GENETIC AND SELECTION

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ABSTRACT

Leading countries in sport and sport medicine have for years been conducting research and experimentations on genetics to improve sport performance. Most of these studies have been kept in secret for years and are being used as an instrument for having advantage during hug competitions that moves from the political scene to the sport one. The curtain of these genetic studies in the area of high performance sports is gradually rising and the information is becoming more and more accessible to the sports community.

Researchers' efforts are focused on two directions: on the one hand, work on the genetic potential and prerequisites for choosing sport discipline, based on the main motor qualities such as strength, speed, flexibility and endurance. On the other hand, it is mainly aimed at identifying the individual's genotype related to his hereditary burden, metabolic and traumatism profile. Parameters from these studies support specialists in preparing training process, the right diet, the medication needed for rapid recovery and, last but not least, the prevention of health conditions and traumatism, especially in top level sports.

The main goal of the sports specialists in the field of sports genetic is to shape the individual from the beginning of his training and competitive career. Defining the core qualities at an early stage enables coaches to make the correct selection and direct athletes to a specific discipline in a given sport.

Today, even the scientifically defined genotype of an individual cannot be categorical and the only factor in making decisions about the scientific process of selection of young athletes.

Key words: genetic, selection, genotype, profile

How to find the perfect athlete and can we create such?

Are the genes enough to drive the athlete to the outstanding results?

INTRODUCTION – IN SEARCH OF SPORT GENES

Who we are, how the human body is created and is our nature suitable for tremendous physical activities? Can any of us be a good athlete and is our destiny predetermined? What are the capabilities of our ancestors? Is it coded in our genes to be an athlete, the family and the way of living determines the path to become such or just not yet undiscovered social-economical phenomena?

In the center of each cell of our body lies the curved DNA ladder, which defines us as human beings. Particularly, more than 23 000 genes are rooted in the ladder which determines the human genomes and us as Homo-sapiens. After years of profound research and analyzes, today we can claim that the human genome is almost decoded.

In general, the genetic science gives answers and analyzes in details the maxim "the quantitative ac-

cumulation leads to qualitative changes..." The climate, the social-economic changes, the technical revolution have a significant impact also on the human genome, particularly for the last half a million years of the human evolution. The craftsman has become engineers; the hunter became an athlete. However, the main question still stands: which genes a better and much stronger – of the craftsman on of the hunter? Of the hero of the day or of the one who's potential hasn't been recognized?

How science finds out that talent exist and further helps to develop it? For many years the scientists have been trying to find the exact sport gene researching only individual markers. However, the focus of research moves to a group or a combination of genes which affect the athleticism. Furthermore, researchers and scientists have started interpreting how the endowment together with the strenuous training and hard work will impact the athleticism.

Sprinter or long distance runner, long of high jumper? The leading sport countries for many years already gather and rely on the genetic code to prove the sport achievement. Looking for answers many researches are chasing the truth in order to find out which are the best genes that build the best athlete. Big part of those analyzes are kept in deep secret are further used as an advantage at the big stage of both sports and politics. The iron curtain of the genetic researches was partially lifted and became more and more accessible for the sport society.

Genetic endowment or tremendous efforts

Do you remember the classmates in the primary school which were always ahead, always proactive and which always difficult to be controlled? Do you remember the obese blond girl, which was the best in the math class? Do you remember the young teenager running faster than all the seniors? „She isn't good enough, don't invest in her!“ or „He was unique athlete, but the luck wasn't on his side! “.

We have all listened to the in-depth analysis of a number of specialists, but one of the most annoying things I've ever encountered is: „You are not born to be a sprinter, you have lost the last 10 years for nothing!“.

How does it feel when the athlete hears similar words from a specialist who has analyzed the results of his or her genetic test? How does feel a person who has dedicated the last ten years of his or her life for hard work, discipline, deprivation and injuries? How will a competitor feels, already at the sunset of his career, reaching the finals of European and World Championships, but never touch the Olympic glory, never stand on the podium, side by side with the best athletes?

