VYTAUTAS MAGNUS UNIVERSITY
FACULTY OF ECONOMICS AND MANAGEMENT

SAŠKA, IVANOVA

IMPLEMENTATION OF GENETIC TESTING INTO SPORT INDUSTRY

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Supervisor: Assoc. prof. dr. Renata Legenzova
Defended: Assoc. prof. dr. Rita Bendaravičienė

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Abstract

Identification of future sport talent and enhancement of sport performance of the athletes by optimization of training sessions and reduction of sport/related injuries or illness is of paramount interest for many sport stakeholders. Recent scientific studies in genetics opened the door for sport genetic testing with the promise of predicting individual’s sport predisposition and optimizing training and nutrition regimes. This study aimed to provide baseline of recommendations on how to implement sport genetic testing into sport industry. The study surveyed the athletes and sport support staff to assess the occurrence of the genetic testing in sport and to collect the opinions of the main players in sport. In addition, different direct to consumer companies were analysed, case study of different nations (China and Egypt) and a club (FC Barcelona) was carried out, and lastly cost-efficiency analysis of implementing the gene testing in sport was performed.

The research identified key areas of considerations that have to be taken into account before implementing genetic testing into sport. Moreover, the study proposed guidelines for implementation of genetic testing into sport on different levels, namely sport governing bodies, federations and club’s level.
INTRODUCTION

**Relevance and topicality.** Over the centuries, different strategies and technologies were used in order to improve sport performance. Spectators are hungry for better sport results, unbelievable world records and outstanding sport entertainment. Some of the sport improvement is clearly due to the new technologies independently of the athlete’s capabilities, like for example faster athletic tracks, improved sportswear and equipment (i.e. tech suits in swimming, lighter javelin, etc.). However, major part of sport performance is due to the athlete’s abilities, which are multifactorial and depend on a complex and not fully understood interactions between environmental and genetic factors. Athletes are a mix of sociocultural, psychological, anatomic and physiological factors; this is why physical performance is a combination of inherited genes and the overall environmental circumstances (Davids and Baker, 2007). In past twenty years significant amount of studies confirmed the correlation between numerous genetic loci and sport performance (Barton-Davis et al., 1998; Wang et al., 2004; Ahmetov and Rogozkin, 2009). Latest biological and medical research caught the attention of different sportspersons and their trainers in order to gain unfair advantage in the race to be the best by using so called gene doping; i.e. inserting DNA for the purpose of enhancing athletic performance (Filipp, 2007). Nevertheless, World Anti-Doping Agency (WADA) immediately reacted and banned gene doping already in 2003, and broaden the regulation of gene doping in 2018.

This advance in biomedical research, on the other hand, could be used for genetic testing for sports performance, disease and injury prevention and talent identification. Instead of using prohibited gene doping, sport industry could employ examination of the individual’s genetic information to separate "winners from losers". Several studies suggest that elite athletes have variants of specific genes that gives them advantage over people that do not have them (Woods, 2009; Yang et al., 2009; Ma et al., 2013). Moreover, there are also people with inherited gene mutation that are not common in human population, but can give advantage in sports. For example, Finnish Nordic skier and 1964 Olympic gold medallist Eero Mäntyranta had unusually high amounts of red blood cells, and a boy born with a myostatin dysfunction has larger than normal weight-lifting capacities (Schuelke et al., 2004). In the case of Eero Mäntyranta, genetic testing would actually show that his high levels of red blood cells are not due to doping, but rather due to rare mutation. Several companies have recognized the economic potential of genetic testing in sport and are already offering so called sport genetic testing (i.e. 24Genetics, DNAfit, smgenomics, etc.). Furthermore, although the practise of genetic testing in sport is currently not common, there are examples of genetic information being used by sports clubs and governing bodies to make decisions about an athlete’s capability to perform (Patel and Varley, 2019); and also about athlete’s training, nutrition and recovery regimes. Since the technology is quite new and somewhat controversial, especially from the human rights, legal and
regulatory point of view, there are no specific regulation or guidelines by federations/leagues or clubs on how genetic testing should be used and/or implemented in sport. Considering that individual’s genetic information is a very sensitive information that could be misused, there is an increasing and immediate need to regulate, manage and set the framework on how genetic testing should be properly implemented into sport. Even more so since the use of genetic information is not forbidden (but gene doping is), there are sport gene tests available and more importantly there are first reports of nations, clubs and sportsmen using this technology in order to improve sport performance.

The obvious question that arises here of course is, how to monetize genetic testing in sport and how would it bring money to the sport industry. Clearly if genetic testing in sport will eventually be able to screen for genetically the most suitable individual’s and discriminate between the winners and losers, it will also save a lot of money invested in genetically less suitable athletes. In other words, it will reduce the costs sport industry invests in talent identification and selection. The other possible benefit of genetic testing is in reducing injury risk. Huge amounts of money are being lost on the salaries of injured athletes sitting on the benches, without even taking into consideration all the medical, sponsors and other costs related with it. Moreover, genetic testing could reveal susceptibility to some medical conditions, like for example sickle cell trait; and rare heart condition that could lead to sudden death, particularly for athletes (Quick, 2009; Bland, 2011; Wagner, 2012; Roberts et al., 2017). In this cases, physical examination reports are fine, but there is a ticking bomb in their genes, which might take away all the invested money, years of effort and in extreme cases also lives. Nevertheless, there is a confrontation between protecting health and well-being of the sportsmen, safeguarding the financial costs of the teams and clubs, and potentially discriminating against the athletes by restricting their employment opportunities (Patel & Varley, 2019).

**Research problem** addressed in this paper is “What are the issues of sport genetic testing from different stakeholder’s perspective and what view should be taken regarding the implementation of sport gene testing into sport industry?”

**The object** of the paper is the implementation of sport genetic testing into sport industry.

**The aim** of the paper is to give baseline suggestions on implementing and managing sport genetic testing on governing bodies/federation/league and club’s level. The proposals given in this study would consider all the pros and drawbacks of genetic testing as well as legal and ethical issues associated with it.
To reach the aim of the paper the following objectives were set:

1. To review the literature on sport genetic testing, and identify its players.
2. To discuss the genetic fair play.
3. To collect the opinions of athletes/sportsmen and sport support staff on genetic testing in sport and compare them with already published study on opinions of elite athletes and support staff in relation to genetic testing in elite sport within the UK (Varley et al., 2018).
4. To analyse practices of the companies that offer sport gene testing and already known cases of genetic testing in sport.
5. To assess the frameworks and procedures for genetic testing in sport on sport governing body/federation/league and clubs level, if existing or possible.
6. To analyse the benefits and drawbacks of sport gene testing from clubs and players perspective.
7. To provide baseline frameworks, procedures and recommendations for federations/leagues and clubs on how to implement genetic testing into sport.

The paper is structured in 3 main parts.

The first part will cover the literature review on sport genetic testing, which includes scientific background and definition of genetic testing, types of genetic testing, the current use of genetic testing in sport, the stakeholders involved, the World Anti-Doping Agency (WADA) regulation and position on genetic testing, regulation of genetic testing in general in sport as well as ethical and legal considerations of it. The literature review will also discuss the genetic fair-play in sport.

The second part will focus on current status of genetic testing in sport. First, the occurrence and opinion of athletes and support staff will be collected by online survey, next companies that offer sport genetic testing will be analysed, and case studies of nations and club that already use genetic testing in sport will be done. Finally, predictions of the costs of genetic testing will be done and cost-efficiency analysis of the implementation of the genetic testing in sport will be carried out.

