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## GENETICALLY DESIGNED VICTORY? – GENETICS, SPORT AND LEGAL DETERMINATION

### Abstract

Certain gene variants in the human genome (polymorphism of common genes or alleles) give a comparative advantage in performing physical activities. Some of them are directly related to the structure of skeletal muscles and the ratio of white and red fibres in them. It has long been known that marathoners have a different body constitution than sprinters and that the endurance of the former is based on lower body mass and a high percentage of red muscle fibres, and the explosive power of the latter on larger muscles and the percentage of white muscle fibres. The genetic makeup of muscle is not the sole source of athletes' advantage - genes that control the stress response affect physical strength but also the mental ability to cope with the pressure. Moreover, in sports competitions, genetic variations that lead to disease and, ultimately, injury or even death may give some specific physical advantage. In the case of Marfan's syndrome, persons with this gene variant have long limbs and elastic joints, which is preferred especially in ball sports, which makes them recognized by coaches. Unfortunately, playing sports increases their risk of dilatation and dissection of the aorta, as a result of its wall extensibility.

The human body has its physical limits that can be pushed by strenuous training. This stretching of the 'physical boundaries' must be paired with a suitable

regeneration time for the effect to be greater than the possible damage. Also, time for regeneration is often denied to the most successful competitors who enter from one round of the competition to the next - as is the case at the World Cup. In not a small number of cases, success is traded with speedy metabolic aging and deterioration of health due to injuries. Given the fact that the value of top athletes is measured by money and reputation, there is an understandable interest in new methods to make their careful selection and personalize their training and diet. Our current understanding of molecular genetics is at such a stage that its application is possible and a tempting option for top sports. The advent of CRISPER technology goes a step further and allows the design of as yet unseen human abilities. Although the genetic application is in its infancy, sport organizations have to take a stand on which tests and which interventions are bioethically justified in sport competition and which are absolutely unacceptable. The big question is – what is a legal regulation of the same issue? Is the law following (bio)ethically determined state? Generally speaking, it is important to investigate what are exact legal acts that are connected with genetics in the first place and then with the combination of genetics and sport. Also, the goal is to examine whether the relevant legal acts are forbidding any kind of altering of human genome, in general and in sport. Can it be said that the law is one of the major stopping-mediums and “dams” of the complete genetic research that tends to be transhuman?

**Keywords:** sport competitions, genomic analyses, athletic genes, sport science, law, legal protection

## Introduction

Sport science (kinesiology) and sport medicine are branches of basic and clinical science that closely deal with the physical performance and impact of various sport activities on health. The traditional interest of these professions, ever since ancient times and the origins of modern medicine, was to improve physical endurance and health (Hill, 2019). The link between proper nutrition and sport performance, as well as the link between the type of sport and possible sport injuries, was recognized very early on. Also, the benefits of physical training for mental health (Paluska and Schwenk, 2000) and vice versa, the benefits of mental training for athletic performance, were well studied (Vealey, 2007). In breaking some world records, it was not only the fitness and mental stability of athletes that was crucial but also the attitude of the profession towards human boundaries. One of the best-described examples was the mile run world record by Roger Bannister who, on May 6, 1954, at the Iffley Road (UK) racetrack, ran under four minutes (3:59.4) (World Athletics, News, Bannister, first man to run a sub-four-minute mile, dies)<sup>1</sup>. Since the first precisely measured running tracks were built in the 1850s, the men's record time dropped from 4:28 (Charles Westhall, 1885 in London; in: Schiffer, 2008) to 4:01.4 (Gunter Hägg, 1945 in Malmö; World Athletics, Athlete Profile - Gunter Hägg)<sup>2</sup> over the course of a century. Four minutes was considered the physiological limit of the human body and the record held for nine years. Bannister himself was a medical doctor with a deep interest in human physiology, autonomic nervous control of cardiovascular function in particular (MacAuley, 2005). After competing in the 1952 Olympics in Helsinki, which he considered a personal failure (fourth place in the 1500 m final), he set himself a new goal - to run a mile under four minutes. Bannister was deeply convinced that the 4-minute limit was imaginary and used inventive 400 m interval training combined with periodic total rest for the purpose of recovery (supervised by his coach - Franz Stampfl) to break it. His record was broken in just 46 days (John Landy, June 21<sup>st</sup>, 1954 in Turku) in support of the hypothesis that the 4-minute figure was a tempting mythical limit just because it was very close to the record at the time and because it was round. The current men's mile record is 3:43.13, held by Hicham El Guerrouj since 1999. Given that it has not been broken in 21 years (World Athletics, All-time Top lists, One

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1 URL: <https://www.worldathletics.org/news/iaaf-news/roger-bannister-obituary>, Retrieved: 2020-07-30

2 URL: <https://www.worldathletics.org/athletes/sweden/gunder-hagg-14862355>, Retrieved: 2020-07-30

MileMen),<sup>3</sup> it is probably closer to the physiological limit of human endurance in this discipline.

