



REVIEW PAPER

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How genetic predispositions may have impact on injury and success in sport

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Abstract

Introduction. Studies investigating the determinants of physical endurance were initiated nearly 30 years ago. The research was inspired by the curiosity to find out about the nature of talent for sport and why some athletes are better than others, despite the same or even greater effort in training routine, diet and the supplementation. An attempt was therefore made to determine the genotype of a perfect athlete, but conducted research showed that it is a very difficult task. Although 140 genes were proposed to affect of ideal sportsman fitness, scientists are still far from formulating answers about the nature of physical abilities and genotype.

Aim. Our main goal was to review the literature about the selected genes and polymorphisms which are most often investigated in the context in relation to injury in sports.

Materials and methods. Analysis of literature from US National Library of Medicine, National Institutes of Health, PubMed, Google Scholar.

Results. We review the selected genes and polymorphisms which are most often investigated in the context in relation to injury in sports, we also present the function of genetic variants prevalent in athletes which are able to achieve better physiological adaptation during the training.

Conclusions. There are probably more than 140 genes involved in physical performance. Changes in even one nucleotide within the gene (SNP) can improve the body's adaptation to better physical performance and the frequency of injury to athletes.

Keywords. sport, genetic predisposition, endurance performance, aerobic capacity, injury

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The list of abbreviations:

LDL – low density lipoprotein, SNPs – single nucleotide polymorphisms, VO_{2max} – maximal oxygen uptake, GDP – guanosine diphosphate, ATP – adenosine triphosphate, ADP – adenosine diphosphate, RLC – regulatory light chain, CK – creatine kinase, Mb- myoglobin, mTOR – mammalian target of rapamycin kinase, BMD – bone mineral density

Introduction

Individuals who are able to master new movements during exercise within only few repetitions and who need little time to learn new motor skills are commonly referred to as physically gifted individuals.¹ A question arises what factors may influence the occurrence of such abilities in some people, while a complete lack of these abilities is observed in others. A similar question can be asked in relation to professional athletes, i.e. why do some athletes gain greater benefits from training than others? The explanation could be provided by Darwin's theory of natural selection declaring that people with more favourable traits have a better chance of survival and transfer the relevant trait to their offspring. It should also be noted that every person has certain limits in task execution. This is caused by the adaptation of the organism to the environment and the prevailing conditions. Phenotypes characterized by endurance, which may be linked with genetics, include: muscle performance, extensibility and strength of tendons and ligaments and physiological attitude towards training.² Increasing the performance of one function may impair the development of another. These assumptions were supported by research focusing on world-class athletes. In one of the experiments it was shown that persons cultivating a strictly defined sport on a daily basis may be predisposed by nature to succeed, yet the same individuals are not able to obtain equally good results in other disciplines.² It has also been shown that players who are very successful in static sports, such as weightlifting, cannot boast of equally good results in dynamic sports e.g. sprint running, and *vice versa*.³ The importance of the genetic factor has long been studied in many laboratories worldwide. One of these projects is "the HERITAGE family study" focused on health, risk factors, training exercise and genetics, which began in 1992. This project was designed to determine the impact of individual genes on the performance of the cardio-respiratory system, metabolism, and hormone balance during aerobic exercise. The analysis also covered such parameters as: maximal oxygen uptake, blood pressure, concentration of glucose and concentration of free fatty acids. The results of this project have shown that changes in metabolic processes during training have influence on the results achieved by athletes and in some patients this increase amounted even 50%, but no relationship

between initial level of VO_{2max} and the change in this parameter after training was found. Changes in metabolic process are a result of the regulation of gene expression and the variability of non-protein and protein products.⁴ Roth et al. suggests on an interest in a particular sport discipline is affected by genetic factors, and additionally, it was noted that the specific physical characteristics, such as body shape are important and unique for particular sport disciplines.⁵ Hereditary traits were also examined in children whose parents were professional athletes. During this study the hereditary sport-related traits like skeletal muscle was shown to range from 55% to 65% frequency, but height frequency of the body was found to be the most hereditary trait of all, reaching the up to 85%. Aerobic capacity was measured in terms of a submaximal rate, of the measured value and as the maximum factor (eg. VO_{2max}). This trait has proven to be much less heritable compared with the abovementioned genes, heredity oscillated in the range of 40-50% frequency. Each of the attributes described above can help to increase the endurance of the organism.⁵ However, looking at the last 20 years, and the studies carried out during that time, it can be seen that they mostly focused on isolated genes, as well as a small number of chromosomal regions.⁶