In the third part I will be giving baseline recommendations and plan of action, based on the literature review and field analysis done, how to implement and properly manage genetic testing in sport on the governing body/federation/league and clubs level, covering among others, ethical, financial and regulatory aspects of it.

Research methods. This paper, to accomplish the objectives set, used research methods as literature review, survey conducted on athletes/players and support staff, as well as internet based analysis (of the webpages) of the companies that offer sport genetic testing and personal knowledge
and experience in science and genetic testing. Moreover, case analysis of two nations and of FC
Barcelona club, that already use genetic testing, and cost-efficiency analysis of implementation of
 genetic testing into sport was conducted.

Information sources. The study used peer review published journal articles and books as
well as verified internet sources to gain information related to the topic of the study. In addition, the
survey was conducted on athletes and support stuff in Europe about genetic testing in sport. Official
webpages of the direct to consumer companies that offer sport genetic testing were examined for
collecting the data about their practices.
I. LITERATURE REVIEW ON SPORT GENETIC TESTING

1.1 The concept of sport genetic testing

Human Genome Project in 2003 completed the sequencing of human genome and opened the whole new era in the understanding and better interpretation on how genetic information can explain individual’s characteristics and predisposition to variety of conditions. Since then genetic ancestry, susceptibility to some diseases, responses to drugs or nutrients is widely being studied through genetic testing. There is no consensus on the definition of genetic testing, but in general it involves an analysis of the sequence of human DNA in order to detect specific genetic traits and variations, or susceptibility to different conditions (Patel & Varley, 2019). In healthcare genetic testing already improves medical provisions, patient choice for effective disease treatment and even disease prevention (Gostin & Hodge, 1999). Nowadays genetic tests are easily accessible with many companies offering it directly to the consumer (DTC), and people are becoming increasingly interested in their genetic information for variety of reasons, among them most popular being health, ancestry, nutrition and sport performance.

The advances in medicine, science and technology are helping to understand athletic ability. Collecting and analyzing biometric and biomechanical athlete data is a key feature of sport practice (Osborne, 2017). Increasing demand for excellent sport performance further pushes the boundaries in the search for what makes an athlete elite and the inborn genetic traits might reveal future sport stars (Patel & Varley, 2019). The degree to which genes (genotype) can influence sport performance is still under debate. On the other hand, there is an increasing amount of evidence of association between specific genetic loci and/or gene variations (so called SNP; Single-Nucleotide polymorphism) and favorable athletic performance and injury risk (Bouchard et al., 1992; Bray et al., 2009; Wang et al., 2013; Collins et al., 2015). However, at the moment, there is no enough evidence demonstrating that certain genes can clearly predict athletic success, training responses or injury susceptibility.

Even though there is a lack of predictive research, the potential of genetic testing in sport should not be underestimated, especially since with more elite athletes have their genomes sequenced, more it becomes obvious the existence of predisposition to better sport performance or that elite athletes have higher frequency of some gene variations than general population (Jacob et al., 2018). There are several reports of different clubs, national teams or even nations using genetic testing or intent to use it in the future years (Varley et al., 2018; Jacob et al., 2018). One of them is China, who according to a document posted by the Ministry of Science and Technology of China in 2018 plans to establish a laboratory standard for the “selection of athletes by genetic markers” (South China Morning Post, 2018). China’s athletic hopefuls for the 2022 Winter Olympics in Beijing will have to...
undergo genetic screening for a chance to represent the host nation, and it seems that genes will decide who actually enters the national team (South China Morning Post, 2018). Moreover, some European football clubs have reportedly used genetic testing to evaluate muscle damage risk for individual players, whose muscles are less resistant to inflammation, so that team doctors may develop more effective ways to treat or prevent injuries (South China Morning Post, 2018).

Altogether, genetic testing is already being used in sport and will most probably change social, financial, ethical and economic aspects of it. Therefore, there is an imminent need for frameworks and baseline regulation of genetic testing in sport.

1.1.1 Procedure of genetic testing

The procedure is extremely simple for the customer side as he/she or his/her doctor has to swab the inner side of the cheek with the tip provided by the test. The sample is then shipped to the chosen company and the results are received within few weeks.

In the company/laboratory DNA is extracted from the sample and short, specific sequences called single nucleotide polymorphisms (SNPs; acceptable variances in the DNA) or mutations (any changes in the DNA away from the normal) are being analyzed in order to determine different potential health risks, predispositions, etc. Depending on genetic testing acquired (see section 1.1.2), different parts of DNA or genes will be analyzed. In general and as seen in Figure 1, the customer does not get the list of genes tested but rather the results are displayed as risks/sensitivities (low, medium, high), intolerances (tolerant/intolerant), needs (normal, raised), etc. In some cases, like for example in sport genetic testing or for confirmation of some disease as cystic fibrosis, the customer gets the list of the genes tested and the corresponding variations (SNPs) or mutations.

1.1.2 Types of genetic testing

There are different types of genetic testing available on the market and in healthcare, the most common being the following ones.

- **Ancestral genetic testing.** Here the customer gets the information of his ancestral background or more precisely which ethnicity his/her forefathers were.

- **Potential genetic health risk or so called predictive testing.** This kind of testing identifies possible risks for different diseases, most commonly used in cancer, but also in neurodegenerative diseases like for example Alzheimer’s disease. The most known case of predictive testing is Angelina Jolie’s case, who has a strong family history of breast and ovarian cancers. Mutations in one gene, named BRCA1, are clear predictor of breast and also ovarian cancer. Angelina Jolie tested herself and has a mutation in BRCA1 gene, which is why she decided to undergo double
mastectomy (breast removal) and ovary removal in order to reduce the risk of getting breast or ovarian cancer (Cancer Research UK, 2013).

- **Diagnostic testing.** Diagnostic testing is used when there is a suspicion for a certain genetic condition based on signs and symptoms of an individual. Clear example is cystic fibrosis. Patients first presents symptoms and signs of the disease, which is confirmed by the genetic testing of mutations in an explicit gene, which are direct cause of a disease.

- **Carrier testing.** Normally used if there is a family history of a genetic disorder. For example, cystic fibrosis occurs only if both alleles (we have two copies of each gene in our DNA, one we got from the father and the other one from the mother) are mutated, meaning that if we carry one gene mutated, but the other one is fine, we will not get cystic fibrosis, however, we are a carrier of a disease. The problem arises if we have children with a person, who is also a carrier of cystic fibrosis, since in this case there is a 25% chance that the child will get both faulty genes and thus have a cystic fibrosis.

- **Prenatal and preimplantation testing.** These genetic screenings are used when there is a risk of genetic disease or strong family history of a genetic or chromosomal disorder.

- **Newborn screening.** In the US they automatically screen newborns right after birth for around 30 genetic disorders. If detected, some of them can be treated without causing problems (Genetic Counselors). Similar genetic screening of newborns is not taking place in the Europe, however, there is an EU initiative to harmonize genetic testing across Europe, suggesting that in the near future newborn screening might be implemented (EuroGentest).

- **Forensic testing.** This type of testing uses individual’s DNA for legal purposes, most commonly for identifying crime or catastrophe victims, ruling out or implicating a crime suspect and for establishing biological relationships between people (i.e. paternity).

- **Sports genetic testing.** The sport genetic testing companies promise to provide individual’s response to exercise, fitness, and sport; to help decide genetically fit sports career and to assist with preventive measurements in order to prevent or decrease the injuries. Moreover, customers get information if genetically they are more fit for endurance or power performance, how good is their V(02) maximal, injury risk and recovery speed (Figure 1; DNAfit). The information that is provided varies from company to company and more tests or different genetic traits are added with time.

