



GENETICS WORKING FOR SPORT: FACT OR FICTION?



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The sports performance can be influenced by several factors: training programs, the diet, psychological factors and the use of ergogenic aids. But even when athletes adopt similar behaviors while preparing for the competition, only a few succeed in their sports, collecting medals and establishing a solid hegemony. But how to explain the fact that athletes who have the same training regimen, the same type of diet, and often live in the same clubs, achieve different sport competition outcomes? In this context, the scientific and technological advances are enabling the assessment of further analysis, such as the humans' genetic profile (genotyping)^{1,2}. This type of analysis may help detect individuals more responsive to sports training^{1,2}. Moreover, the genetic profile will help to explain the individual variability in response to specific stimulation related to sports training. By the year 2005, the genetic maps for physical performance and health-related phenotypes have included 165 candidate genes². Some of these genes are targets of extensive studies related to their influence on the athletic performance in long- or short-duration sports. For example, the gene that contains the information for the production of the alpha-actinin 3 protein (ACTN3) may present a mutation^{3,4}, resulting in a lack of this protein in the homozygous individuals. The ACTN3 is a sarcomeric protein, which interacts with muscu-

lar actin and constitutes the main structural component of the Z line. However, only the fast-twitch muscle fibers, and particularly, the fibers with lower oxidative capacity IIb (IIx), have information to produce ACTN3. Although the gene mutation does not impair muscle function, genotyping studies have shown that the mutation frequency is much lower in athletes that succeed in high-intensity activities⁴ (sprints or movements that require great muscle power). This may suggest that this protein plays an important role in skeletal muscle, involved in this kind of activity. Conversely, the mutation is very common in athletes who perform prolonged activities (endurance), indicating that the lack of ACTN3 can benefit these individuals, enhancing aerobic performance³.

Another gene related to endurance events performance is the one that encodes the transcription factor, known as PGC-1 α ⁵. Several evidences indicate that PGC-1 α exerts an important function in the acquisition/development of oxidative metabolic properties in skeletal muscle⁵. For instance, the PGC-1 α is found in greater amount in slow-twitch fibers (oxidative), reinforcing the hypothesis that this gene is involved in oxidative metabolism. Studies carried out on experimental models, in which the transgenic animals are bred to produce great quantities of PGC-1 α , show that in these mice the mitochondrial biogenesis is increased. Moreover, there is a great conversion of fast-twitch fibers to slow twitch fibers in those animals. Although it is not possible to reproduce these models in humans, it is known that endurance training can increase the concentration of PGC-1 α in skeletal muscle, favoring the adjustments mentioned above.

Several genes have shown some influence or relationship to sports performance, but when we talk about elite athletes, the more extensively studied is the gene that encodes the angiotensin-converting enzyme (ACE)^{6,7}. This gene can take different isoforms among individuals, resulting in the production of proteins with different characteristics. One of these forms, known as ACE I, is related to increased ability to perform endurance activities, while the other form (ACE D) is related to improved performance in events that require high power output. A study carried out on 64 Australian rowers showed a higher frequency of ACE I when com-

pared with the control group with 144 healthy subjects⁶. Conversely, another study showed a higher frequency of ACE D isoform in 35 elite short distances swimmers⁷. These studies suggest that the ACE gene different isoforms have different effects on athletic performance.

The scientific and technological advances have enabled the development of new strategies for the detection and preparation of elite athletes, which leads the elite sport to a level never previously achieved. The molecular biology tech-

niques (e.g. genotyping), which can detect, for example, genes mutations that have some kind of influence on performance, might be a helpful tool to select young athletes. Thus, genotype knowledge could help the detection of individuals with greater potential (trainability) to respond to certain activities, maximizing the training programs effects. Regarding the question rose in the title of this article, recent evidence shows that the use of molecular biology tools in sports is now a reality!

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