Polymorphisms of genes described below may be associated with predispositions to various types of sports. Pescatello and Roth presented the relationship between gene tested in terms of predisposition for sport as overlapping parts of circles, which indicates that one gene does not affect only one function in the human body.⁷ In addition, increasing number of scientific publications show a multigene character of performance *attainment* in *sport*, thereby departing from investigating isolated genes only.

Genes and gene variants implicated in determination of endurance

Adrenergic- β 2 receptor (ADRB2)

Adrenergic receptors are located in cell membranes, they are activated by adrenaline or noradrenaline, and are able to activate the G protein which is responsible (along with GPCRs) for activation and directing of B and T lymphocytes and monocytes towards immune response and for enhancing their proliferation and in addition phosphorylation of GDP. Beta receptors are involved also in cardiac and smooth muscle tone (in the muscle they are expressed in 99% of cells).^{4,8,9} ADRB2 is responsible for the performance and endurance of the body because it regulates energy and lipid levels in human adipose tissue.¹⁰ It is responsible for relaxation of the smooth muscle, strength and frequency of myocardial contractions, dilation of blood vessels and decrease of blood pressure.⁴ Research also showed the inverse correlation between initial concentration of noradren-

aline and work performed or maximal oxygen uptake, and positive correlation between the expression of the *ADRB2* gene before training and work performed as well as maximal oxygen uptake.¹¹ Beta adrenergic receptor is the main target of illegal anabolic agents which are used by athletes. Agonists of these receptors cause e.g. expansion of the bronchi. In combination with corticosteroids the receptors significantly enhance the anti-inflammatory activity. Additionally people with the *Gly16* (*rs1042713*) variant show a rapid decrease in $\beta 2$ receptor activity and loss of sensitivity to agonists. On the other hand people with the *27Glu* (*rs1042713*) allele will be resistant to these effects.¹² Wolfarth et al in their study mainly focused on the *Arg16Gly* polymorphism (*rs1042713*).¹⁵ The study group included 313 athletes: runners, cyclists, rowers, triathlons and others. The results were compared with the control group of 297 male subjects. The findings showed that the *Gly16* allele was not responsible for the athletes' high endurance but for their strength. Tsianos et al. showed the *16Arg* allele influenced the results (times) obtained by marathon runners, whereas in gymnasts the *ADRB2* gene may be associated with low body weight and adipose tissue.^{14,15}

Guanine nucleotide binding protein β polypeptide 3 (GNB3)

The *C825T* polymorphism (*rs5443*) in the *GNB3* gene, which encodes the $\beta 3$ subunit of the G protein is associated with increased transduction signal between the receptors coupled to this protein and intracellular effectors presented in almost cells of the human body.¹⁶ The product of the *GNB3* gene can also affect hypertension by enhanced exchange of Na^+/H^+ activity which is mediated by G- protein.¹⁷ The T allele of *GNB3* gene (*rs5443*) is associated with higher arterial blood pressure in Korean men.¹⁸ There is also evidence that the T allele of this gene may enhance effectiveness of certain drugs, e.g. used as treatment of pulmonary hypertension.¹⁹ This polymorphism has also been investigated in the context of sporting achievements.

