

Kinesiogenomics: The Genomic Era in Kinesiology

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Abstract

The application of genomics research in kinesiology may be characterized as kinesiogenomics or sport genetics. Therefore, kinesiogenomics is generally known as the study of genetics in the various discipline of kinesiology which refer to the study of human movement. In the research field of Kinesiogenomics, the scientific goal is to clarify the role of genes in sport performance, identification of various genes with their different alleles that affect to the response and adaptation of the body systems and metabolism .

By help of the these genetic research studies in various disciplines of kinesiology, it will be possible to use genetic testing to predict sport performance or to individualize exercise prescription with the potential for genetic therapy. It is supposed to enhance the sport performance because of that reason that the Interindividual variation of kinesiogenomic-related trait such as maximal oxygen consumption, muscle fibre composition and trainability has a strong genetic basis.

Genetic research has a large potential for developing the area of kinesiology with the contribution of genetic factors for various phenotypes related to sport performance including aerobic and anaerobic performance, muscular endurance and strength, motor performance, and some determinant of performance such as morphological, cardiac and skeletal muscle characteristic. In the area of kinesiology, up till now kinesiogenomics research studies have moved into a new era utilizing well-phenotyped, large cohort and genomewide technologies. The main aim of this review is to summarize the most recent and significant genetic finding in the various discipline of kinesiology and to predict the future expectation and possibilities for kinesiogenomics.

Keywords: Kinesiology, kinesiogenomic, exercise, sports genetics, sport performance, genomics

Kinesiogenomik: Kinesiolojinin Genomik Çağı

Özet

Genetik bilim dalının kinesioloji alanındaki uygulamaları kinesiogenomik veya spor genetiği olarak adlandırılabilir. Bu nedenle, kinesiogenomik terimi, kinesiolojinin çeşitli disiplinlerindeki genetik bilim uygulamaları olarak tanımlayabiliriz. Kinesiogenomik alanında yürütülen araştırmaların temel bilimsel amacı sportif performansın genetik yönlerini açıklayabilmek ve farklı genlerin farklı allelleriyle vücut sistemleri ve metabolizmasının yanıt ve uyumu üzerinde gösterdikleri etkileri açıklayabilmektir.

Kinesiolojinin çeşitli disiplinlerindeki genetik bilim uygulamalarıyla, aynı zamanda performansının saptanmasında genetik testlerin kullanılmasını olası kılacak ve sporcu performansını geliştirmeye yönelik bireysel egzersiz reçetesi ve gen tedavisini beraberinde getirecektir. Maksimal oksijen kullanımı, kısa fibril tip yapısı, antrene edilebilirlik gibi bireysel özelliklerde genetik yatkınlık ağır basmaktadır.

Genetik araştırmalar, sportif performansla ilişkili genlerin fenotipik katkıların açıklanmasıyla; aerobik, anaerobik kapasite, kalp ve iskelet kası morfolojileri, kas gücü ve dayanıklılığı ve sinirsel ileti özellikleri gibi kinesiogenomik bağlantılı özelliklerinin etkisiyle kinesioloji alanında yeni gelişmelere neden olmaktadır. Bugüne kadar yapılan kinesiogenomik araştırmalarında, kinesioloji alanında geniş kohortlu, fenotipi iyi belirlenmiş geniş katılımlı gruplar üzerinde yapılan genom çalışmalarıyla bir yeni çağ açılmıştır. Bu derleme makale çalışmasının amacı, kinesioloji alanındaki çeşitli disiplinlerde son dönemde yaşanan önemli genetik uygulamaları özetlemek ve kinesiogenomik açısından spor bilimi ve kinesioloji alanında gelecek için bilimsel beklenti ve olanakları öngörmeye çalışmaktır.

Anahtar Kelimeler: Kinesiyoloji kinesiogenomik, egzersiz, spor genetiği, spor performansı, genomik

Introduction

The most important studies in modern age have been going on in genetic field. In 2003 when researchers completed genome map, this was thought to be a huge success for genetics. Although thanks to this there have been many developments about usages of genes, there are also many unknowns and still researched situations about gene (Mara Diegue A, 2008; Montgomery HE, Marshall R, Hemingway H, et al, 1998; Gayagay G, Yu B, Hambly B, et al, 1998).