The main goal of gene sports specialists is to profile the individual right from the start of his or her professional career. Defining biological gifts at an early stage enables professionals to make the right selection and to direct competitors to a particular discipline in a sport. The wide profile of genetic studies allows us to establish at an early stage the development of the individual and the level of his predisposition in general to motor activity by examining the genes as DRD2 and ACE.

The availability of such information largely provides clear guidance on where to direct a competitor

- to a discipline that prioritizes endurance or explosive power, speed, or high aerobic capacity. This avoids the loss of years of effort and wrong-headed preparation. Such genetic studies make it possible to analyze not only the muscular profile for the purpose of establishing the dominant muscular fibers of the musculoskeletal system, but also the possibilities of the cardiovascular system, aerobic and anaerobic capacity in order to establish the thresholds for permissible loads, fatigue and recovery rate.

GENOTYPE – BUILD A BETTER TRAINING PROCESS

Nowadays, more than 200 genes inform us about the physical activity of the athletes, their aerobic capacity, endurance, explosive power, speed, heart's work, soft tissues and the supporting motor apparatus. Here we are going to go through those genes, which are useful for those who aim to increase the sport results. Let's not talk about magical trainings and secret recovery recipes. Let's talk about the pure functional, individual peculiarities of the human organism and it's genetical predisposition.

The efforts of the scientists, working in the area of the sport's genetic concentrate in three main directions: *establishing the genotype of the individual, it's metabolic profile and the traumatic sustainability of the organism.*

The first part of the gene's test aims to establish the qualities of the cardio-vascular system, the musculature and the breathing abilities, which have vital importance for the development of every athlete's abilities.

The second part helps the specialists to create the right food regime and the supplement recovery process after heavy physical training.

Last but not least, the third part informs about the traumatic sustainability of the organism and the prevention from risky for the health situations, which are frequent in the professional sport.

The early establishment of the genotype of every athlete can be excellent instrument of early profiling, sports and disciplines choice.

CARDIO-VASCULAR PROFILE

The cardio-vascular profile shows the specifics of the possibilities and abilities of the heart to satisfy and supply with blood the organism during differ-

ent activities, different thresholds of recovery rates of the pulse frequencies and levels of the blood pressure. The establishment of the hereditary burden and the increased chance of heart attack and other heart diseases are highly important.

In order to define the working capacity of the individual and his stamina, molecular genetic analysis for polymorphism insertion/deletion (I/D) in the ACE gene has to be made. The polymorphism ACE has an impact on the Bradykinin and is in association with the aerobic endurance. This indicator makes direct connection with the possibilities for the vertical jump of the athlete, typical for sports like basketball, volleyball, vertical jumping in the athletics, ski jumping and etc.

The ACE gene also provides instructions for making a protein called angiotensin-converting enzyme, which converts a hormone called angiotensin I to another form called angiotensin II. Angiotensin II helps control blood pressure and may also influence skeletal muscle function, although this role is not completely understood. Allele I has an increased endurance, while Allele D has increased speed. Therefore, based on this analyze we can define the following genotypes:

- Genotype I/I – endurance type
- Genotype D/D – speed – power type
- Genotype I/D – universal type

The fast oxygen transmission to the muscles and the high level performance of aerobic capacity of the athletes has always been an object of attention not only of the trainers, but also for the sport medicine, which is connected with the recovery process of the teams. During the last few years the sports genetics pays special attention to a study called HERITAGE (Health, Risk Factors, Exercise Training and Genetics). In this case 21 different genetic variations, important for the development of maximum aerobic volume VO₂ max are tested. When we talk about training and results we have to keep in mind a lot of factors, however this test shows that the athletes who have 19 out of these 21 genes achieve three times better results than the ones that have 10 out of 21 genes. The main gene that gives information regarding the condition of the circulatory system and the way the blood cells transport the oxygen to the muscles is the EPASL gene. The main indicator connected with increasing of the aerobic volume and the development of the athlete is ATP1A2.