- **Diet genetic testing - nutrigenomics.** In the diet or nutrigenomics testing the customer gets its optimal diet type and different food and drink sensitivities, intolerances, needs and predispositions. Different companies offer different types of food and drinks testing, one example of such testing could be seen in Figure 1 (DNAFit).
• **DNA testing for skin care - dermagenetics.** Here the testing promises to determine the propensity for collagen production, sun exposure risk, inflammation control, and certain antioxidant needs. The test should help with choosing the right products based on your DNA instead of trying one thing after another with little or no results (FutureDerm, 2014).

![DNAFit example report](https://www.futurederm.com/wp-content/uploads/2014/01/DNAFit-Diet-Optimal-Diet-Type.png)

**Figure 1:** Example of sport and diet genetic testing report.

Source: DNAFit webpage (DNAFit.com)

There are also other types of genetic testing, that are not further specified here (for example hair genetic testing), and in the future human race will probably be able to DNA test ourselves for about everything, nevertheless, the most important and relevant ones in the scope of this MBA thesis were named here. It has to be emphasized that given genetic testing does not tell you with certainty that you will be better at some sport or that you will definitely get a breast or ovarian cancer even though you carry mutations in genes like BRCA1, however, it does indicate that the person with the mutations in BRCA1 gene has 80% more probability of having cancer then the person not having mentioned mutations. Our genes are telling us the probabilities we have for certain things, but are definitely not deciding our future as the other major factor is the environment (our life style, eating habits, alcohol consumption, etc.).
1.1.3 Sport gene heritability

Through history there were/are families with obvious sport predisposition or colloquially said, in which sport runs in their veins as for example the Maldinis, the Gasols, the Williams or the Manings. But what actually runs in those families are heritable athletic genes or traits, with following percentages accountable for the heritability of athletic traits (Genetic Literacy Project, 2015):

- Mix of “slow twitch” and “fast twitch” muscle fibers (basically, whether your muscles are better at endurance or sprinting): about 45% heritable.
- Aerobic fitness: about 40-50% heritable.
- Strength and muscle mass: about 50-60% heritable.
- Height: about 80% heritable.
- Competing in sports, at all: 66% heritable.

Trainability itself might have a genetic factor also. Two people starting from the same fitness level with the same program will most probably never respond the same to the training and one of them will end up being stronger and fitter. Even though the genetic advantage might be small, on the elite sport level this small advantage can distinguish between winning a medal at the Olympics versus watching them at home since you came in 4th at the trials (Life Hacker, 2015).

1.1.4 WADA and gene doping

In the year 2004 two breakthrough papers showed that just by manipulating one gene scientists could create so called “marathon mice” (Wang et al., 2004) and “Schwarzenegger mice” (Barton-Davis et al., 1998; Lee et al., 2004), that outperformed control group in the endurance running (marathon mice) or muscle building (Schwarzenegger mice). Shortly afterwards, scientist received calls from multiple athletes in a search of a doping via their genes (CNN, 2018) – i.e. gene doping. Gene doping is an outgrowth of gene therapy, however, instead of injecting DNA in person’s body for medical purposes (i.e. disease treatment), it is used for enhancing sport performance. According to the list of prohibited substances published by WADA (World anti-doping agency) in 2008, gene doping has been defined as: “nontherapeutic use of cells, genes, genetic elements, or modulation of gene expression, having the capacity to enhance athletic performance” (WADA, 2008). From 2018, the list also banns “gene editing agents designed to alter genome sequences and/or the transcriptional or epigenetic regulation of gene expression” (New Scientist, 2017; WADA, 2017). Nonetheless, the main problem WADA faces at the moment is how to accurately detect gene doping, probably this is why there has not been any single case reported, even though the technology is out there and the scientist have confessed to be approached by the sport industry.
Gene doping is forbidden, but, it has to be stressed out and clarified that genetic testing in sport is not banned by the WADA. Actually, there is no consensus in the sport community if the genetic screening of the athletes should be done. The difference is that in genetic testing the individual’s genetic material that he/she was born with is being read and based on inherited genetic information specific predispositions could be detected (i.e. for different diseases, ancestry, etc.). In the case of gene doping, individual is changing its genetic information in order to improve his/her sport performance. It could be argued that advances in sport genetic testing would increase usage of gene doping by sportsmen who lack so called “sport genes”, however that is out of the scope of this MBA thesis.

1.2 Current use of genetic testing in sport

Genetic testing in general is new method, especially in sport industry, nevertheless, there is reported use of genetic testing in sport. In addition, recent study done in UK revealed that genetic testing does occur in British elite sport, although sporadically (Varley et al., 2018). Altogether this confirms that genetic testing does take place in sport (Table 1).

Table 1: Timeline of genetic testing in sport.

<table>
<thead>
<tr>
<th>Year</th>
<th>Genetic testing</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td>1966-1999</td>
<td>Y-chromosomal testing as a part of sex segregation policy of IAAF and IOC</td>
<td>Foddy and Savulescu, 2011</td>
</tr>
<tr>
<td>2001</td>
<td>Professional Boxing and Martial Arts Board of Victoria considers compulsory genetic screening for APOE4 variant in boxers</td>
<td>Jordan et al., 1997</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Robotham, 2001</td>
</tr>
<tr>
<td>2003</td>
<td>WADA prohibits methods of gene doping</td>
<td></td>
</tr>
<tr>
<td>2005</td>
<td>18 Australian male rugby players were tested for 11 genes;</td>
<td>Dennis, 2005</td>
</tr>
<tr>
<td></td>
<td>The Chicago Bulls attempt genetic testing of free agent,</td>
<td>Litke, 2005</td>
</tr>
<tr>
<td></td>
<td>Eddy Curry, for the purpose of ruling out hypertrophic cardiomyopathy</td>
<td></td>
</tr>
<tr>
<td>2009</td>
<td>23 and Me Company analyzes DNA samples from 100 current and former NFL players</td>
<td>Assael, 2012</td>
</tr>
<tr>
<td></td>
<td>MLB begins using genetic testing with prospective players from the Latin American Countries</td>
<td>Schmidt and Schwarz, 2009</td>
</tr>
<tr>
<td>2010</td>
<td>The National Collegiate Athletic Association implements mandatory sickle-cell trait screening</td>
<td>Zarda, 2012</td>
</tr>
<tr>
<td>2011</td>
<td>An English Premier League soccer team analyzes players' DNA samples at 100 genetic loci</td>
<td>Marsh, 2011</td>
</tr>
<tr>
<td></td>
<td>The National Football League screens for genetic conditions Sickle Cell Trait and G6PD under the 2011 NFL collective bargaining agreement</td>
<td>Siegel and Alvarez, 2010</td>
</tr>
<tr>
<td>2012</td>
<td>English Institute of Sport expresses interest in the integration of genetic technologies to “tailor the training.</td>
<td>Watts, 2012</td>
</tr>
</tbody>
</table>
conditioning, and preparation” of Britain’s Olympic and Paralympic athletes

<table>
<thead>
<tr>
<th>Year</th>
<th>Event Description</th>
<th>Year</th>
<th>Report/Source</th>
</tr>
</thead>
<tbody>
<tr>
<td>2014</td>
<td>Two Barclay’s Premier League soccer teams commission tests of their players’ DNA for 45 variants</td>
<td>2014</td>
<td>Williamson</td>
</tr>
<tr>
<td>2016</td>
<td>FC Barcelona DNA tests their football players</td>
<td>2016</td>
<td>Miller</td>
</tr>
<tr>
<td>2018</td>
<td>Gene-testing company partners with Egyptian Olympic and soccer bodies China to select Winter Olympics athletes by their genes</td>
<td>2018</td>
<td>Holmes</td>
</tr>
<tr>
<td></td>
<td>WADA prohibits gene editing</td>
<td>2018</td>
<td>South China Morning Post</td>
</tr>
</tbody>
</table>

Source: Adopted and updated from Goodlin et al., 2015.

