers to the capacity to do or execute physical tests with the least energy expenditure by understanding the performance and physical condition as different concepts: the physical condition is the sum of all the physical qualities and the performance as the conditional and coordinative capacities [1]. Other factors associated with the environment and training conditions can influence these characteristics. However, finding its relation with genetics has a great importance, since it allows us to explain many of the characteristics of development in athletes. In the present review, we offer a compilation of the most recent studies that analyse phenotypic and genotypic traits in high-performance athletes, as well as a description of some of the genes that influence characteristics like muscle growth, metabolism changes directed to the production of biochemical energy and adaptations to the physical activity.

2. Genetic traits found in high-performance athletes

There are many physical traits that determine the athletic capacity of an individual, mostly the strength of the skeletal muscle and the predominant kind of fibres on it. Other traits include the maximum capacity of oxygen that the body can deliver to the tissues (aerobic capacity), muscle mass, height, flexibility, coordination, intellectual capacity and personality [2]. On average, 66% of the variation in athlete status can be explained by genetic factors. The remaining variation is due to environmental factors, such as deliberate practice, nutrition, ergogenic help, place of birth, availability of medical and social help, and even luck [3]. Some of the genetic factors that we found include the ACTN3 gene, located in the chromosome 11 and responsible for encoding the α-actinin-3 protein (αA3) in the Z-line of the muscle sarcomere of the fast-twitch fibres [4]. The mutations in this gene can occur in the R and X alleles, which allow synthesising the αA3 protein. The combination of these alleles produces the RX, RR and XX genotypes. RR and RX genotypes provide a greater stability to the contractile structure in the muscle fibre, leading to a greater strength

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production. There is an important association between the RR genotype and athletic performance of speed/power, however, its effectivity manifests in the strong anaerobic contractions, besides, this genotype increases the area in the transversal section of the IIa and Ix fibres to favour the short efforts with intensity of 95% or even more. The RR + RX genotypes are related to the athletic status of both speed/power athletes and to the long-distance runners. About the XX genotype, it is very probably its contribution to the aerobic resistance performance by giving a physiological advantage to the bearers of this gene in the competences when the aerobic resistance and muscle efficiency are much more important than the speed and power, and also contributes to modifying the muscle structure to increase the adaptation to exercise and reduce the risk of suffering injuries [5, 6]. In order to discover the genetic markers, which associate more to high-performance athletes, it was carried out a review of a whole-genome sequencing (WGS) [7] and of an exome-wide association study (EWAS) that involves athletic cohorts [8]. However, because the sample size was limited, these research and later replication studies have resulted in the identification of only 13 genetic markers.

3. A brief description about the main genetic variances found in athletes

The high-performance athlete's genotype presents a series of polymorphisms (currently called "*genetic variances*") which are different between the strength and endurance sports. The success in the sports is determined by many genetic traits such as transcriptomic, biochemical, histological, anthropometric, physiological and psychological traits [3]. Many single-nucleotide polymorphisms (SNP) have been discovered that are related directly or indirectly with sports, which are responsible for the development of many physical characteristics found in athletes. There are other factors that influence the presence or absence of polymorphisms, one of them is the race, since it has been demonstrated that the rs671 G/A aldehyde dehydrogenase gene (ALDH2) polymorphism is associated to strength in athletes and non-athletes in the Japanese population. The ALDH2 gene is located in chromosome 12 and is responsible for encoding one of the most important enzymes needed for the hepatic metabolism of the alcohol. The previously mentioned rs671 polymorphism produces lower levels of acetaldehyde through its elimination and the subsequent accumulation in the organism [3, 9], and also, this enzyme has a great importance that lies on its participation to attenuate the oxidative stress and the elimination of endogen lipid aldehydes, while its inactivation leads to effects like muscle loss [10]. In addition, other genes like APC rs58013 genotype are related to the RT (reaction time), which is especially important in wrestlers because it helps to anticipate the movements of their opponent; besides, there have been identified 8 RT alleles of this genotype [11].