Forms, functions of gene and its genetic features inheritance are investigated by genetics and genomics. With the increase of gene studies, there have been also many questions and answers about gene applications on exercise and kinesiology. Among these questions, there are some important ones: Is there genetic applications in kinesiology? For what and how is genetics used? How should be genetic factor used in picking talents and talented athletes up? When genetic applications contribute to athletes' conditions, is there a doping effect? Is a genetic development being used in sportive activities is right for moral and ethics?

Usually 3 methods are applied for genetic studies. The first one is researching certain physical features' inheritances, the second one is mapping genes of big groups having coherent features and the third one is researching specifically nominated genes which are thought to effect physical features (Brutsaert, T. D; Parra E. J, 2006).

Genes are important for functional and structural characters that enables performance. Genetic factors are important because they effect the form of muscle-skeleton system, distribution of muscle types, metabolic activity, reflex capacity, lung capacity and using energy efficiently. Athlete can show an outstanding performance thanks to his genetic foundation. Supports of some genes like ACE, ACTN3, VDRL, EPO, PPAR-gama, PPAR-y, PEPCK, IGF-1, BMP, eNOS have been researched for success in sports (Gayagay G, Yu B, Hambly B, et al, 1998; Myerson S, Hemingway H, Budget R, et al, 1999).

It's still a question of debate that whether exercise capability is innate or acquired. It's also another question that whether performances with these talents have limits or not. Although sportive performance is thought to be understood like completing necessary tasks and activities to be successful, despite positive and negative facts, performance is a product of athlete's capacity, quality and sportive talent. While in some sportive activities like athletics, performance is running, jumping and throwing, in some performance have different features. So it's thought here genes affect sportive success (Brown L.E, 2000).

Because studies reveal the importance of gene, usage of genetic technics will be also important in the future. However, community should accept it for its usage. For example, embryo manipulation and some other genetic technics are important for blocking serious diseases but if community is biased against usage of genetic technics, this method will not be appropriate for ethics. But it's supposed that majority of community would support genetic technics that are used for treatments of important health problems (Breivik G, 2005; Miah A, 2005). This approval will lead to that genetic technics are used also in sportive activities. Gene map studies in sportive activities are used for determining the location of genes establishing performance features. The basic of these studies is determining huge community's own phenotypic features, stating genetic determiners and making statistic of these studies. When gen maps are made, more than one gene's phenotypic features on performance are also stated (Işık, A, 2008).

On the other hand, when we review the relationship of kinesiology and genetics, we can have idea about common health field. Thanks to genes reacting positively to trainings, people who don't sport, have healthier metabolisms than others despite the fact that they don't exercise. Besides, genes supporting marathoners about storing energy effect negatively health of people who don't exercise (Foody B. and Savulescu J, 2007)

Gene studies also make contribution to obesity. As a result of studies, interaction between genetics and nutrition and body weight is also important. In human genome, there are 2 copies of each gene one coming from mother and the other one from father. Gene sets among individuals have differences which are called Single Nucleotide Polymorphism. Among these differences there are such genetic changes that they can effect reactions of body against life style and nutrition (Gardner, 2007; Rankinen T, Zuberi A, Chagnon Y.C, Weisnagel S.J, Argyropoulos G, Walts B, Pérusse L, Bouchard C; 2006).

Before the project started, the gene number was known as 100.000. As a result of the study, gene number is few so gene numbers is not important, but its features are (Ünal M, Ünal D.Ö, 2003).

Genetics field is very important for every field especially health. In this article, we'll discuss gene's effect on sports, gene doping and gene-sports relationship with an ethical way. Because, human genes play a role in forming structural and functional character in sports performance. When reached to aimed point in gene studies, it's thought that they will find out athletes would be successful in which sports branch and develop their performances according to that and whether it's ethic or not. It's also a question of debate that the truth of using gene studies and gene therapy on doping. This situation affects genetic studies and so especially sports doctors, managers, everybody have a task about it (Montgomery HE, Marshall R, Hemingway H, et al, 1998; Gayagay G, Yu B, Hambly B, et al, 1998; Ünal M, Ünal D.Ö, 2003).

When Human Genome Project existed, they found out that the number of genes was not important but their features were (Mara Diegue A, 2008; UNESCO, 2009). Started in 1980, project has some aims like defining DNA array, the importance of DNA for human health and DNA differences between individuals. But the most fundamental aim was mapping human DNA. By this way, the password in human genome would be solved and when that would happen, many applications from human innate talents to their tendencies to diseases, gene treatment before birth, when that child grew up in which branches he/she could be successful were thought to be made (Mara Diegue A, 2008).