The current reported use of genetic testing in sports are as follows.

1.2.1 Age identity. In the Major Baseball League (MBL) genetic testing was initiated in order to authenticate player’s identity and age, since some prospective players, especially from Latin America, were falsifying their age to be more appealing to the MBL clubs (Hebbar, 2011; Zitelli, 2011). Genetic testing does not verify the age, but it does track the family line by testing the players and their families (Zitelli, 2011). Even though genetic testing can violate privacy and lead to genetic discrimination, MBL defends it as a mean to guard the essence of sport (Schwarz, 2009; Stevens, 2011).

1.2.2 Doping. WADA is the responsible body for the anti-doping testing. With the introduction of the Athlete Biological Passport they are monitoring different biological variants to find potential effects of doping. WADA prohibited gene doping and gene editing and is considering to require athletes to submit a copy of their genetic information in order to detect any changes in the athlete’s DNA (Niiler, 2018). At the moment WADA will have to show that this is necessary to ensure the integrity of sport, however, with the current advances in the gene therapy and also in gene editing (Le Page, 2017), is just a matter of time, legal and ethical boundaries, that will have to be set in near future (personal opinion).

1.2.3 Injury and medical conditions. In the study done by Varley et al., 2018, they found that the majority of the athletes surveyed would like to know if they carry genetic variations associated with sport performance and injury susceptibility. The use of genetic testing could help sport staff to minimize the player’s chance of injury by optimizing individual’s training schedules (Goodlin et al., 2015), and also by protecting their lives/lifestyle. For example, the ApoE4 (variant 4 of the ApoE – protein that helps carry cholesterol and other types of fat in the bloodstream and is also a most prevalent genetic risk factor of Alzheimer’s disease) has been associated with the increased risk of brain damage and dementia in professional boxers (Jordan et al., 1997). There has been discussion of introducing compulsory genetic screening for ApoE4 variant in boxing and martial arts, however, the opinions are split on the subject. Some
boxers would not take the risk of getting or further increasing probability to suffer brain damage, dementia or Alzheimer’s disease; nonetheless, a lot of professional fighters would not believe the test or be willing to drop their career and years of dedication just because of ApoE4 variant (Springer and Maugh, 1997).

In 2005, Chicago Bulls basketball club tried to force Eddy Curry Jr with diagnosed benign arrhythmia to take the DNA test to determine the susceptibility to the rare heart disease that could lead to sudden death, especially in athletes. The player declined to take the test and sued the club on the privacy bases. Before the court ruling, he was traded to the New York Knicks, where he was not required to take the test. The Chicago Bulls stated that their intention was to protect the Curry’s well-being, nevertheless, there were insinuations that the club was on a witch-hunt and just protecting their interests (The New York Times, 2005). A year later, in 2006, the opposite scenario unfortunately took place. American football student player Dale Lloyd II died during a training due to complications associated with the sickle-cell trait (people who inherit one sickle-cell gene and one normal gene have sickle-cell trait), but they usually do not have any of the symptoms of sickle-cell disease and in rare cases have “pain crises” or might undergo sudden death (CDC)). The Lloyd family sued the university and the National Collegiate Athletic Association (NCAA) for their son’s death, accusing them of negligence and failure to warn Dale of being a carrier of a sickle-cell trait and risks associated with it (Quick, 2009). In 2010 NCAA implemented mandatory sickle-cell trait screening for the athletes (Zarda, 2010).

The promise to use genetic testing for sport injury prevention is also increasing. One example is the variation in the collagen alpha chain type 1 gene (COL1A1), the predominant protein component of the ligaments and tendons, which affects the level of its production. People that have the so called TT variant of a gene have increased collagen I expression and thus decreased risk for anterior crucial ligament (ACL) rupture and Achilles tendinopathy (Khoschnau et al., 2008; Ficek et al., 2013; Collins et al., 2010). In addition, there are other genes and their variants associated with other athletic injuries, time of recovery from the injuries and better response to the treatment (Gayagay et al., 1998; Yang et al., 2003; Zanoteli et al., 2003; Maffulli et al., 2013; Pruna et al., 2013). It is too early to measure the effect of genetic testing on reducing the incidence of injuries or inducing behavioral changes that will promote health and/or prevent injury, but clearly there are many genetic variants that provide information about risk for sports-related injuries and performance related conditions. Athletes, coaches, and medical practitioners can use this information to generate personalized training regimens for athletes (Goodlin et al., 2015).
1.2.4 **Talent identification and selection.** In theory, genetic testing could identify genetically favorable athletic traits and predict better sport performance of an individual having those traits. Since talent identification is of extreme importance in sport, genetic information could make it easier, more exact and quantitative. Currently, at least publicly, nobody admitted to use genetic testing for talent identification and selection of players for the squad/team. In 2008 it was reported that at least one English football club contacted scientists for possible screening of players for genetic predisposition to athletic excellence (Scott and Kelso, 2008). Moreover, there are several reports of elite athletes or teams working with DTC companies in order to prepare for different competitions (Watts, 2012; Holmes, 2018). And most recently China decided to select their Winter Olympic Games 2022 in Beijing representatives (also) based on their genetics (South China Morning Post, 2018), which raised a lot of concerns, beside legally and ethically, also scientifically, since at the moment we are not quite there. Over the decades more than 150 different sport related genes and their variants have been identified, but their exact connection, significance and importance for sport performance remains elusive. Sport performance is a polygenic trait (meaning that many different genes contribute to the sport performance and there are very few (if any real at the moment), that could directly and clearly predict sport excellence). Having said that, there have been variants within genes that have been identified as predisposing individuals to elite endurance or elite power performance (Jacob et al., 2018). The ACE and ACTN3 gene variants, for example, among other functions also influence the type of muscle fibers (fast- or slow-twitch muscle fibers) and have been linked to endurance and strength (Myerson et al., 1999; Zanoteli et al., 2003; Yang et al., 2003). Beside athletic traits (power, endurance, and strength), other traits like skill acquisition and development influence success in sport, especially team sports like basketball and football (Jacob et al, 2018). Several genes have been shown to impact on motor coordination and learning of complex skills (Fritsch et al., 2010; Huertas et al., 2012).

1.2.5 **Gender verification.** In some cases, when the testosterone levels in women are high (IAAF hyperandrogenism rule) or the women athlete appearance is suspicious, female athletes have to undergo genetic test to verify their sex. Historically, this rule was implemented to prevent gender fraud and regulate unfair sport advantage (Patel, 2015), nonetheless, it excludes the athletes with inherited genetic traits such as high testosterone levels (Patel and Varley, 2019). Most recent case of high levels of testosterone (3 times more than is expected in women) is Caster Semenya, South African Olympic medalist in middle-distance, who was forced to undergo genetic testing by the IAAF before allowed to compete in 2010 due to her rare condition (presumably she is genetically male, but was born and raised as women with intersex
condition due to the disorder in sex development (DSD) – athlete with DSD; The New York Times, 2019; Savulescu, 2019; Jones 2019).