The marathon is the next discipline that occupies the human quest for boundaries. This tests the endurance of a completely different type from the one required for short and middle distance races. Marathoners risk metabolic collapse (the same one that cost Pheidippides, the famous Marathon victory herald, his life) if they do not manage to develop the necessary metabolic flexibility through long and persistent training and allow themselves a long enough period of regeneration between two races. International Association of Athletics Federations (IAAF) standardized marathon distance in 1921 to 42.195 km. However, the first listed record dates back to 1908 when Johnny Hayes took the place of a disqualified Italian runner Dorando Pietri who got help in front of the finish line (London, UK) (Marathon Guide, Men's World Record Times - 1905 to 1911).<sup>4</sup> The men's record (2:55:18.4) back then was broken by more than 50 minutes in the next 110 years (2:01:39, Eliud Kipchoge, Berlin Marathon 2018). The circumstances surrounding this record were very similar to those of the Roger Bannister era – 2:00:00 is a round figure very close to a possible human physiological limit. The most likely record striker, Kenyan long-distance runner Eliud Kipchoge, adopted an intermittent style of training which originates from Roger Bannister. Also, he is one of the three front-faces of Breaking2. Nike campaign announced in November 2016 (Runner's World, Nike's Audacious Plan: Break the 2-Hour Marathon Barrier in 2017).<sup>5</sup> While Bannister fought against mainstream myth using knowledge of physiology and psychology, Kipchoge added technology to this. Nike's research team developed personalized shoes The Nike ZoomX Vaporfly 4% and studied the influence of resistance and humidity of air to enhance the prospect of breaking the 2-hour limit. The first attempt to attack the record took place on May 6, 2017, in Monza, Italy. With the help of six pacers in a triangle formation who watched the big clock display and followed a green laser light indicating proper speed, Eliud Kipchoge reached the time of 2:00:25 and broke the world record of that time. The attempt was successfully repeated on October 12, 2019, at Prater Park, Vienna when he broke the imaginary line and achieved 1:59:40. Neither result was recognized by

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3 URL: <https://www.worldathletics.org/records/all-time-toplists/middle-long/one-mile/outdoor/men/senior?regionType=world&page=1&bestResultsOnly=true&firstDay=1900-01-01&lastDay=2020-08-13>, Retrieved: 2020-07-30.

4 URL: <http://www.marathonguide.com/history/records/popupSummary.cfm?RID=MMAR19080724>, Retrieved: 2020-07-30.

5 URL: <https://www.runnersworld.com/news/a20837736/nikes-audacious-plan-break-the-2-hour-marathon-barrier-in-2017/>, Retrieved: 2020-07-30.

IAAF due to the fact that these competitions were not open. Rather, all runners were carefully selected and responded to the invitation. In the meantime, Nike developed the next generation of breaking record shoes - the Nike ZoomX Vaporfly Next% - the use of which is controversial in races of this type because they give an undoubted advantage to competitors. The recognized world record still stands at 2:01:39 (Eliud Kipchoge, Berlin Marathon 2018) (World Athletics, All-time Top lists, Marathon Men).<sup>6</sup>

The Breaking2.Nike initiative showed that victory can be designed and the design consists of choosing the right competitors, their personalized programmed diet, training regimen, developing positive habits that contribute to metabolic flexibility/mental stability and understanding the environmental factors that contribute to choosing the right time to start the competition. If we add the genotyping of the most successful long-distance runners to this as the basis for the selection of competitors in some subsequent competitions, success is almost guaranteed.

The first goal of this paper is to give a brief overview of a recently discovered tool for advancing victory - genomic tests. The authors will also discuss possible gene-oriented personalized interventions affecting athletic performance and the dangers and bioethical problems that lie in their application. The second goal of this paper is to examine the legal situation of the whole issue. This especially pertains to the relevant legal acts and the provision of the same acts that are regulating the whole issue of a combination of genetics and sport to give an answer to the question is altering a human genome for the purpose of sport a legally valid action and in what way?

### **Review of previous research**

All organisms have a genome that interacts with the environment to produce a phenotype. Genes for the one and the same trait come in different variants (alleles) and give rise to genomic variability (reflected, for example, as skin colour). Environmental factors further affect the appearance and variability of a phenotype (tanning of the skin under the influence of the sun). In the case of many physical characteristics (e.g. height or weight) the total population variability has a normal distribution and can be described by a Gaussian curve.

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<sup>6</sup> URL: <https://www.worldathletics.org/records/all-time-toplists/road-running/marathon/outdoor/men/senior?regionType=world&drop=regular&fiftyPercentRule=regular&page=1&bestResultsOnly=true&firstDay=1900-01-01&lastDay=2020-08-13>, Retrieved: 2020-07-30

When searching for some extreme traits (e.g. intelligence) scientists are usually interested in a small percentage of individuals who are at one end of the curve.