A study of 155 Israeli athletes has demonstrated that there are no significant differences in the allele frequency in three groups (controls, endurance runners and sprinters), regardless of their proficiency level, for genotype distribution and for allele frequencies. However, Eynon et al. did not find the correlation of the *C825T* polymorphism of *GNB3* gene and the $-9/+9$ polymorphism of *BDKRB2* gene with the endurance performance.²⁰ Sawczuk et al. confirmed that the *C825T* polymorphism had no differences in genotype distribution between endurance oriented athletes and strength/power in Polish athletes.²¹ However, previous studies, conducted by Eynon et al. demonstrated that TT genotype was more frequent in elite endurance athletes than in sprinters and that the *GNB3* gene was associated with

$\text{VO}_{2\text{max}}$ in people who were not athletes.^{22,23} Additionally the results reported by Gülyaşar et al. suggest that the T allele can be used as a genetic marker of poor capacity for sporting achievement, because basketball players with this genotype obtained lower results related to muscle strength than the remaining study participants.²⁴

Genes and gene variants implicated in determination of muscle endurance

Creatine kinase (CK-MM)

Creatine kinase supplies ATP molecules to the heart and to the skeletal muscle.⁴ It is a catalyst in the reversible transfer reaction of a phosphate group from phosphocreatine to ADP, which is produced during muscle contraction; as a result of this process ATP molecules are formed.²⁵ CK-M is associated with myofibrils M-line which is located in sarcomere. Creatine kinase also regenerates ATP molecules in that location.²⁶ Rivera et al. suggest that the *CK-MM* gene may contribute to individual differences in the $\text{VO}_{2\text{max}}$ (which was measured during cycle ergometry test before and after 20 weeks of endurance training) response during training ($n=495$).²⁷ The A/G (*rs8111989*) polymorphism of the *CK-MM* gene affects athletes' endurance.²⁸ The AA genotype may be associated with rhabdomyolysis.²⁹ A allele was detected with higher frequency in athletes' strength.³⁰ Additionally, the GG and GA genotypes in women ($n=85$), who practice sports as well as aerobic and anaerobic exercise, are related with maximum oxygen uptake. The G allele in turn has a positive effect on $\text{VO}_{2\text{max}}$.³¹ Moreover, as a more common gene variant can be associated with the mechanism protecting muscles against damage.²⁹ Studies of Russian athletes have shown that the AA genotype is more common in endurance athletes, e.g. in rowers, which may be associated with higher $\text{VO}_{2\text{max}}$. On the other hand the GG genotype was more frequent in weightlifting athletes.³⁰ In contrast to the study presented above, Döring et al. did not find a link between the A/T (*rs344816*), C/G (*rs10410448*), C/G (*rs432979*), T/C (*rs1133190*), T/C (*rs7260359*), G/T (*7260463*) and C/T (*rs4884*) polymorphisms and elite athletes.³²

α -actinins-3 gene (*ACTN3*)

Alpha actinin belongs to a group of actin-binding proteins.³³ There are two genes that encode skeletal muscle alpha-actinin. In humans *ACTN2* which is expressed in all muscle fibres and *ACTN3* expressed only in Type 2 fibres. The genotypes associated with *ACTN3* are related to the size of the muscle mass and the use of glycogen during exercise.³⁴ Alpha actinins are responsible (as opposed to the CK-MM) for the Z-line in sarcomere, and play a role in the regulation of metabolism and signalling pathways.^{3,35} Different genotypes of the *ACTN3* gene are associated with different physical fitness in hu-

mans relative to specific environmental conditions. The strength linked with type 2 muscle fibres (high speed and rate of movement) is expressed as the ability of an individual to adapt to training; moreover, a protein, which is a product of the *ACTN3* gene, is necessary for the normal structure and muscle function.^{8,3} The *R577X* polymorphism (*rs1815739*) of the *ACTN3* gene is often described in the context of sporting achievements. Replacement of arginine at amino acids 577 causes a generation of a premature stop codon and result in complete lack of production of a protein in actinin 3.^{36,37}