1.2.6 Nutrition optimization. Nutrition considerably impacts on sport performance and individuals diversely respond to the same food, nutrients and supplements intake, shifting the paradigm towards personalized nutrition in general, and also in sport. Personalized nutrition’s objective in professional sport is to optimize health, body composition, and exercise performance by coupling dietary needs of an athlete to its genetic profile (Guest et al., 2019). Nutrigenetics is a scientific field that studies how different genetic variations influence the response to the nutrients, macro and micronutrients uptake, metabolism and health (Schwartz, 2014). One of the main goals of the nutrigenetics is to figure out how the individual genetic background and underlying genetic polymorphisms control their response to diet (de Roos, 2013; Schwartz, 2014). As seen in Figure 2, the same food, drink, nutrient, or bioactive compound can exert completely divergent response on health or sport performance depending on the gene variant that individual has (SNP; single nucleotide polymorphisms). The most studied example is caffeine, and different studies revealed that inter-individual differences are at least in part due to variations in the gene CYP1A2 (responsible for caffeine metabolism), which determine if someone metabolizes caffeine fast or slow. The fast metabolizers of caffeine have improved response or performance to caffeine, while slow metabolizers have no effect or even impaired performance from caffeine use (Guest et al, 2018; Guest et al., 2019). Moreover, other nutrients like vitamin A, vitamin C, folate, lactose, iron, etc., have been shown to have similar effects on performance as caffeine, i.e. depending on different variants in proteins responsible for its uptake, metabolism, utilization, or excretion (reviewed in Guest et al, 2019). Based on this several DTC companies already offer some kind of nutrigenetic testing in the sport genetic testing (Figure 1), where among others things different nutrient needs and sensitivities are being checked. In addition, body weight, body composition, fuel needs for training and competition (protein, carbohydrates, and/or fat) have also been shown to depend on individual’s genetic variations (reviewed in Guest et al, 2019). Of note, coupling nutrigenetics with standard biochemical blood test are the best way to personalized athlete’s nutrition regime.

Genetic information is growing rapidly, producing new information faster than majority other fields. Sport and exercise genomics is in its early stages, nevertheless, there is considerable amount of ongoing research on individual’s variation in response to exercise, injury susceptibility, genetic prediction of sport talent, etc. (Mann et al., 2014; Pitsiladis et al., 2016; Guest et al, 2019). Probably in the next few years the power of genetic testing to help predict sport talent, likelihood of sports-related injuries, personalized sport nutrition regimes and training optimizations will grow.
exponentially. With advances in machine learning and artificial intelligence, tools, algorithms and apps will be developed that will automatically integrate published data on genetics and sports with given athlete’s genetic information in order to help sport staff to gain competitive edge (personal view – it might sound as a science fiction, but we are definitely already in the genomic era).

**Figure 2:** Nutrigenetics approach to sport nutrition.

Source: adopted from Guest et al., 2019.

**1.3 Stakeholders involved in sport genetic testing**

Genetic testing in sport is new and still developing process, where all the stakeholders are probably not yet fully identified. However, the potential that some interested groups see in it and based on all the literature reviewed, allows me to propose or summarize the stakeholders in sport genetic testing as follows.

**Nations.** The governments are one of the main players as they might support or not the implementation or use of genetic testing in sport. Clear example is China, who went much further than most of the countries and is actually thinking of selecting their Winter Olympic Games representatives in Beijing 2022 based on genes. Of course, if this actually happens, it will have an enormous impact on sport industry, especially if it proves that winners can be chosen by their genetic information. On the contrary, none of the countries, to my knowledge, has prohibited use of genetic testing in sport, however, majority of them are cautious about the subject and against Chinese policy due to the lack (at the moment) of clear prediction of elite athletes based on their genes and ethical issues. Nevertheless, most nations are allowing gene testing as additional tool for better preparation of the sportsmen.

**Public.** The society and especially sport fans are hungry of great sport performances, amazing games or plays and world records breakings. If genetic testing in sport can help in talent identification and better player’s preparation, this would lead to better sport performances and even
more unpredictable results, which would increase the amount of expected viewers and thus the income through sales of tickets, merchandise, hotel accommodations and other expanses generated as a result of the sport event. Having said that, there are no actual studies on how and what society or sport public feels on genetic testing in sport.

**Federations/Leagues.** Similarly as nations, federations/leagues are responsible for approving and giving the frameworks and baseline recommendations for the use of genetic testing in sport. So far and to my knowledge, there was no clear statement about sport gene testing from any of the European’s leagues or federations. Moreover, there are also no frameworks or guidelines on how to properly conduct genetic testing in sport in Europe. One of the few leagues so far that has confirmed the use of genetic testing is the Major League Baseball of United States (MLB) to help clubs corroborate age and identity of the players (The New York Times, 2009; Stevens, 2011). Gender verification is another use of genetic testing that has been conducted in several leagues on "suspicious" female athletes or female athletes with above the normal levels of testosterone.

**Clubs.** At the club’s level is where the gene testing in sport is predominantly being conducted as they are the ones responsible for the sport talent identification, athlete training and nutrition regimes. Several clubs in Europe allegedly already used gene testing for different purposes, among them several football clubs, 2 from Premier League and FC Barcelona (Daily Mail, 2014; The Mirror, 2016). Unfortunately, there is little or no literature on how these specific cases of genetic testing were done, which genes exactly were looked on, who has the access to the individual player’s genetic information, were the players properly informed about the pros and cons of genetic testing.

**Players/athletes.** They are the main stakeholders as the testing is actually done on them. They can benefit or lose from the genetic testing depending on the purpose of it. As already said, if used for talent identification, the athletes that do not have variants of genes looked at might be excluded from persuading sport career. Additionally, probably majority of sportsmen do not understand or are not properly informed about the gene testing and all the consequences that come along with it. There is also no information if the ones who have undertaken genetic testing by their clubs or federations have signed adequate informed consent.

**Genetic testing companies.** They are the ones who test the DNA for different genes and reveal the genetic information of specific athlete. There are several companies offering DTC sport genetic testing (i.e. 23Genetics, DNAfit, smgenomics, etc.). To my knowledge, there is no accredited laboratories for sport genetic testing in Europe nor there is a consensus on which genes to include in sport performance, injury risk or nutrition profile categories.

**Sponsors.** Sponsorship deals in the future might rely also on the genetic information, especially in the individual’s sports.
**Scientific community.** Scientists carry on the scientific research in the field of sport genetics and contribute to the correlation between specific genes and sport performance, injury risk and individual’s nutrition needs.

**Ethical comities.** With the genetic testing in general, as well as sport genetic testing, several ethical questions have been raised around genetic discrimination, privacy and consent in employment and insurance, right to open future, etc. Suitable ethical committees should provide ethical and legal boundaries to genetic testing in sport.

1.4 Regulation of genetic testing in sports

As genetic testing in sports gains momentum, it is important to develop best practices to protect the legal, ethical, and social rights of the athlete. Genetic testing raises concerns about genetic discrimination, privacy and consent in employment and in insurance (Patel and Varley, 2019). Athletes are commonly "discriminated" based on their physiological or phenotype characteristics, sex segregation being one of the best known ones, but also age, different disabilities, and in some sports even weight (boxing for example). All these exclusion rules in sport are seen as necessary and legitimate for fair competition, even though some of them might be incompatible with human rights principles.