The similar logic is applied in the genomic selection of elite athletes. We assume that their exceptional skill is the result of their genetic makeup and environmental factors (training, diet) and they are at the very end of the Gaussian curve. The curve can be drawn for any interesting characteristic, such as running speed, strength, flexibility, maximum oxygen uptake, etc. In Genome Wide Association Studies (GWAS), scientists tried to compare the genomes of elite athletes (selected on the base of their exceptional physical characteristic and sport success) with the general population and look for gene variants that are significantly more often found in athletes than in general population. The success in pinpointing candidate genes is greater if the term 'elite athlete' is better defined. The larger the population of these athletes, the larger the general population tested. How large these numbers need to be in order to be able to draw reliable conclusions based on them can be seen from the most successful GWAS made so far for the purpose of determining the genetic basis of chronic polygenic diseases such as cardiovascular disease and diabetes. Two large consortia of scientists, CARDIoGRAM (Schunkert, Erdmann and Samani, 2019) and C4D (Peden et al., 2011), have joined forces to find risky gene variants (polymorphisms) involved in the development of cardiovascular disease. Their joint CARDIoGRAMplusC4D study (Nikpay et al., 2015) identified 56 at-risk gene loci, and analysed more than 180,000 participants, about half of whom had cardiovascular disease. GWAS done for the purpose of identifying diabetes risk included 62,892 diabetics and 596,424 healthy controls, and 143 risk loci were identified (Xue et al., 2018). For comparison, one of the most recent GWAS studies of endurance athletes included 1520 samples of athletes and 2760 samples of the general population (Rankinen et al., 2016). A metabolic study designed to identify genetically-influenced metabolites associated with athletic performance was performed on samples of 490 elite athletes who passed doping control at international competitions, and the study did not include samples of the general population (Al-Khelaifi et al., 2019). In addition to the problem that the studies of athletes were made on a small number of samples, the search for candidate genes is complicated by the fact that success in different sports depends on different phenotypic characteristics. For example, sprinters are more successful if fast-twitch fibres predominate in their muscle mass, while marathoners are the exact opposite – slow-twitch fibres should predominate in their muscle mass (Trappe et al., 2015). In addition, the classification of the elite

(successful) from the so-called non-elite (unsuccessful) athletes or the general population is complicated by the fact that, in spite of genetic make-up, proper training is crucial for sport success (Guth and Roth, 2013). It is, therefore, not surprising that none of the 249 identified genes that may represent an advantage for athletic performance are a good predictor of the success of the person who possesses it (Camporesi, 2013).

A good example of such a genetic trait is alpha-actin skeletal muscle isoform 3 or F-actin cross-linking protein (ACTN3) whose polymorphism was linked back in 2003 to success in sports that prefer a sprint body type (Yang et al., 2003). The gene encodes a protein that participates in the assembly of the contractile apparatus of a muscle fibre. Two variants of this gene were investigated, of which the first, R577X, is present in almost 45% of the world's population, and the second, 577XX, is found in 20% of the world's population (Amorim et al., 2015). People with the first variant express the protein in the fast-twitch muscles that make up a larger proportion of the sprinter's muscle mass. Carriers of the second variant do not express the protein encoded by this gene at all. Namely, it is a mutation that does not cause disease, but significantly affects muscle development and is an example of very rare mutations in the loss of function in the human genome that give some advantage to their carriers (MacArthur et al., 2012). To make things more interesting, the first variant is associated with the success of sprinters, but the second variant is associated with the success of marathoners and scientists even consider that it gave a selective advantage in the movement of modern humans from Africa (Lee, *et al.*, 2016). Nevertheless, both variants account for only 2% of total muscle performance while the remaining 98% are attributed to environmental impact (Eynon et al., 2011). This is why the genotyping of this gene locus has little predictive value (web source: McArthur D. (2008) The ACTN3 sports gene test: What can it really tell you? Genetic Future).<sup>7</sup>

Another example is the ACE gene encoding angiotensin-1 converting enzyme. This enzyme is possessed by a number of cells, but its role in vascular endothelial cells and participation in the renin-angiotensin system, which is responsible for controlling blood pressure by regulating body fluid levels, is considered particularly important. Two variants of this gene have been described. The first has an insertion of 287 base pairs associated with less enzyme activity (so-called variant I). In the second case, this sequence is missing (deleted or D variant)

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<sup>7</sup> URL: <http://scienceblogs.com/geneticfuture/2008/11/30/the-actn3-sportsgene-test-what-it-really-tell-you/>  
Retrieved: 2020-07-30.

and the enzyme is less active (Rigat et al., 1990). The claim that people with the ACE I / I genotype have an advantage in sports that require great endurance while people with the D / D genotype are better in sports that require strength has prompted a number of conflicting opinions (Ma et al., 2013). If we take into account the fact that 90 out of 100 elite marathoners of all time are of Kenyan or Ethiopian descent, who hold six consecutive world records in this discipline (International Association of Athletics Federations, 2018), there is no significant difference in the frequency of ACE I / D alleles between them and their respective general population (Scott et al., 2005). Therefore, it is obvious that a larger number of genes are responsible for their athletic success. Ahmetov and co-workers hypothesized that there were at least 93 genes (Ahmetov et al., 2009) but none of them were specific to the marathon alone (Moir et al., 2019).

In addition to select future elite athletes, genetics tests are also used as a tool of sports nutrigenomics to personalize nutrition in athletic populations. Since World War II (Ministry of Food (1951) *The Urban Working-Class Household Diet 1940 to 1949. First Report of the National Survey Committee*. His Majesty's Stationery Office, London),<sup>8</sup> it has been common to issue 'one-size-fits-all' dietary recommendations and food rationing to ensure a nutritional minimum that ensures the health of the population and fair distribution of available food. This is still applicable today in emergencies in its advance form (web source: UNHCR, UNICEF, WFP, WHO. *Food and Nutrition Needs in Emergencies*).<sup>9</sup> Recently, dietitians got a more complete understanding of individual genetic differences regarding absorption, uptake, metabolic conversion, nutrient utilization, and excretion that are within physiological limits or belong to pathological variants. Lactose intolerance, caused by a relatively common genetic variant, has no severe consequences and is easily controlled (Storhaug, Fosse and Fadnes, 2017). Dietary fructose intolerance and phenylketonuria, which are less common, have more severe consequences and require a special diet (Macleod and Ney, 2010). However, if the patient adheres to them, health consequences are preventable (Jurecki et al., 2017). Examples of genetic variants which result in the inability to exploit an energy resource have led to the realization that 'one-size-fits-all' recommendations do not suit everyone and can seriously impair the quality of life of those who bear them. So-called personalized diets today are mostly used in the treatment of various diseases (e.g. metabolic and cardiovascular), but also in achieving optimal athlete performance (van Ommen et al., 2017).