Lack of this protein is quite common (18% people have the XX genotype) and may impact the increase in calcineurin signalling in alpha actin 3.^{38,35} The result is the release of the calcineurin from inhibitory effect of calstabin 2 and leads to a reduction in strength, muscle mass and fast-twitch muscle fibres.^{35,39} This increases the metabolism of the skeletal muscle and the quantity of slow-twitch fibres; additionally, exercise-induced increase of phosphorylation of mTOR (which is important signalling pathway in regulation of muscle mass) was lower in individuals with the XX genotype than in those with the RR or RX genotype.⁴⁰ This indicates a lower predisposition to hypertrophy in people with the XX genotype.³⁴ A study carried by Mills et al. on a group of Russian athletes which doing various types of sports, including volleyball, basketball, boating and others. The study found a correlation between the R allele and high level of serum testosterone in male group, in female. Moreover, the authors observed a deficiency in α actinin 3 and muscle hypertrophy as well as power athlete status of the holders of these allele.⁴⁰ On the other hand a study of Japanese athletes has demonstrated the influence of the *R577X* polymorphism on muscle strength, but only in male subjects. The authors of this study suggest that the RR and RX genotypes are associated with significantly higher affects peak muscle power in men during anaerobic exercise then in athletes with XX genotype.⁴¹ In another study on Japanese runners it has been shown that sprinters with the RR and RX genotypes achieve better timing than those with the XX genotype because the former genotypes give benefits which are linked with the endurance of the organism.³³ No such genetic relationship was identified in the case of runners who compete over a distance of 400m.⁴²

Myosin light-chain kinase (MLCK)

RLC catalyzed by MLCK plays a significant role in the development of muscle strength. The binding of Ca^{2+} to troponin and tropomyosin is a fundamental regulator of skeletal muscle contraction.⁴³ It is possible that MLCK has the ability to change the phosphorylation of the RLC type 2 fibres, reducing the ability to withstand loads during long-lasting muscle contractions. The *MLCK* gene can be helpful in providing answers to the varia-

bility of muscle injury during exercise. The studies presented below investigated two polymorphic sites *C49T* (*rs2700352*) and *C37885A* in this gene. The first of these is responsible for the increase in CK (creatin kinase) and Mb (myoglobin) after four-day exercise. It was observed that the largest increase and activity in the above proteins was demonstrated in carriers of the TT genotype. The second polymorphism is associated with higher concentrations of CK, however only after 7 days of effort. But heterozygotes exhibit greater concentration of this protein than homozygotes after 10 days of training; on the other hand the *C37885A* polymorphism was associated with decreased strength (which was determined by level of Mb) during training. Additionally, heterozygotes showed greater loss of strength after training, compared to the homozygous individuals.⁴⁴ People with the *37885A* allele are more susceptible to rhabdomyolysis than homozygotes with the C allele, however, it may also happen that individual carriers of the *49T* and *37885A* genotypes may in the future be affected by this muscle disease.^{44,45}

Angiotensin I converting enzyme (ACE)

Angiotensin I converting enzyme gene (*ACE-I*) was one of the frequent studied genes in regards to the physical performance because their physiological function.⁴⁶ *ACE* is located on chromosome 17, it is composed of 25 exons and 26 introns.⁴⁷ This gene impacts the renin-angiotensin system (RAS). It plays an important role in the regulation of blood pressure, sodium, water and an increase in muscle tissue.⁴⁸ The reduced amount and activity of ACE circulating in serum may be associated with an insertion polymorphism (inserting 287 bp of Alu sequences) in intron 16 of the *ACE* gene.⁴⁹ Angiotensin I converting enzyme is a genetic marker which is also used in determining the risk of kidney or cardiovascular disorders. With regard to this gene there may be the following genotypes: homozygous I/I or D/D and heterozygote I/D (*rs4646994*).⁵⁰ The insertion (the I allele) is associated with an improvement in the organism's performance in response to an applied endurance training. It has also been shown that the polymorphism is associated with prolonged exercise capacity at high altitudes. This was demonstrated during surveys of British mountaineers (who had ascended beyond 8,000m without oxygen) no one was homozygous with for D allele. Similar studies were carried out taking into account rowers, runners, and cyclists which have most frequently I allele compared with controls.^{51,48} The I/I genotype is characterized by low activity of this enzyme in the tissues and allows to keep a positive energy balance during long and intense exercises. It turned out that the athletes with an insertion allele obtained better results at distances longer than 200m. In contrast to the I allele, individuals with the D allele obtain better results in short-distance