Formal regulation of genetic testing in sport seems to be non-existing, most probably due to relatively new technology that is not even properly regulated in the field of medicine and health. There have been few statements by some sport organizations, i.e. British Association of Sport and Exercise Sciences (BASES), which recognizes that genetic research is becoming increasingly important in sport and exercise science, however, research has to be made in an "ethically acceptable manner" (Williams et al., 2012). Another statement was given from the Australian Institute of Sport (AIS), basically saying there are "currently no scientific grounds for the use of genetic testing for athletic performance improvement, sport selection or talent identification" (Vlahovich et al., 2017). However, the AIS also affirms in the same position statement that there are valid roles for genetic research and that they support the genetic research which aims to enhance the understanding of athlete susceptibility to injury or illness (Vlahovich et al., 2017). And recently, Chinese Ministry of Technology and Science publicly admitted that they intent to use genetic testing on their athletes as official athlete selection process for 2022 Winter Olympic Games (South China Morning Post, 2018).

It is obvious that with the growing list of DTC sport genetic testing companies on the market and sport industry interest in the genetic testing, there is an almost immediate need for sport authorities to develop policies and guidelines on the use of genetic information in sport. In order to do that, several aspects have to be considered and are reviewed as follows.
1.4.1 **Legal aspect.** Since there is no clear regulation for genetic testing in sport, hypothetically current sport practices rely on legal standards. Nonetheless, there has always been a complex interconnection between sport and the law. Sport is unique as it helps shape society and cultures, and contributes to economic growth, which is why sport regulations are often under great scrutiny and receive immediate media and public attention (Boyes, 2017; Patel and Varley, 2019). In general, sport is considered as self-regulating private sphere, where line between formal law and normative rules of sport are blurred (Harlow and Rawlings, 2009). Sport governing bodies often see the law as a threat to the essence of the sport, thus favoring internal sports regulation (Patel and Varley, 2019).

- **On the international level** The Universal Declaration on Human Genome and Human Rights (UNESCO, 1997) declares human genome as "heritage of humanity" (Article 1) and tries to balance the advances in the science with the protection of human rights and dignity. Moreover, The Declaration states that everyone is entitled to a "right to respect for their dignity and for their rights regardless of their genetic characteristics" (Article 2) and no one should be subject to discrimination on the basis of those genetic characteristics (Article 6). Consent and confidentiality of genetic information are also included in The Declaration (UNESCO, 1997). However, it has to be said here that The Declaration is providing the guidelines to countries on how to make legislation and their policies on genetic information, but they have no legal accountability. In 2017 WMA (World Medical Association) stated the importance of an informed consent of the individual when genetic testing is being performed (WMA, 2017).

- **On the European Union (EU) level** the regulation of the genetic testing is under The European Convention on Human Rights (ECHR) and Biomedicine Convention (Oviedo Convention, 1997), which is so far the only legally binding document on the protection of human rights in the field of biomedicine (Council of Europe, 2015), aiming at protecting dignity and identity of all human beings and guaranteeing everyone, without discrimination, respect for their integrity and other rights and fundamental freedoms with regard to the application of biology and medicine (Article 1). The convention establishes the right to privacy of information in the health field (Article 10), prohibits any form of discrimination against an individual on grounds of his or her genetic heritage (Article 11) and bans predictive tests for reasons other than health or health-related research, even with the consent of the person concerned; therefore, it is forbidden to do predictive genetic testing as part of pre-employment medical examinations, whenever it does not serve a health purpose of the individual (Article 12). Article 12 would thus exclude the genetic testing for sport performance and talent identification. However, sport has a special treatment when it comes to the law, which actually lead to the birth of a distinct discipline called sports law. Later on, in 2016, Council of Europe recommended member states
to adopt proper measurements to ensure respect for the fundamental rights of persons, without
discrimination, in the context of the insurance contracts (Recommendation CM/Rec(2016)8). In addition, two more EU regulations have impact on the genetic testing in general, The General Data Protection Regulation 2016/679 (GDPR), which protects the processing of personal genetic, biometric or health data (Article 9 of GDPR); and new Regulation on Medical Devices adopted in 2017 (The Medical Device Regulation (MDR) and The In Vitro Diagnostic Medical Device Regulation (IVDR)), that aims to better protect public health and patient safety and include the predictive genetic tests within their scope – which will place obligation on the DTC genetic testing companies to ensure that their procedures are accurate and validated before getting on the market. This might impact sport clubs and teams that use DTC services for the genetic testing of their athletes.

• **On the country/domestic level** majority of the EU states recognizes the EHCR as domestic law. Moreover, many European countries have banned the use of genetic testing in employment and insurance (Van Hoyweghen, 2015). In the US in 2008 the Genetic Information Non-Discrimination Act (GINA) was implemented, that bans genetic discrimination in health insurance and employment.

Taken together, there is an increasing awareness for regulation of genetic testing on all levels. It remains unclear if these measures directly apply to genetic testing in sport, especially since the global nature of sport could put testing outside the jurisdictional reach of any legislation.

### 1.4.2 Ethical aspects

Several ethical dilemmas arise from the use of genetic testing in sport.

• **Discrimination/denied open future.** Current genetic tests are not powerful enough to clearly predict sport performance. Thus, some athletes (especially young ones) might be discriminated based on their genetic information, which is against all the Non-discrimination and Human Rights Acts mentioned in the section 1.4.1. Should sports industry be able to screen the talent, hire and fire based on genetic variations? Should individuals whose genetic data differs from scientifically proposed as the most suitable one for the given sport be excluded from pursuing the sport career?

• **Imposition of genetic testing.** There is a great possibility that genetic testing might be imposed on the athletes, especially on the young ones, and is potentially susceptible to abuse. It is unclear how players could deny consent to genetic testing if asked by their medical staff, coach or franchise/club and how this denial would influence their future. Nevertheless, even if consenting to genetic testing would follow ethically acceptable manner, what steps should follow from different test results, especially since testing itself does not guarantee unequivocal prognosis (McNamee et al., 2009).
• **Insurance of athletes.** Genetic data of athletes might change insurance policies of an athletes, especially the injury and death policies. The question here is, should insurance company have an access or request the athlete’s genetic information.

• **Impact on salary.** Similar as with insurance companies some genetic variations could influence the actual salary of the athletes like for example high injury risk. Let’s hypothesize that Gareth Bale, who has a lot of injuries in his career, does genetic testing and it predicts high injury risk, this information would most probably decrease his transfer fee and also salary in Real Madrid and the actual decision to buy him.

• **Confidentiality and data protection.** Confidentiality is one of the big concerns, especially who can have access to the genetic information of the athletes, which genes can be looked at, to whom could this information be revealed, etc. Another problem is also data protection or how to properly protect the genetic information from being disclosed, how to properly guard/save it and protect it from possible genetic espionage.

• **Social impact.** Some genetic testing results might reveal predispositions to different diseases; if and how this is communicated to a player might impact not only his sport career but his life in general. For example, predisposition to Alzheimer disease might provoke anxiety, stress, change in a life style, etc., this is why any genetic testing result should be given by properly trained staff and in parallel with genetic counselling.

1.4.3 **Genetic fair-play.** One of the key elements in sport is fair-play and prevention of unfair advantage of an athlete or a team in a competition. In order to ensure fairness in sport, different segregations during the history have been made, sex separation being one of the most evident ones. In order to ensure "fairness", sport governing bodies have introduced other measures like The Athlete Biological Passport (WADA) and all year around whereabouts of the athletes (anti-doping measures). In addition, financial fair-play in some competition has been implemented. With growing evidence that some athletes are genetically "better equipped" than others, like for example Eero Mäntyranta, that naturally had more red blood cells (The Science of Sport, 2013), that gave him probably "unfair" advantage over the others; and most recent case of Caster Semenya, that naturally produces more testosterone than allowed values for women in sport. In her case it is considered unfair advantage and she was advised to use medication to lower testosterone levels (The New York Times, 2019; Savulescu, 2019; Jones 2019). Most probably genetic testing will reveal more cases like this, and the question that arises is, should sport force these individual’s to somehow abolish this genetic advantage they were born with in the name of fair-play, or should sport governing bodies be fair to the athletes that have this genetic variants? Third option would be to segregate athletes based on their genetic, however, at least at the moment, this seems unfeasible.
1.5 Economic impact of genetic testing in sport

Genetic testing in sport might have economic impact on different sport areas. In my opinion the following processes/areas in sport would definitely be affected by sport genetic testing.