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8 Available at: [https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment\\_data/file/549165/The\\_Urban\\_Working\\_-\\_Class\\_Household\\_Diet\\_1940-1949.pdf](https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/549165/The_Urban_Working_-_Class_Household_Diet_1940-1949.pdf).

9 URL: <https://www.unhcr.org/45fa745b2.pdf>.



A joint position statement from the American Dietetic Association, Dietitians of Canada, and the American College of Sports sets out views on the impact of diet on athletic performance and recovery from exercise (American College of Sports Medicine, American Dietetic Association and Dietitians of Canada, 2000). The release lists evidence-based analyses regarding the satisfaction of energy requirements, the use of macronutrients and micronutrients, and optimal hydration. It is especially emphasized that physical activity increases the need for micronutrients that participate in energy production, regenerate muscle tissue, blood cell production, reduce the effects of oxidative stress, maintain the immune system, etc. Gene variants that reduce the ability to replenish micronutrients can significantly reduce athletic performance (Guest et al., 2019). Nutrigenomics allows the design of a precise personalized diet that corrects such deficiencies, not only to improve performance but also to preserve health or maintain reproductive fitness, especially in the case of women.

Dietary recommendations for athletes usually also include daily doses of supplements. As much as it is harmful to the health of athletes to omit supplements, it is even more dangerous to use them in excess. A good example is an iron replacement in athletes who have unfavorable variants of the *TMPRSS6* (increased risk for low levels of transferrin and hemoglobin saturation), *TFR2* (increased risk for decreased red blood count), and *TF* (increased risk for low ferritin) genes regulating hormone hepcidin expression which is crucial for the absorption of iron from the digestive tract. Due to the deficiency of iron absorption, these athletes have an increased risk of developing anemia (Guest et al., 2019), a condition that requires 3-6 months for reversal. In contrast, athletes with a pathological variant of the HFE gene are at risk for the chronic disease hemochromatosis due to iron overload. Although people with this disease have a genetic advantage in various sport disciplines (Semenova et al., 2020), iron replacement in their case pushes them even more into a pro-oxidative state and exacerbates the underlying disease (Zoller, 2004). Knowledge of individual genetic variability, along with biochemical tests to check the actual condition, allows for optimization and has the greatest effect on athletic success.

Today, dietitians have a whole list of genes that are interesting in terms of giving accurate dietary recommendations (genes that affect liver metabolism, absorption of vitamins and micronutrients, or even body composition). It has also been proven that disclosure of genetic information can motivate behavioral change and help introduce new habits (Nielsen, 2012). It should be noted that there are still a small number of studies that have investigated the interaction of

genes and diet in large enough cohorts of competitive athletes and controls that their results could be considered strong evidence.

## **Discussion**

From the point of view of sport medicine, genetic screening tests have the greatest justification if they are used for the purpose of predicting sport-related injuries or performance-related conditions. Sudden cardiac death, usually of younger athletes, is not common, but it is devastating. It is a consequence of hereditary structural heart diseases or congenital electrical disorders (arrhythmias, canalopathies) associated with known genetic variants (Sarquella-Brugada et al., 2013). Such events cannot be completely avoided, but the superficiality of clinical examinations of young athletes is still surprising. Genetic tests that would predict such incidents have not yet been applied to athletes due to the rarity of these diseases but became a medical standard for preventing sudden death within affected family. In the last decades, there have been several sudden deaths on sport fields (Hank Gathers, Reggie Lewis, Haris Charalambous – Magic Harris) in athletes with Marfan syndrome - an autosomal dominant connective tissue disease associated with a mutation in the *FBNI* gene (Loeys et al., 2010). These athletes more often engage in sports in which long limbs (basketball, volleyball, baseball) or elastic joints (skating) are an advantage. Diagnosis is a sufficient reason for their disqualification from the further competition (as it was the case with Isaiah Austin or Kayla Burt) for the association of this mutation with aortic dilatation and sudden cardiac death (Braverman et al., 2015).

Recently, there has been an increased interest in the genetics of sport injury prevention. Goodlin et al. (2015) reported single-base polymorphisms (SNPs) in various genes that increase the risk for low bone mineral density and stress fracture (67 SNPs), osteoarthritis (7 SNPs), anterior ligament tear (4 SNPs), and Achilles tendinopathy (6 SNPs). The same paper lists 39 SNPs associated with poorer athletic performance due to low circulating levels of iron, calcium, magnesium or vitamins E, D, B12, B6, B9, or risk levels of phytosterol and plasma homocysteine (Goodlin, et al., 2015). Genetic tests to identify these variants (polymorphisms) would be justified if done solely for the purpose of designing a personalized diet and injury prevention, but the possibility of their misuse to lower the price for a professional athlete or increase the amount of insurance for him or her cannot be ruled out.