Genetic and Scienfical Ethics Discussions

The possibility of forcing children to change sports from hobby to carrier and making gene tests on children have been question of debate recently. And this may lead a child to take a dislike to sports (Savulescu J, Foddy B, 2005).

It's also an ethical problem to use gene transfer aiming doping. Being untalented not so important for sports, it's also a truth that there might be ethical problems in sport competitions considering serious investments (Tural Ş, Tural, E, Kara N. & Ağaoğlu S.A, 2011).

Privacy of athletes, gene tests on athletes and discriminations are also very important because they are big parts of ethics of this subject. So there have been legal adjustments about

protecting genetic rights in United Nations Gene Map and Universal Declaration of Human Rights (UNESCO, 2009; Lea D.H, 2008).

Genetics' Relationship with Exercise and Kinesiological Parameters

Genes have important roles in lives of people. For example, it's thought that athletes' being successful or some people's being unsuccessful in sports is related to genes. Sir Roger Bennister who runs a mile under 4 minutes said "athletes are born unequally". This quote is question of debate but people's ethnic origin and genetic structures may be effective on success. It's known that West African athletes are good at short distance, East Africans are good at marathons, Asians are good at swimming. Today the number of gene studies increase and studies will find out that gene have a role in determining tendency to certain sports branches of individuals (Varlet-Marie E, Audran M. and Ashenden M, 2009).

Genetic screening in early ages can lead to canalize in sports that child may be successful and be potential to make personal training program and thanks to them genetic tendency will be increased and special training methods will be picked up to develop (Tural Ş, Tural, E, Kara N. & Ağaoğlu S.A, 2011).

In the studies of sports doctor field, some factors like individual's gender, age, genetic structure, anatomic features, physiological balance, neural system, cardiovascular structure are standing out. Especially genetics is used as a tool by sport doctors to determine sportive talent. Genetic background determines performance potential however contemporary performance capacity exist by training much more, obtaining practices, being motivated for his sport branch, gaining regular nutrition habit and effected by other factors (Maughan R.J, 2005).

Information about Some Genes

Forming genotype, genes reveal individual's phenotype by enzyme and protein synthesis. The event which is important for sports doctor is that genes that effect exercise capacity , kinesiological parameters and sports phenotypes as a result of studies are found. Genes repairing injuries and effecting sportive performance are ACE, ACTN3, BMP, EPO, PPAR-gama, PPAR-y, PEPCK, IGF-1, VDRL, eNOS and others (Myerson S, Hemingway H, Budget R, et al, 1999). According to branches, athletes should have different biological features. Among genes that mostly reviewed, Angiotensin converting enzyme (ACE) exists. Starting with ACE, many genes were observed (Goa K.L, Wagstaff A.J, 1996).

Some genes like Erythropoetin (EPO), perocsizom proliferator activating receptor (PPAR-gama, PPAR-y), phosphoenolpyruvate carboxicinase (PEPCK), insulin like growth factor 1 (IGF-1), myostatin, follistatin bone morphogenic protein (BMP), vascular endothelium growth factor (VEGF), endothelium nitric oxide synthase (eNOS), aktinin binding protein 3 (ACTN3), endorphin have features effecting kinesiological performance in sports (Ruiz Jonatan R, et al, 2010).

The Relationship between Genes and Performance (Performance Gene)

Copenhagen Muscle Survey Institute proved that environment factor effected %25 of athletes' successes, while %75 of it depended on genes. When the number of studies about genes increase and unknown features are discovered, its effect on sports and kinesiological performance also found out. Each gene has a huge effect on performance. Phenotype features determine the performance under the effect of more than one gene. And gene map studies are done to determine performance features (Brutsaert, T. D, Parra E. J, 2006; Perusse L, Rankinen T, Rauramaa R, Rivera S. M, Bouchard C, Wolfarth B, 2003).

In a study, it's reported that maximum oxygen consumption value's genetic transmission rate is %50, explosive muscle strength genetic transmission rate is %67, and muscle fibre distribution genetic transmission rate % 40-50, cardiac output genetic transmission rate is % 42-46 (Calvo M, Rodas G, Vallejo M, Estruch A, Arcas A, Javierre C, Viscor G, Ventura J.L, 2002).