- **Reducing costs of talent identification and selection.** Talent identification and selection is a costly process, which not always gives the expected results. Talent identification is a prediction of future sport performance based on current physical, technical, tactical and psychological qualities. Different techniques and prediction methods exist, however, none of them is 100% accurate. Genetic testing for sport performance gene variants might be an additional tool, however, it should not be used as the only or predominant one, in the process of helping to identify future sport talent. If genetic testing could improve the spotting of high-potential athletes by only few percent, this would also directly reduce the costs of talent identification process.

- **Reducing costs of injuries.** Costs of injured players include their salary for the time they are not playing, treatment and rehabilitation of the injury, adaptation to trainings after the injury and in some cases club has to consider buying another player to fulfill missing position. There is a lot of money being lost in sport yearly on injuries. Genetic testing for injury susceptibility might help reduce risk for injuries by modifying training regimes and taking precautions according to genetic profile, which would directly reduce the money lost on injured players.

- **Optimizing nutrition of athletes.** Nutrition of the players can clearly influence their physical performance. Personalized nutrition would optimize player’s dietary and supplemental strategies based on genetic data, thus improving athletic performance. Better athletic performance or winning is positively correlated with revenues (Bradbury, 2016). In addition, disclosure of genetic information enhances provided dietary recommendations (El-Sohemy, 2017; Guest et al., 2019).

- **Avoiding being sued due to negligence.** There have been several cases in the US, where families have sued different universities or sport governing bodies due to negligence to inform a player of life threatening condition (as for example Lloyd case mentioned in the section 1.2.3), some of them could be screened by genetic testing.

- **Employment or outsourcing for genetic testing and counseling.** Sport club or in the case of China the nation, that wants to conduct genetic screening of its athletes will have to invest the money first in the genetic screening per se, the adequate personnel to conduct the collection of the samples, interpretation of the given results, implementation of the results into training, nutrition and other regimes, and genetic counseling to the athletes.

- **Sponsors.** Most probably if genetic profiles become publicly available or at least some parts of it or if some athletes want to reveal their genetic data, sponsorship deals might favor the more "genetically fit" athletes. Like for the insurance companies, there is an ethical dilemma, if they can directly ask for genetic information.
• **Public and spectators.** As mentioned in the section 1.3, public and spectators want to see always better performances, outstanding results and unpredictable games. Genetic testing might help improve talent identification, reduce injuries, optimize the nutrition, etc. of the athletes, which would directly impact the sport performance and increase the attendance to the games and all the others revenues related to fans and spectators of the games/matches/races.

To summarize the first part, different types of genetic testing were introduced as well as sport gene heritability and WADA view on the gene doping and genetic testing were explained. Moreover, current use of genetic testing in sport was reviewed, namely for what kind of testing it is being used, stakeholders in sport genetic testing were identified, and regulation of genetic testing in sport was reviewed, especially legal and ethical aspects of it. And lastly, different economic impact of genetic testing were found, as for example reducing costs of injuries.
II. ANALYSIS OF CURRENT STATUS OF GENETIC TESTING IN SPORT

2.1 Research design

**Background.** Some countries, leagues and clubs have expressed interest or even conducted some type of sport genetic testing, however, exact frameworks, protocols and guidelines followed are vague. The potential of genetic testing in sport appears to be huge and promising in terms of helping identify sport talent, decrease the risk of injuries, personalized training and nutrition regimes, etc.; which might lead to great sport performance, better prepared and less injured players, probably more equal and unpredictable games, and finally more revenues. Legal boundaries and frameworks are currently under construction even in the health care system, where genetic testing is more common and accepted. Nevertheless, sport has a unique role in global economic growth and social life worldwide, giving it the power to be at least in part a private self-regulating sphere that sets its own internal regulations.

Sport genetic testing is in its beginnings, with a lot of unresolved issues, especially ethical ones, but also practical as at the moment it is not clear how it is being done, which protocols are being followed, how it will change the sport itself, and which actual benefits and drawbacks it will have.

**Goal of the field analysis.** The objective of the field analysis is to assess the current status of genetic testing in sport, how leagues/clubs conduct genetic testing in the absence of clear regulation, which are the problems encountered and bottlenecks that need thorough consideration before genetic testing is widely implemented in sport and before frameworks, guidelines and protocols are set by sport governing bodies.

**Research method/tools.** The study first assesses how common genetic testing is in sport by doing a survey on athletes and support staff on the current use, opinion and how they envision genetic testing in sport. Survey of this study was compared to the survey done on the elite sport in the UK (Varley et al., 2018). The qualitative aspect relied on previous literature and reports by different clubs/leagues/nations and DTC companies on the protocols they used for the genetic testing of athletes. And lastly, the economic impact of genetic testing in sport is assessed by cost-efficiency analysis of implementing genetic testing into sport.

**Research scope.** The study examined the opinions of the elite athletes and support staff in Europe on current status and their opinions on the genetic testing in sport, and ethical considerations related with it. Furthermore, known worldwide sport genetic testing companies and known nations (Egypt, China) and FC Barcelona’s practices were scrutinized in order to identify possible problems and considerations that have to be taken into account before proposing how to implement the genetic testing into sport industry. Possible benefits and drawbacks of implementation will be assessed by
cost-efficiency analysis of implementing genetic testing in sport. The limitations of this study are the relatively low number of elite sport and support staff respondents to the survey and the difficulties of contacting them (several people at the positions to forward the survey denied to do it). Furthermore, Ricard Pruna (medical doctor conducting genetic testing in FC Barcelona) never replied to my mails, neither did the CEO of Made of Genes/Genomcore nor the sport genetic company S.M.Genomics. In addition, Ignacio Muro, doctor at EuroLeague, denied to share the statistics of injuries of the players in the Turkish Airlines EuroLeague. The general limitation of this study is the scarcity of the data available by the nations and clubs that are already doing genetic testing of their athletes, and the actual benefits and drawbacks they see on different sport areas due to genetic testing implementation.

2.2 Survey on athletes and sport support staff on genetic testing

Ethical, legal and social concern have to be taken into account when implementing genetic testing into sport (Camporesi et al., 2016; Loland, 2015; Varley et al., 2018). There is a lot of discussion going on about the implications of sport genetic testing, however, with no specific guidance to follow, there is a lot of ambiguity and confusion. Thus, the aim of this survey was to assess the current use and to collect the opinions and beliefs about the genetic testing in sport of elite athletes and support staff in Europe.

- **Materials and methods.** Athletes and support staff of Europe clubs, national selections of different sports, and governing bodies were contacted via mail and by word of mouth via personal and professional contacts, especially the ones I made by working as a volunteer at last five Turkish Airlines EuroLeague Basketball Final 4, several swimming and water polo elite level competitions (including World Championship in Swimming 2013, European Championship in Water polo 2018) and UEFA Championship Final 2019. Participants were asked to complete an anonymous and internet based survey, which had 16 multiple choice questions (Table 2 and 3) and were adopted, upgraded and extended from the published study on the current use and opinion of elite sport on genetic testing in UK only (Varley et al., 2018). Athletes were classified as professional if they regularly competed on national or international level, and sport staff that works with professional athletes met the inclusion criteria. The completion of the survey by the athlete or support staff was taken as a consent to be a part of the study. Completed surveys were analyzed by frequency based percentage to a given question.