After genetic testing in recent years, many examples show that already proven athletes in sprint disciplines are arranged to be better athletes for mid or long distances running. And vice versa: after genetic studies of young athletes who show exceptional aerobic ability at long-range performance, they definitely have dominant muscle fibers and cardio-vascular profiles of sprinters. This has definitely gone unnoticed by the coaches and is a serious fact that should be addressed in the future preparation and profiling of competitors with such indicators.

STRONG ENOUGH OR FAST ENOUGH – NO, JUST BE SMART

The speed of the muscle fibers and the skeletal muscle is defined by the molecular genetic analysis for polymorphism RS77X in the genes ACTN3 and ADRA2B. The gene ACTN3 or so called Speed Gen codes the sarcomere protein α -aktinin-3 which expresses in the fast, glycolic, type II fast twitch muscle fibers and plays an important role in generating of the “Explosions” speed-power muscle contractions. There are three main genotypes for the type of muscle fibers and skeletal muscle:

- Genotype – C/C (RR) – higher percentage of fast twitch muscle fibers (Type II) – typical for the speed-power type athletes
- Genotype T/T (XX) – higher percentage slow twitch muscle fibers (Type I) – typical for the endurance type athletes
- Genotype C/T (RX) – equally divided between the two main types of muscle fibers (universal type)

During the last few years the research targets specific muscle groups for establishing the potential of the individual. An example can be the complex of genes that give information about the qualities of the lower limbs like BDKRB2, which defines the power of the quadriceps femur and the efficiency of the muscle contraction. Together with the other already popular genes for muscle fiber and supporting motor apparatus ACTN3 and ACE, this test gives very precise picture regarding the genetic predisposition of the athletes, who want to develop themselves in sports which use mainly the lower limbs like football, athletics, weight lifting and tbc.

For athletes that want to specify in sports which needs the strength and density of the bone system we can use the VDR gene. An example can be sports that required strong forearm grip like djudo, climb-

ing, canu-kayak and sports gymnastics and etc.

METABOLIC PROFILE – CUSTOMIZE THE NEEDS, DOSE THE RESPONSE

Metabolic disorders occur due to the failure of metabolic pathways of several different molecules, namely carbohydrates, aminoacids, fatty acids and organic acids. A metabolic profile is also made to provide a better view of the genotype of the sport individual. This profile provides guidance on the metabolism of athletes, their genetic predisposition to lose weight, or to rapidly gain weight during preparation. This profile builds a picture of insulin and glycogen sensitivity, identifies levels of cholesterol, testosterone and other important hormones that affect athletic performance. This line of research is very well developed, as it affects not only elite sport but also a very broad segment of the sporting society.

Observing the study of the gene ADRA2B, a precise basal metabolic rate (BMR) is built, which is strictly individual and depends on many factors as height, weight, sex and age, as well as our genes. Based on a properly constructed metabolic profile, we can control and properly dose of loading the competitor, especially in cases where we have preliminary information about the genetic burden of muscle hypertrophy and a sudden increase in weight. This is especially important in sports whose competitions are held in categories where body control is of the utmost importance. An excellent example of personal weight control is a canoe-kayak sport, where the relative strength of the athlete per kilogram is of great importance for the movement and speed of the boat.

Exactly here is the place to pay special attention to the „Sports Recovery“, which has already been commented in sports science in two ways: first, recovery in the sense of the body's ability to recover energy loss and recovery from fatigue accumulated as a result of physical activity on the body. Second, the rate of recovery of injured tissue in the body due to trauma.

The genotype, which has the potential for faster recovery in both directions, will be preferred by specialists and dominant in the sports race. Confirmation of a clinical diagnosis through genetic testing allows for genetic counseling and may lead to adequate sport medical management. Generally,

one of genes for determining the metabolic profile is the ADRA2B gene, utilizes markers for the liver's detoxification system, which influences the lipid profile and a number of genes that determine the structure of proteins and fatty acids.