4. Epigenetic changes associated with physical activity

The athletes' development, as well as the rest of the people, is a process sensitive not only to internal factors, but also to others external that influence, through DNA modifications, the way in which the genes are expressed, as well as the functions they will play later. Although there has been identified a great percentage of athletes that have genetic traits that allow them to stand out in many activities [3], it is not possible to ignore the epigenetics' importance, who takes and important role in the appearance of characteristics that contribute to the development of these individuals through modifications in the genes expression involved in the physical performance [12]. The epigenetics is a science that, in a short time of having emerged, allowed to establish a link between elements that seemed so distant from each other, like the environment and the genetic material, so we could understand how the first one influence on the appearance of physical characteristics or phenotype in the living beings. One of the most studied epigenetic changes due to exercise is the increase of muscle strength, a process regulated by molecular mechanisms that promote the expression of genes coding for contractile proteins in response to the physiological phenomena experienced during the physical activity [13]. This particular effect is of interest, for the changes that happen in the exercise (e.g. the increase of body temperature) are not properly exogen in character, but they induce to a molecular response that alters tissue functions, like the gluconeogenesis rate and the contractile-protein synthesis, mostly α-actinin-3 [14]. As can be seen, the tissue that most changes experiments during exercise is the skeletal muscle that must adapt to constant stress to continue the contraction during long periods; such changes happen not only at the level of the contractile machinery, but also through the increase of the expression of the genes associated to enzymes that belong to many metabolic pathways in order to keep a constant energy production. In this context, the muscle tissue can be stimulated by the release of hormones and growth factors; besides, some DNA regions that encode enzymes necessary for the metabolism experiment a reduction in methylation (associated to the phenomena called *"gene silencing"*), which causes a higher rate of protein synthesis [13, 15]. In addition to the metabolic changes after the exercise, there is also a size increase on myocytes, which can be partially explained by the previously mentioned effects and because of the proliferation of transcription factors associated with cellular growth. A recent study [16] analysed biopsies of skeletal muscle on different moments after an endurance exercise routine and found that the MYC transcription factor (a protein found in the nucleus of the muscle cell) accumulates in the cell and promotes the protein synthesis and ribosome biogenesis, therefore, it is considered a pro-anabolic factor that contributes to muscle hypertrophy.

5. Relation between nutrigenomics and sports performance

As mentioned before, there are many environmental factors that can influence the athletes' development according to their individual genetic characteristics, among which includes the nutrition. Usually, diets are planned without considering characteristics that can affect the way in which the nutrients are used by the organism, or its capacity to absorb them. It is here where nutrigenomics takes a very important role by establishing a link between these variables and the athlete's genetics, with the purpose of taking full advantage of the individual characteristics through nutrition. Although diets are planned in a personalised way, it is not enough only to consider the energetic requirements of the people, especially in the case of the high-performance athletes, for which there have been implemented techniques like the genetic tests that cover aspects such as the organism's response to the feeding and the way in which the metabolic pathways process the nutrients supplied. The classic examples of these genetic variances are the lactose intolerance and phenylketonuria, which makes it difficult to develop nutrition plans adaptable to a wide range of the population and, besides, demonstrates the importance of individualising the nutrients supply. Retaking the example of phenylketonuria, there are individuals that cannot metabolise other molecules, which results in a risk for their health that can produce serious consequences [17].

Nutrigenomics has seen its progress enhanced by the great genetic diversity expressed in changes like the SNPs, capable of provoking a wide range of metabolic responses. The effects of the physical activity on the metabolism changes and muscle adaptations are due to chromatin changes in different levels, such as DNA and histones hypomethylation, which promotes the transcription and protein synthesis [17, 18].

6. Importance of genome knowledge in athletes' health and performance

Most of the epigenetic changes provoked by physical activity are related to the adaptation of the individual through the improvement of their physiology and endurance, which happens at the level of many tissues and metabolic response. Besides, there are other aspects that, in most of the time, are not considered and are capable of provoking serious damages to the athlete's health. One of these cases is sudden death in young athletes, which constitutes a tragedy because of its unexpected appearance, most commonly in sport competitions. The pathophysiology of this event includes the use of anabolic steroids, stimulants and also endogen factors, such as hypertrophic cardiomyopathy and arrhythmogenic right ventricular dysplasia [19]. There have been identified mutations in genes responsible for encoding ion channels proteins, that were found in many forms of long QT syndromes with familiar origin, which indicates the need of genetic tests in apparently healthy individuals to discard the presence of these anomalies [20]. Furthermore, injuries like tendinopathies are some of the most common manifestations with a genetic origin, mainly due to mutations in collagen encoding genes, related to fractures, osteoporotic fractures and, more frequently, injuries in Achilles and quadriceps tendon, and anterior cruciate ligament rupture [21]. The previously mentioned pathologies represent some of the reasons for which it is necessary to identify the genetic characteristics in athletes, to prevent their manifestations that can constitute a threat to the athlete's performance and even their life.

In order to discover the markers associated with the best performance in athletes, there have been developed many techniques directed to sports-related genetic testing, which allows to identify potential athletes with the most appreciable traits necessary to stand out in a certain discipline [22].

Even when the genes have a great importance in the development of the athlete's characteristics, it is not possible to base their selection only in this aspect, since there are populations that have better characteristics that lead to a greater success and it can lead to an unfair process. A recent review [23], in which they compared many traits found in different races (e.g. the Anglo-Saxon race that stands out in swimming) proves that genetics is an important factor that contributes to determine performance between individuals from different populations.

7. Conclusion

The presence or expression of genes that encode proteins necessary for contractile machinery or metabolic pathways usually show variants that provoke changes in the physiology of the athletes, which is responsible for the genetic traits previously described. The activation of these genes is partially influenced by environmental factors that help the athlete to experiment a higher development of some traits, like increase of muscle mass, necessary for some sport disciplines. The knowledge of the genetic differences is essential to understand the importance of individualised follow-up of athletes, which contributes to pushing them and taking advantage of their own characteristics; besides, identifying high-risk markers in the athlete's genome could help to prevent injuries or more serious consequences that may affect their health.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

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