sports, which require greater muscle strength and more rapid shrinkage of type II muscle fibres these findings were confirmed e.g. for swimmers and sprinters running at a distance of 200m.^{48,50,52} Researchers also found that the D allele is a factor contributing to the uniqueness of some players, because its significant part participates in the conversion of angiotensin I to II. The latter is prevalent in the skeletal muscle but it is also found in the myocardium; it has also been demonstrated that angiotensin II is involved in the repair of this tissue.⁵³ The I/D polymorphism may be associated with vascular diseases such as hypertension, myocardial infarction, and left ventricular hypertrophy. This polymorphism has no effect on the level of oxygen uptake or regulation of muscle contraction but it is associated with the increase in the size of type I muscle fibre. The D allele is associated with higher production of angiotensin II and aldosterone, as well as decreased half-life of bradykinin in comparison with the I allele.⁵

In contrast to the I/I genotype, the D/D genotype is more common in athletes who have to deal with anaerobic exercise (sprinters, short-distance swimmers). In these individuals the ACE levels in tissues is more than twice as large as in people with the I/I genotype.⁴⁹

In general, left ventricular hypertrophy is a common trait of athletes. Exercise activates the renin-angiotensin system, which can regulate the growth of the heart muscle. Angiotensin II stimulates the synthesis of proteins in this muscle, whereas bradykinin, which plays the opposite part, inhibits this process. It has been proven that the increase in the left ventricular mass in each of the subjects was different; this suggests that the genetic factors can regulate this trait. Moreover the D allele is associated with the ACE protein concentration and thus angiotensin II in plasma and tissues.⁵⁴ The highest concentration of this protein was demonstrated in individuals with the D/D genotype and the lowest in those with the I/I genotype.⁵⁵ Increasing the ACE level in the organism can promote angiotensin II; the consequences include an increase in the amount of superoxide anions decomposing nitric oxide (which regulates the cardiovascular system and more specifically is directly related to vascular relaxation; it retains a resting tension of blood vessels); additionally, ACE can promote the degradation of bradykinin.^{54,56}

Bradykinin β 2 receptor (BDKRB2)

BDKRB2 encodes the bradykinin β 2 receptor responsible for increased glucose uptake in the skeletal muscle during exercise. Activation of bradykinin leads to the production of nitric oxide (NO) from arginine. Under the physiological conditions NO regulates mitochondrial metabolism and optimizes the ratio of oxygen consumption to the produced energy.⁵⁷ *BDKRB2* is responsible for the regulation of arterial blood pres-

sure and for the performance of muscle contraction; it may be associated with bradykinin which is activated by *BDKRB2* (B2R).^{58,59} The polymorphism investigated in this context -9/+9 (*rs5810761*), located in exon 1. The -9 allele, in contrast to the +9 allele, is associated with increased expression of *B2R* gene; moreover, this allele is associated with higher muscle metabolism efficiency and enhanced performance and endurance in athletes.⁵⁵ Additionally, the -9/-9 genotype is more frequent in Caucasian triathletes than in the control group and also together with the G allele of the *NOS3* gene impacts better results at the finish compared with the control subjects.⁶⁰ Furthermore, the homozygous -9/-9 genotype showed associated with better muscle growth after short force-training in comparison with the homozygous +9/+9.⁵⁸ On the other hand a study of 125 Israeli athletes by Eynon et al. showed that the -9/+9 polymorphism was not related to endurance in athletes.²⁰ A study on Greek athletes showed a predominance of the +9/+9 genotype; moreover Sgourou et al. suggested that the joint influence of the D/D genotypes of the *ACE* gene, +9/-9 *BDKRB2* and G/A *LEP* gene may be associated with better feat achieved by athletes.⁶¹ On the other hand a study of Polish swimmers (157 subjects) suggested that the -9/+9 *BDKRB2* gene polymorphism had no effect on sport achievements.⁶²