Cardiovascular performance is an important determiner of athletical success. However physical performance depends on muscle strength and muscle performance change so genetic studies in sports are mostly about cardiovascular field. Training increases cardiopulmonary capacity. The limits of increase depend on genetic background. It's thought that mitochondria number and muscle cells' using oxygen efficiently and forming ATP capacity depend on genetic features (Wolfarth B, Bray M.S, Hagberg J.M, Perusse L, Rauramaa R, Rivera M.A, Roth S.M, Rankinen T, Bouchard C, 2004).

In reported study short distance runners have Type 2 muscle fibers, while marathon runners have Type 1 muscle fibres (Savulescu J, Foddy B, 2005).

Reports about results of performance gene studies are related to common human health. For example while some genes that effect positively athletes, enable to form a healthier metabolism by exercises, some genes that protect energy for a long time for marathoners, may lead to some problems like obesity, heart, diabetes and etc (Perusse L, Rankinen T, Rauramaa R, Rivera S. M, Bouchard C, Wolfarth B, 2003).

Genetic background has a huge effect on power in exercise, resistance, type of muscle fibres, muscle massive rates, kinesiological parameters and lung capacity. Apart from cardiovascular area, some genes about other systems are also observed and researched their effects on exercise. As result of studies, athlete body's reactions to training, nutrition and other factors are found out. According to this situation, athlete who has a low potential in resistance will be more successful if he reacts to training better when we compare with an athlete who has a high genetic potential (Perusse L, Rankinen T, Rauramaa R, Rivera S. M, Bouchard C, Wolfarth B, 2003; Wolfarth B, Bray M.S, Hagberg J.M, Perusse L, Rauramaa R, Rivera M.A, Roth S.M, Rankinen T, Bouchard C, 2004).

Gene Therapy and Gene Doping

Transferring genetic material to human cells which are used for blocking diseases and failures is called gene treatment. Gene treatment is not only used for health aims but also for treatment and being normal in the situations of muscle and meniscus injuries and bond and tendon ruptures. The principle of gene treatment depends on compensating anormale or lack gene and sending treating gene to cell. (Haisma and Others) is still researched so there are many risks

that may exist in healthy people. Among them, there are heart attack, stroke, embolism (Halit E, Hayrettin G, Alpaslan K, 2013).

There is not yet success in gene treatment. This method having a low success possibility has high risks. Therefore there are restrictions in some countries like USA, UK, Germany, France, Italy, Switzerland, Japan, China and Australia. Thanks to Genome Project, genetic codes are resolved and there's an opportunity to be treated by gene therapy. While there are many ethical debates about gene therapy, the main aim of this therapy is compensating an abnormal or lack gene and sending treating gene to cell. With gene therapy athletes will have better performances and their bodies will be reformed (Ünal M, Ünal D.Ö, 2003).

There's a huge debate about that gene therapy would be used as doping. Gene and cells being used in a untreated way and increasing athlete's kinesiological performance is called gene doping. Proliferation of doping will rule fair competition out and harm athlete's health and sports soul. Gene doping is not only the newest one but also its harms are not known yet (Tural Ş, Tural, E, Kara N. & Ağaoğlu S.A, 2011).

Among factors that force athletes to use doping are success wish, being forced by trainers and managers, praising audiences for success and etc. When we think about elite sports sources distributions, it's already unfair structure. Because of gene doping, this situation will be more outstanding and it will be like Formula 1 races (Tännsjö T, 2005).

When gene doping's effects are researched, athlete who makes gene doping will force others to do the same thing. Even if an athlete doesn't use doping, the doping user athlete's fluid will pass the other one by physical contact and it will harm the other athlete (Schneider A.J, Friedmann T, 2006; Haisma H.J, de Hon O, Sollie P, Vorstenbosch J. 2004).

And religionist athletes will never use gene doping because it's against their religion and they will lose the competitions at the beginning. This will destroy excitement and competition feeling in sports. Genetic interference will lead to commit a sin by religionist athletes, leave the competition and other situations (Tännsjö T, 2005).

Gene doping is that target gene is transferred into cell. Presenting target gene to athlete is made by a method intracellular (in vivo) or extracellular (in vitro). Presenting intracellular gene doping to athlete is made by biological, physical or chemical ways. In extracellular method, cells taken from athlete are reproduced in laboratory and genes which are wanted to transfer are transferred into cells and given to the individual (Tural Ş, Tural, E, Kara N. & Ağaoğlu S.A, 2011).