The flooding of the market with direct-to-consumer (DTC) genetic tests for sport performance aimed primarily at coaches and parents has prompted the reaction of a number of national sports organizations (Webborn et al., 2015; Vlahovich et al., 2017; Loland, 2015). These tests are mostly resented due to the lack of quality control and genetic counselling for the interpretation of genetic data to the consumer (Roth, 2012). However, even more objections should be made for the lack of evidence that either one of the tested genetic variants predicts an athlete's performance better than the quality and length of training (Camporesi, 2013). Early talent directing and long-term training are known combinations for success, so it is not surprising that some parents feel the urge to provide their children with a head start using DTC genetic tests. On the other hand, reaching for such tests takes away the ethical and educational dimension of sport - it turns sport into a purpose in itself. In addition, children are deprived of the role of the subject in making their own choices. Even more frightening is the possibility of designing a baby. CRISPER technology opens the door to such possibilities. This new tool in molecular biology can be used to correct point mutations in both fetuses and adults, with some limitations related to target tissues. Correction of unfavourable traits is just one possible application which could be followed by upgrading and rearranging genetic material. All of the above could substantially change the way that we look at sport and society in general.

### **Legal view on the stated issue**

#### *Bioethical thought in a legal framework*

The main mission of the law is to regulate all possibly predicted acts, behaviours, and events in modern society. The legal norm must be the pure reflection of society itself or to be precise about its general moral views, trends, and needs. Bioethical challenges provide a big assignment for law in general, which is to regulate all activities which arise from modern bio-research. Since the appearance of that kind of research, many different ethical issues arose. Now, the law has another complicated assignment – to deal with ethics. The relationship between the law and ethics/moral is one of the most crucial, but sometimes very controversial, parts of legal philosophy and jurisprudence. The legal aspect can give some sort of guidelines in solving ethical obstacles, but, very often, ethical and moral obstacles cannot be solved merely by the law (Nedić, 2020: 257). The Latin and Roman law maxim stated, *non omne quod licet honestum*

*est* (not everything what is legal, is also honest/just/moral; see: Petrak, 2010: 95), dividing the law (in the way of legal positivists, as opposed to natural view of law) from all other entities: moral (ethics), justice, and freedom. However, due to all the differences between the terms, the fact is that the legal and ethical norms should be harmonised as best they can (Nedić, 2020: 257).

In that sense, there are two bioethical approaches of the researched problem. The first is the conception of genetic testing used for the purpose of improving health or preventing the occurrence of diseases and sport injuries. The second one is the usage of genetic tools for the purpose of creating the so-called designed baby and for the purpose of 'improving' the performance of adult athletes or, as we can say, a tendency to create some sort of superhuman in the field of sport. There is no doubt that, from an ethical and legal perspective, we can mark the first approach as purely justified, but can we do the same with the second one? If a genetic tool can cause severe damage to human health just in order to be successful in sport, can human society justify it, first in an ethical way and then in a legal one? In the ethical way, there has already been genetic and (bio)ethical research done which has underlined the ethical question and debates of these kinds of ventures (e.g. Zajc Petranović et al., 2019). According to authors in the field of philosophy and bioethics of sport, when discussing the ethics of doping in general, but also in relation with human genetics, "four principles should be applied: harm principle ("are the procedures harmful to the athletes and are they disrupting and damaging the athlete's health?"), fairness principle ("gaining of an unfair advantage that needs to be assessed"), paternalistic principle ("should every athlete of age have the freedom to decide whether they want to change their genes?"), and coercion principle ("how can we be sure that the athletes have made their decisions autonomously and without coercion?")" (Škerbić, 2016; Schneider, 2016). However, it is also very important to emphasize that there is indeed the possibility that correcting some of the genetic variants that necessarily lead to the disease could improve the health of the person who has the same variant, which can also affect that person's physical ability. In this case, treatment has priority and the 'side effect' of treatment, which is seen as better athletic success, should not necessarily be interpreted as undesirable genetic engineering.

When it comes to the "property" of human body, Michel Aramini (2009: 266) underlines the principle of autonomy and principle of defence of physical life. Aramini sees the human body as unavailable human property, not the fact that the human body is indeed in any kind of disposition (principle of autonomy)

where, in a Kantian way, the human being is always a purpose and never any sort of medium (principle of defence of physical life) (Aramini, 2009: 254). What is the reflection of these kinds of thoughts in relevant legal regulations?

### *International law and the issue of human health*

As we can see it here, one of the central issues of this kind of approach is related with the elaboration of human health. The big question is how does law (especially acts of international law) perceive the whole situation relating to health?

The first concept is in an argument of legal protection of human life and human health. The right to health is a right that does not apply exclusively to the individual and his right to be “healthy” (individual right), but also the right that applies to the whole group of people (collective right), in a way to enable the collective a healthy life, healthy environment (environment), sustainable development, etc. Although there is no explicit term ‘right to health’ in international and European legal acts, the same term is itself inclusive, which means that it includes several important legal concepts. The right to health includes certain individual rights of the first (personal and political rights and freedoms) and second (economic, social, and cultural rights and freedoms) generation, but also collective rights of the third generation (such as the right to a healthy life and environment). According to Art. 25. of The Universal Declaration of Human Rights (United Nations, General Assembly resolution 217 A, 1948) “everyone has the right to a standard of living adequate for the health and well-being of himself and of his family, including food, clothing, housing and medical care...”. Art. 12 of the International Covenant on Economic, Social and Cultural Rights (United Nations, General Assembly resolution 2200A (XXI), 1966) states that “the States Parties to the present Covenant recognize the right of everyone to the enjoyment of the highest attainable standard of physical and mental health.” It is the same with the regulation in the acts of the Council of Europe – the European Convention on Human Rights (Council of Europe, 1950) and (Art. 11) of the European Social Charter (Council of Europe, ETS No.035, 1961). There is another important concept that the constitutions of all modern states provide (Art. 23 of the Croatian Constitution, Official Gazette 56/90, 135/97, 08/98, 113/00, 124/00, 28/01, 41/01, 55/01, 76/10, 85/10, 05/14) – forbidden medical or scientific experimentation without consent. According to Art. 7 of the International Covenant on Civil and Political Rights (United