Insulin-like growth factor-1 (IGF-1)

Insulin-like growth factor 1 (IGF-1) is redundant and structurally similar to the insulin receptors. Just like myostatin, IGF-1 is produced in the skeletal muscle and liver; it is also a modulator promoting muscle growth and strength. IGF-1 impacts the anabolic effects such as the increase in the level of protein synthesis.⁶³ An increase of IGF-1 in the bloodstream following physical effort may be an evidence of effective training and good health; moreover, free form of IGF-1 may be absorbed by the tissues which are involved in exercise.⁶⁴ The potential benefits possibly caused by IGF-1 include: an increase in lipolysis, lipid oxidation, glycogen synthesis, a decrease in muscle degrading proteins, an increase in the opposing, synthesizing proteins and an increase in the synthesis of collagen in tendons.⁶⁵ The polymorphism of this gene, potentially associated with the genetic determined sporting accomplishment is *C1245T* (*rs35767*) which is located in the *IGF-1* gene promoter. This polymorphism was investigated in the context of the muscle size and function. It has been shown that in elderly women the C/C genotype is associated with increased body fat compared with the C/T genotype; in addition people with the C/C genotype have lower level of muscle tissue and fat-free mass than those with the T/T genotype.⁶⁶ The T/T genotype is only found in athletes, but not in the control group. It may also be associated with athletes' strength and endurance. Moreover,

the T allele occurred only in top athletes.⁶⁷ The polymorphism associated with the P1 region in the *IGF-1* gene promoter is characteristic for endurance athletes, and it may also be associated with the organisms' adaptation to performance.^{68,66} Thesecond investigated polymorphism is *A275124C (rs1464430)*; the study published by Ben-Zaken et al. shows that the incidence of the A/A genotype is much higher in the control group than in whole swimming athletes group, this is due to reduce of frequency A/A genotype in short-distance swimmers.⁶⁷ On the other hand, there was no difference in the prevalence of this genotype in athletes competing in national and international tournaments, but in top athletes this genotype occurred less frequently than in athletes competing in national events only.

Myostatin (MSTN)

Myostatin is a highly conserved member of the TGF- β family (transforming growth factor- β), functioning as a negative regulator of muscle size and mass, moreover it is often expressed in skeletal muscle but lesser in adipose tissue and cardiac muscle.⁶⁹⁻⁷¹ Polymorphism of *Lys(K)153Arg(R)* in (*rs1805086*) myostatin gene consist in replacement nucleotide 2379>G, which causes the exchange of amino acids included in mature myostatin protein. This may affect the proteolytic processing or binding affinity to the extracellular activin type II receptor which causes activation of the SMAD pathway, inducing myoblast proliferation and muscle mass growth.⁷² Studies of untrained individuals suggest that this polymorphism is associated with influences muscle strength and an increase in the thickness of triceps and biceps. Another study involving a group of 316 Caucasian endurance athletes has shown that the *K153R* polymorphism influences an increase in the thickness of the biceps and quadriceps muscle, but the gene in question cannot be considered as a marker of organism's endurance.^{72,73} On the other hand a study of a young African-American population proved that the 153R allele influenced the maximum isometric contraction in both sexes.⁷⁴ On the other hand, Seibert et al. conducted research focusing on people aged 70-79 and the study yielded different results: people with the 153R allele had less muscle strength than the other participants of the study.⁷⁵