IGF-1 gene transfer is used for doping. Human growth hormone increases IGF-1 level. Because myostatin blocks muscle development, it's thought that anti-myostatin injections are used for gene doping. Gene doping methods and HTC increase with EPO way, increase of blood stream with NEGF way, increase of muscle strength with IGF-1, increase of muscle sizes with a way inhibiting myostatin, endorphines are used for killing pain. It's too hard to understand gene doping and special gene which is transferred into body, have some results visible or measurable (Tural Ş, Tural, E, Kara N. & Ağaoğlu S.A, 2011; Livanelioğlu K, 2008).

Discussion

There are many against and supporting views about genetics. Especially these ethical views are much more in rightwinger environments. The most important reason of this is thoughts of religionists that genetic engineers interfere in God's works (Fukuyama F, 2003).

It has been always a debate that whether athletic talent is innate or not. With studies about genes on sports, it's added in the debate that talented children are picked up in early ages and personal training programmes are made for them to be successful. Because by this way equality will not exist.

It's understood from studies that sexual activation effects negatively sportive performance. Because sleep disorders, alcohol and behaviours against morality and rules results in this. Thought of "sexual activation harms" is coming from that despite sexual alert activation doesn't happen and it effects negatively on performance. The important thing is that under right circumstances activation's happening and this situation effects positively performance (Bayraktar B, Kurtoğlu M, 2004).

Doping in sports, especially gene doping is the most debated one. Being in WADA's exclusion list in 2003, there are still many different views about gene doping. Although the certain effects of gene doping are still unknown, they are against it because they state it it's used for increasing physical performance (Schneider A.J, Rupert J, L, 2009). In sports, although methods aiming treatment are not considered as problem, when it's beyond normal, in other words increasing performance it's restricted. However the limit is not determined so it's a question of debate (Breivik G, 2005).

If we give an example about it, genetic treatments and gene therapies aiming repairing muscles and regrowing may make muscle stronger than past. Or repairing anterior cruciate ligament effects directly athlete's performance. Athlete's knee may be stronger. Although it's acceptable, if muscles are used for strenghtening, it's not acceptable. Because in this point, gene treatment changes into gene doping (Breivik G, 2005; Schneider A.J, Rupert J.L, 2009). We mentioned about gene doping's effects being unknown. By this way, its harms are also not certain. However according to some views, individual is free to decide by his own. Explaining risks right, if it doesn't harm anyone, there must be no view against gene doping of athlete. Some scientists think that gene doping must be restricted and healthy athletes mustn't be damaged by gene doping (Breivik G, 2005; Schneider A.J, Rupert J.L, 2009; Simon R.L, 1984; Friedmann T, 2010).

Tamburini is also against restricting gene doping and states that the number of climbers who died are much more than the number of doping users and boxers also have many health problems after even they are retired (Tamburrini C.M, 2005).

Conclusion

Thanks to genetic studies and tests, nutrition and exercise methods are made for personal and this leads to a positive effect on athletes increasing performance and on obesities controlling weigh. When studies have results, athletes will be motivated for their talented branches in early ages and be very successful. And active individuals apply their personal performance increasing exercise programmes thanks to gene tests. Genetic background determines the potential which is needed for surpassing in sports. Even though an athlete have a genetic

background to break a record and be successful, he can't be successful with insufficient exercise, irregular nutrition and irregular life. But when an athlete having a limited genetic potential, he will be successful if he have a regular exercise and life. To understand changes of performance, interactions from mother's womb to professional sports life should be observed, in other words each genetic and enviromental factors should be observed and researched coorelation. Recently performance genetic has concentrated on candidate genes. Here it's so important to choose the best candidate gene, analyse true functions of genes with detailed informations in gene map. Therefore after choosing candidate gene, changes in this gene should be researched in huge communities and analysed for right informations. In 2002, WADA prohibited some methods which are told to be performance increasing works about gene by considering them doping. Considered as doping in WADA list, gene doping should be prohibited by all countries to competete fairly. If genetic engineering effects sports, this will lead to unethical behaviours. Resolving these behaviours will not be possible with moral values. A project including the whole world countries is needed to solve the problem. Those who are against prohibiting gene doping claim that elite sports don't have already a fair environment now. Is it really true that the ones who use gene doping are more successful than who don't and making elite sports unfair considering social and economical factors? On the other hand, sports doctors making studies on children in early ages and determining personal trainings and their talented branches may have very positive results for countries who care about tthis and may also results in inequality and destroying competitions in sports. Therefore the one who attach importance to sports, should not only keep up with genetics but also be careful about ethical ways of its effect on sportive applications and its reflection on daily applications.

Conflicts of interest

None declared by authors.

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