Nations, General Assembly resolution 2200A (XXI), 1966) “...no one shall be subjected without his free consent to medical or scientific experimentation”, which is a concept that emanates from Art. 5 of the Universal Declaration of Human Rights, Art. 3 of the European Convention on Human Rights.

If international legal acts are protecting the whole concept of human life and health, then the conclusion must be clear. The use of genetic tools for the purpose of creating the so-called designed baby, but also for the purpose of ‘improving’ the performance of adult athletes, is a concept that violates the basic principles of human rights, more precisely the right to life and health standards of life. However, we are coming to an issue that the concept of human rights and human health cannot deal with. Genetic improvement of adult athletes is not something that is strictly violating the law in the aspect of right to health. As we saw, the only forbidden aspect are the medical and scientific pieces of research performed without consent. However, what if an athlete gives his written consent? In addition, there is a well-known legal and bioethical question of the status of the person for an unborn child and the fact that an unborn child is not the subject of law and has no legally determined rights. Meaning, if in some countries it is legal to “kill” an unborn child (abortion), is it really so bad to turn him into a superhuman? The legal concept of human health is only the first (of two) “sieves” of solving this kind of issue.

### **Legal protection of genetic research**

These two above mentioned aspects and questions of an adult athlete and unborn child are no longer related to human rights and health, but to legal protection of genetic research, which is the second “sieve” of solving this issue. The first problem, when it comes to law and genetic research, is the fact that there are not many concrete, unified, and universal legal acts which regulate this kind of issue. This leads us to the issue that many countries are legally solving and regulating genetic research in their own way. For example, although it is stated that “a large body of law exists in the member states of the European Union regarding the implementation of genomics technologies”, in its own *Overview of EU National Legislation on Genomics* (European Commission, JRC Science for Policy Report, 2018: 4-5), the European Commission (respectively JRC Science for Policy Report) underlines three report highlights:

- “1. The existing national legislations seem to focus on genes, their variations and heritability in living organisms, while scientific interests and

advancements, at the moment, have a more holistic view of the genomes of living organisms. Further reflections are needed to address this important gap in the legislative framework.

2. There is a number of areas that are differentially addressed in the regulatory frameworks of the different member states, such as human somatic and germ line modifications (through the application of gene and genome editing technologies),
3. Different legislative frameworks take into consideration the citizens'/ patients' rights of having their personal genomic data characterised, used, and shared. An online interactive compendium should be produced and released, that would include all the legislative instruments retrieved and analysed for this report."

The same report also underlines that there is "the absence of an explicit EU legal framework on genomics (European Commission, JRC Science for Policy Report, 2018: 74)". On the EU level, the GDPR<sup>10</sup> and Regulation 536/2014 on clinical trials<sup>11</sup> both regulate genetic research to a limited extent (Pormeister, 2018: 709). Pormeister also emphasizes that "it must be noted that Regulation 2017/746 on in vitro medical devices<sup>12</sup> (applied as of May 26, 2022) will regulate genetic testing as well, however, only in a healthcare context and not regarding research."

Comparatively, in Austria (The Gene Technology Act, BGBl. Nr. 510/1994, last amended BGBl. I Nr. 59/2018), Lithuania (The Law on Ethics in Biomedical Research of 2002), and the Netherlands (The Law on Medical Research on Humans of 26 February 1998) there are laws that are explicitly regulating the whole aspect of genetic research. In those places, it is forbidden to perform experiments on humans in a way that changes the germ line of humans (see: European Commission, JRC Science for Policy Report, 2018: 10). In other countries (The German Embryo Protection Act (1990) No 69/1990, The Maltese Embryo Protection Act (2013), The Portuguese Law no 12/2015 of 26 January on

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10 Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation), Official Journal of the European Union, L 119/1

11 Regulation 536/2014 on clinical trials on medicinal products for human use, and repealing Directive 2001/20/EC, Official Journal of the European Union, L 158/1

12 Regulation 2017/746 of the European Parliament and of the Council of April 5, 2017 on in vitro diagnostic medical devices and repealing Directive 98/79/EC and Commission Decision 2010/227/EU. [2017] Official Journal of the European Union L117/76.

Personal Genetic Information and Health Information, The Slovenian Medicines Law (2014), The Swiss The Federal Act on Research Involving Embryonic Stem Cells (2003)), although there may not be any laws that explicitly regulate the issue of genetic research, alteration of the human germ line is also forbidden. The same fact also appears to be the case with the Croatian legislation where no specific and explicit genetic technology legislation could be found. The most relevant act of this issue is the Law on the Protection of Patients' Rights (Official Gazette, No. 169/04, 37/08) where, according to Art. 22 of the same Law "interventions directed at changing the human genome can only be undertaken for preventative or therapeutic purposes and no interventions are allowed with the view to changing the patient's germ line."