Susceptibility to injury

Mutations in the *COL1A1* gene coding the alpha 1 chain of type 1 collagen, which is the principal protein component of connective tissue, can cause many diseases such as e.g. *osteogenesis imperfecta*, excessive bone fracture or Ehlers-Danlos syndrome.⁷⁶ Single-nucleotide polymorphism in the *COL1A1* gene is associated with connective tissue disorders such as increased risk of shoulder dislocation, Achilles tendon rupture, verte-

bral fractures due to diseases causing low bone mineral density (BMD) and anterior cruciate ligament rupture. The latter injury is associated with the +1245 G/T polymorphism (*rs1800012*) of the *COL1A1* gene. A study conducted on Polish skiers showed that the probability of rupture in the anterior cruciate ligament is 1.43 times lower in individuals with the G allele compared with the subjects with the T allele. Moreover the authors found the difference in the genotype distribution (GG vs GT and TT) between injured skiers and controls.⁷⁷ Another polymorphism of this gene which is described in the literature with regard to the same injury is -1997 G/T (*rs1107946*). A study about football players has shown that the higher frequency of the GT haplotype in both of these polymorphisms (-1997G/T and +1245 G/T) acted protectively against a possibility of injury involving anterior cruciate ligament rupture.⁷⁸ On the other hand the *COL5A1* gene encodes the alpha chain of type V collagen.⁷⁹ Moreover, similar to *COL1A1*, it may be associated with Achilles tendon injuries and joint hypermobility (Ehlers-Danlos syndrome).⁷⁶

Polymorphism-/AGGG(*rs71746744*) is associated with adaptation to running, while AGGG/AGGG genotype was significantly over-represented in the group of fast and least flexible runners compared with the -/AGGG and -/- genotypes.⁸⁰ Genetic predisposition to and a higher risk of tendon injury is also associated with the genetic variation in the gene encoding the protein MMP3 (matrix metalloproteinase3) and TNC (tensacine C).The5A/6A polymorphism (*rs3025028*) in the promoter of the *MMP3* gene, like the *COL1A1* gene polymorphisms, may be linked with cracks in the anterior cruciate ligament. Moreover, the 5A genotype may be associated with susceptibility to this type of injury comparing patients from contact sports and non-contact.⁸¹ These genes are also responsible for Achilles tendon injuries in physically active people.⁸² Furthermore, the variant of the G allele of the *MMP3* gene and T allele of the *COL5A1* gene interact and increase the risk of Achilles tendon injuries.⁸³

Conclusion

The article is a review of research papers discussing selected locations of markers used to study genetic determine to sport achievements and to predispose to sustaining injuries.

Another important aspect described here is the psychological adaptation. While considering these issues, it is possible to raise a question regarding the genetic factors which might be responsible for contestants' will to compete, their desire to win, capacity for self-denial, persistence in aiming for a particular purpose, or for the phenomenon in sport frequently referred to as a one-day-predisposition. Despite the positive findings of the many studies, the authors suggest the multi-ge-

netic nature of physical capacity and the dependence of the tested alleles responsible for the organism's strength on this function. The same conclusion which was presented above can also be drawn in the context of muscle performance and susceptibility to injury. Therefore, other genetic markers associated with strength, injury proneness and psychological adaptation tested jointly, can provide new and interesting information about the predisposition to succeed in sport. However, these genes should be investigated in relation to specific sports rather than taking into account similar disciplines jointly. This is because each sport has its own unique characteristics; therefore the training in an obvious way focuses on different domains, which as a consequence are the underlying factors of the competitors' success. Considering the genetic markers of sport achievements we also have to take into account the differences between populations.

New available technologies such as microarrays and new generation sequencing (NGS) could provide new insights on the genetic determinants of sports achievements and predisposition to injury. The role of association studies concerned on human disease is huge and well established. The significance of genetic study focused on physical performance may be useful to the decision by subjects, who do not reached satisfactory results or sustained injuries during training/competition, on which discipline they should concentrate to reach the better results and which efforts they should avoid. However there are probably more than 140 genes involved in physical performance what nowadays makes it difficult to use them in laboratory tests. In addition, besides the SNPs, epigenetic changes like DNA methylation and microRNA expression can modified the ability to athletic effort and sport achievements. There is much probability that in the future the epigenetic and functional study will develop in this science field.

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