Based on the genetic studies of the metabolic profile, a number of nutrition specialists developed programs not only for the elite sport, but also for the amateur and grassroots sport fans. The sports supplements industry has received also a tremendous push, precisely on the basis of a number of genetic studies on a wide range of people. Many of those people are just trying to improve their appearance through physical activity or simply leading a healthy lifestyle.

Selection of the most appropriate genetic tests and also interpretation of the results must be made with careful consideration of the clinical symptoms in each case. It is important to look for combinations of genetic changes in the genes as well as to investigate the relevance of any biomarkers in detail.

INJURY RISK – THINK, PREVENT, TAKE CARE

One of the most important tasks of scientists working in this field is to determine the levels of injury risk. Many studies have shown that specific genotypes suffer from significantly higher injuries. Essentially, the risk-related risk groups are even segmented based on bone and joint injuries, soft tissues - muscle and tendons or combined trauma. Tendons and ligaments within the upper and lower limbs are some of the more common sites of musculoskeletal injuries during physical activity.

The traumatology test allows the elite athletes to know how to train and work preventively at an early stage in their careers in order to reduce sports injuries and the unnecessary risk of interrupting training for a certain period of time. Recent studies have identified genetic markers associated with risk for certain sports-related injuries and performance-related conditions. These markers could be used by individual athletes to personalize their training and diet regimens.

Recently, cases of infarcts of elite athletes during a competition have increased significantly, some of which, unfortunately, are fatal. Efforts are being made to work on the impact of physical activity on reducing cholesterol levels

and heart rate in order to reduce and prevent cardiovascular disease risks. During the long preparation process, it is important not only for high performance but also for the future health of the sports heart that it is important to study two genes: ADRA2B, which provides information on heart rate recovery, and PPARA, which provides us with information about upcoming changes and possible preventative measures to prevent left ventricular hypertrophy, which is characteristic of active athletes.

I would also like to briefly look at another popular case of tendon trauma among athletes. Sequence variants of the tenascin C (TNC) gene have been shown to be associated with Achilles tendinopathies and Achilles tendon ruptures, whereas a variant of the collagen V α 1 (COL5A1) gene has also been shown to be associated with Achilles tendinopathies. Both genes encode for important structural components of tendons and ligaments. The COL5A1 gene encodes for a component of type V collagen, which has an important role in regulating collagen fibre assembly and fibre diameters. The TNC gene, on the other hand, encodes for TNC, which regulates the tissue's response to mechanical load. To date, only variants in two genes have been shown to be associated with Achilles tendon injuries.

Early diagnosis makes it possible to properly structure the workload and helps reduce risky exercises that can lead to serious injuries and even to the end of your sports career.

CONCLUSION

Returning Science to Sport - Task # 1

In conclusion, it can be said that genetics in excellence plays a huge role in selection, early profiling, proper construction of the training process, nutrition and recovery of elite athletes. It is of paramount importance to work towards the prevention of health-risk workouts and workouts, as well as the prevention of severe traumatology, which will put an end to a promising career. De-

veloped societies have not underestimated or neglected the basic principles of sports science. Developed and upgraded their sports training systems, based on extensive research, part of which is genetically focused.

Nowadays, even the scientifically defined genotype of a particular individual cannot be categorical and the only factor in deciding on the career of a young athlete. Reading the human genome to a large extent allows us to build a better workout plan, better individual workout dose, make a proper recovery for the athlete and, last but not least, minimize the injury risk.

Yes, the genotype at its core can provide guidance for better construction of the entire training process, but it cannot be a major criterion, especially when it comes to desire, hard work, talent, discipline, deprivation and self-esteem... or lack thereof. a athlete.

And remember the blond and the fast-running boy from the beginning of the material! The blond girl who „lacked in sport“, became a great sport shooting champion. The fast-running boy with the „unique talent“ never could get on the big track and field. An early orphan, today he builds gyms where to train the elite athletes!

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