However, while there is room for improvement regarding regulations of this kind of issue, in member states of EU and EU law in general there is an applicable legal act on the basis of the Council of Europe.<sup>13</sup> According to the Convention on Human Rights and Biomedicine<sup>14</sup> (Council of Europe, ETS No.164, 1997), in Art. 12 it is stated that "tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counselling." More relevant for the above stated issue, in the next article (13), the Convention is very clear: "An intervention seeking to modify the human genome may only be undertaken for preventive, diagnostic or therapeutic purposes and only if its aim is not to introduce any modification in the genome of any descendants." It is important to emphasize that this Convention is an international treaty ratified by 29 European countries (not only EU members; Council of Europe, Chart of Signatures and Ratifications of Treaty 164),<sup>15</sup> and which is, by the legal force, above the application of the law. According to the same Convention and interpretation of its mentioned provisions, genetically designing a "superhuman" is definitely not acceptable.

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<sup>13</sup> "More specifically, the laws that would need to be evaluated concern:

...

4) Application of gene and genome editing technologies. Besides the above considerations on human applications of gene editing technologies, legislators should also address how to regulate the application of such technologies on animals and plants, especially on species relevant to the food and feed industry."

<sup>14</sup> Full name: Convention for the protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine (with its two Protocols)

<sup>15</sup> URL: [https://www.coe.int/en/web/conventions/full-list/-/conventions/treaty/164/signatures?p\\_auth=hY6c3JTC](https://www.coe.int/en/web/conventions/full-list/-/conventions/treaty/164/signatures?p_auth=hY6c3JTC) , Retrieved: 2020-08-4.



On the basis of the United Nations, it is important to mention UNESCO's Universal Declaration on the Human Genome and Human Rights (General Assembly resolution 53/152 of 9 December 1998), UNESCO's International Declaration on Human Genetic Data (32 C/Resolution 15, 2003), as well as UNESCO's Universal Declaration on Bioethics and Human Rights (32 C/Res. 24, 2005) as one of the most important international legal acts in the field of bioethics in general (see: Kirby, 2009). According to UNESCO's International Declaration on Human Genetic Data: "human genetic data and human proteomic data may be collected, processed, used and stored only for the purposes of: (i) diagnosis and health care, including screening and predictive testing; (ii) medical and other scientific research, including epidemiological, especially population-based genetic studies, as well as anthropological or archaeological studies, collectively referred to hereinafter as "medical and scientific research"; (iii) forensic medicine and civil, criminal and other legal proceedings, taking into account the provisions of Article 1(c); (iv) or any other purpose consistent with the Universal Declaration on the Human Genome and Human Rights and the international law of human rights." In that sense, an act that is directly related to the topic of the human genome is the Universal Declaration on the Human Genome and Human Rights where, in Art.1, it is stated that the human genome is "the heritage of humanity" determining the human being as it is. According to Art. 10, "no research or research applications concerning the human genome, in particular in the fields of biology, genetics and medicine, should prevail over respect for the human rights, fundamental freedoms and human dignity of individuals or, where applicable, of groups of people." It is important to mention Art. 11 where it is stated that "practices which are contrary to human dignity, such as reproductive cloning of human beings, shall not be permitted" and also Art. 5 where it is stated that "research, treatment or diagnosis affecting an individual's genome shall be undertaken only after rigorous and prior assessment of the potential risks and benefits pertaining thereto and in accordance with any other requirement of national law." Although they are so-called *soft law* acts, these acts show a very important tendency of United Nations, as the most important and influential international organization and subject of international law, to legally regulate those kinds of ventures that are related to the human genome and genetics in general. According to the stated provisions, altering the human genome for the purposes of sport performance definitely could not be acceptable.

Designing a superhuman is definitely not acceptable according to another parameter. According to the *Code* of the World Anti-Doping Agency (Art. 4.2.2. of *World Anti-Doping Code*, World Anti-Doping Agency, 2021) in the newest *Prohibited List* (Section M3 *Gene and cell doping*, World Anti-Doping Agency, 2020) it is stated that “the following, with the potential to enhance sport performance, are prohibited:

1. The use of nucleic acids or nucleic acid analogues that may alter genome sequences and/or alter gene expression by any mechanism. This includes but is not limited to gene editing, gene silencing and gene transfer technologies.
2. The use of normal or genetically modified cells.”

It is important to clarify that the World Anti-Doping Agency does not only prohibit substances, but also methods in which forbidden gene and cell doping are part of it. These kinds of methods, like all defined and listed methods in general, are prohibited at all times, not just in the time of competition or in particular sports (*Prohibited List*, World Anti-Doping Agency, 2020). This leads us to the conclusion that designing a “superhuman” in sport is not only unacceptable from an ethical perspective, but also from a legal one. It is not acceptable not only because it may harm human health, but also because of the fact that the same research is violating the pure essence of a human being, making him into something that is not provided by nature itself. Clearly, these two reasons, human health and preservation of the human genome, are two main reasons why the law is not justifying these kinds of ventures.

## **Conclusion**

The distinctly competitive nature of sports and the orientation towards the market and the entertainment industry poses a temptation to athletes and their managers to increase the performance to the very limits by using all possible means. Athletes can achieve a high marketing price so investors use some traditional methods of their selection, but increasingly resort to unconventional ways of identifying potential talents (Breitbach, Tug and Simon, 2014). Few manage to pass the strict control of constitution, fitness, and self-discipline, and marketing mistakes come at a high price. In recent times, sport science and sport medicine had integrated knowledge from genetics (Trent and Yu, 2009) and nutrigenomics (Guest et al., 2019). Genetic tests have a few possible goals; to detect the so-called ‘performance enhancing polymorphism’ for the purpose

of early identification, programmed training, and dietary regime of potential athletic talents, rule out the existence of 'life-threatening polymorphisms' the negative consequences of which are triggered by physical exertion, creating personalized dietary instructions and a personalized training regimen. If they are used for the purpose of improving health or preventing the occurrence of diseases and injuries, the public opinion considers them to be ethically justified.

To date, more than 200 gene loci have been identified that could affect performance, but there is no valid evidence to justify their use in identifying future elite athletes for any of them. That is why early testing and forcing hard workouts is extremely ethically dubious. The use of genetic tools for the purpose of creating the so-called designed baby, but also for the purpose of 'improving' the performance of adult athletes, is dubious. Existing genetic tools still suffer from childhood diseases, but it is to be expected that they will develop rapidly, and the human race will find it difficult to resist the challenge of designing a superhuman.

The elaborated ethical and especially legal unacceptability regarding this issue is, therefore, determined by two issues. The first is the parameter of legal protection of human health (first "sieve"), which of course this kind of research and venture may bring. The second one is the legal regulation of the whole issue of genetic modification (second "sieve") which is not oriented primarily on the protection of human health. In that sense, the law must be oriented towards regulating and preventing not only genetic research that may be harmful for human health, but that kind of genetic research that is changing the whole nature and essence of the human being. Designing a "super-human" in sport and in general, apparently proved to be that kind of research. The law, apparently, really is an important stopping-medium and "dam" of complete genetic research that tends to be transhuman. Only three decades ago, sport organizations were obsessed with looking for potential fraud in the performance of men in women's disciplines. According to the newest research papers, in the near future sport organizations could be prosecuting and chasing "super-humans" who compete in human disciplines.

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## **GENETIČKI DIZAJNIRANA POBJEDA? - GENETIKA, SPORT I PRAVNO ODREĐENJE**

### **Sažetak**

Određene genske varijante (aleli), daju komparativnu prednost kod obavljanja fizičkim aktivnostima. Neki od njih izravno su povezani sa gradom mišića odnosno omjerom bijelih i crvenih vlakana. Odavno je poznato da maratonci imaju drugačiju tjelesnu građu od sprintera te da se izdržljivost prvih temelji na nižoj tjelesnoj masi i visokom postotku crvenih mišićnih vlakana, a eksplozivna snaga potonjih na većim mišićima i postotku bijelih mišićnih vlakana. Genetička podloga sastava mišića nije jedini izvor prednosti nekog sportaša – primjerice geni koji kontroliraju stresni odgovor utječu na fizičku snagu, ali i na mentalnu sposobnost nošenja s pritiskom. Štoviše, u sportskim natjecanjima, varijacije gena koje dovode do bolesti, a u konačnici do ozljede ili čak smrti, mogu pružiti određenu fizičku prednost. U slučaju Marfanovog sindroma, osobe s ovom varijacijom gena imaju dugačke udove i elastične zglobove, koji predstavljaju prednost osobito u sportovima s loptom, radi čega budu prepoznati od strane trenera. Na žalost, bavljenje sportom povećava im rizik od dilatacije i disekcije aorte kao posljedice rastezljivosti njene stijenke.

Ljudsko tijelo ima svoje fizičke granice koje se napornim treninzima mogu pomaknuti. Ovo rastezanje 'fizičkih granica' mora biti upareno s prikladnim vremenom regeneracije da bi učinak bio veći od moguće štete. Također, vrijeme za

regeneraciju se često uskraćuje najuspješnijim natjecateljima koji ulaze iz jednog kruga natjecanja u drugi - kao što je slučaj na Svjetskom prvenstvu. U ne malom broju slučajeva, cijena uspjeha ubrzano je metaboličko starenje i pogoršanje zdravlja zbog ozljeda. S obzirom na činjenicu da se vrijednost vrhunskih sportaša mjeri novcem i ugledom, razumljiv je interes za nove metode njihovog pažljivog odabira, prilagođavanje treninga i prehrane. Naše trenutno razumijevanje molekularne genetike u takvoj je fazi da je njezina primjena moguća i primamljiva opcija u vrhunskim sportovima. Pojava CRISPER tehnologije ide korak dalje i omogućuje dizajn još neviđenih ljudskih sposobnosti. Iako je genetska primjena još u povojima, sportske organizacije moraju zauzeti stav o tome koji su testovi i koji zahvati bioetički opravdani u sportskom natjecanju, a koji su apsolutno neprihvatljivi. Veliko je pitanje i – kakva je pravna regulacija istog problema? Slijedi li pravo bioetički utvrđeno stanje? Općenito govoreći, važno je istražiti koji pravni akti su u prvom redu povezani s genetikom, a zatim s kombinacijom genetike i sporta. Također, cilj je ispitati da li relevantni pravni akti zabranjuju bilo kakvu promjenu ljudskog genoma, općenito a zatim u kontekstu sporta. Možemo li se reći da je zakon jedan od glavnih zaustavnih medija i “prepreka” cjelovitim genetskim istraživanjima koja teže transhumanizaciji?

**Ključne riječi:** sportska natjecanja, genomske analize, atletski geni, sportska znanost, pravo, pravna zaštita