- **Results and discussion.** In total 34 athletes (32 males and 2 females) and 22 support staff (19 male and 3 female) completed the survey between December 3, 2019, and January 9, 2020. Athletes were from following sports: basketball (18), football (2), cycling (2), kickboxing (2), modern pentathlon (1), water polo (8) and athletics (1); from the following countries: Austria (3), Germany (1), Greece (2), Hungary (2), Italy (2), Lithuania (3), Monaco & France (1), Montenegro
(5), Poland (3), Serbia (1), Slovenia (3), Spain (4) and Turkey (4). Support staff was from
following sports: basketball (17), handball (1), football (1), and water polo (3); and from
following countries: Greece (5), Croatia (1), Italy (2), Lithuania (4), Monaco & France (2),
Montenegro (2), Serbia (1), Spain (3), Turkey (1) and Russia (1). Table 2 and 3 reflects the
responses to the questions of the athletes and support staff.

Table 2: Responses from the athletes to the survey about the genetic testing in sport.

<table>
<thead>
<tr>
<th>Athletes/players (n=34)</th>
<th>Yes</th>
<th>No</th>
<th>Unsure</th>
<th>Already is</th>
<th>N.R.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Current use</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Have you ever heard of genetic testing in sport?</td>
<td>24,5 %</td>
<td>47,1 %</td>
<td>24,5 %</td>
<td>n.a.</td>
<td>0 %</td>
</tr>
<tr>
<td>Have you undergone a genetic testing in relation to sport performance?</td>
<td>20,6 %</td>
<td>64,7 %</td>
<td>14,7 %</td>
<td>n.a.</td>
<td>0 %</td>
</tr>
<tr>
<td>Have you undergone genetic testing in relation to injury susceptibility?</td>
<td>17,6 %</td>
<td>64,7 %</td>
<td>14,7 %</td>
<td>n.a.</td>
<td>2,9 %</td>
</tr>
<tr>
<td>Have you undergone genetic testing in relation to nutrition optimization?</td>
<td>23,5 %</td>
<td>61,8 %</td>
<td>14,7 %</td>
<td>n.a.</td>
<td>0 %</td>
</tr>
<tr>
<td>If you have undergone genetic testing, do you recall which genes were looked at?</td>
<td>8,8 %</td>
<td>Informed, can’t remember</td>
<td>50,5 %</td>
<td>n.a.</td>
<td>23,5 %</td>
</tr>
<tr>
<td><strong>Beliefs</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Do you think genetic testing will be part of sport in future?</td>
<td>44,1 %</td>
<td>5,9 %</td>
<td>44,1 %</td>
<td>5,9 %</td>
<td>0 %</td>
</tr>
<tr>
<td>Sport performance is due to?</td>
<td>Genes</td>
<td>Training</td>
<td>Genes&amp;Training</td>
<td>n.a.</td>
<td>0 %</td>
</tr>
<tr>
<td><strong>Potential for the use</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Would you take a genetic test to gain information about sport performance if asked by coach/governing body?</td>
<td>61,3 %</td>
<td>8,8 %</td>
<td>29,4 %</td>
<td>n.a.</td>
<td>0 %</td>
</tr>
<tr>
<td>Would you take a genetic test to gain information about injury susceptibility if asked by coach/governing body?</td>
<td>64,7 %</td>
<td>5,9 %</td>
<td>29,4 %</td>
<td>n.a.</td>
<td>0 %</td>
</tr>
<tr>
<td>Would you take a genetic test to gain information about nutrition optimization if asked by coach/governing body?</td>
<td>67,6 %</td>
<td>2,9 %</td>
<td>26,5 %</td>
<td>n.a.</td>
<td>2,9 %</td>
</tr>
<tr>
<td>Would you want to know if you had a genetic variation associated with sport performance?</td>
<td>73,5 %</td>
<td>8,8 %</td>
<td>17,6 %</td>
<td>n.a.</td>
<td>0 %</td>
</tr>
<tr>
<td>Would you want to know if you had a genetic variation associated with injury susceptibility?</td>
<td>64,7 %</td>
<td>11,8 %</td>
<td>20,6 %</td>
<td>n.a.</td>
<td>2,9 %</td>
</tr>
</tbody>
</table>
Should genetic information help determine selection/employment in your sport? | 35.3% | 20.6% | 44.1% | n.a. | 0% \\
Would genetic information be a valuable tool for talent identification in your sport? | 50% | 5.9% | 44.1% | n.a. | 0% \\
Genetic information could reveal medical conditions and predisposition to some diseases. Would you be comfortable with your club/governing body having this information about you? | 29.4% | 23.5% | 47.1% | n.a. | 0% \\
Do you think genetic testing should be tightly regulated by governing bodies? | 20.6% | 23.5% | 55.9% | n.a. | 0% \\
N.R., no response, n.a., not applicable

Source: Survey made on athletes by the student.

Table 3: Responses from the support staff to the survey about the genetic testing in sport.

<table>
<thead>
<tr>
<th>Support staff (n=22)</th>
<th>Yes</th>
<th>No</th>
<th>Unsure</th>
<th>Already is</th>
<th>N.R.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Current use</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Have you ever heard of genetic testing in sport?</td>
<td>31.8%</td>
<td>59.1%</td>
<td>9.1%</td>
<td>n.a.</td>
<td>0%</td>
</tr>
<tr>
<td>Has your club/organization ever done a genetic testing of its athletes/players in relation to sport performance?</td>
<td>18.2%</td>
<td>68.1%</td>
<td>13.2%</td>
<td>n.a.</td>
<td>0%</td>
</tr>
<tr>
<td>Has your club/organization ever done a genetic testing of its athletes/players in relation to injury susceptibility?</td>
<td>18.2%</td>
<td>58.6%</td>
<td>63.6%</td>
<td>n.a.</td>
<td>0%</td>
</tr>
<tr>
<td>Has your club/organization ever done genetic testing of its athletes/players in relation to nutrition optimization?</td>
<td>22.7%</td>
<td>54.4%</td>
<td>22.7%</td>
<td>n.a.</td>
<td>0%</td>
</tr>
<tr>
<td>If your club/organization has done genetic testing, do you recall which genes were looked at?</td>
<td>4.5%</td>
<td>Informed, can’t remember 18.2%</td>
<td>Not informed 54.5%</td>
<td>n.a.</td>
<td>22.7%</td>
</tr>
<tr>
<td><strong>Beliefs</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Do you think genetic testing will be part of sport in future?</td>
<td>45.5%</td>
<td>9.1%</td>
<td>36.4%</td>
<td>9.1%</td>
<td>0%</td>
</tr>
<tr>
<td>Sport performance is due to?</td>
<td>Genes 0%</td>
<td>Training 9.1%</td>
<td>Genes&amp;Training 90.9%</td>
<td>n.a.</td>
<td>0%</td>
</tr>
<tr>
<td><strong>Potential for the use</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Do you think there is a place for genetic testing in relation to sport performance in your sport?</td>
<td>50%</td>
<td>13.6%</td>
<td>31.8%</td>
<td>n.a.</td>
<td>4.5%</td>
</tr>
</tbody>
</table>
Do you think there is a place for genetic testing in relation to injury susceptibility in your sport? 59,1 % 18,2 % 22,7 % n.a. 0 %

Do you think there is a place for genetic testing in relation to nutrition optimization in your sport? 59,1 % 18,2 % 22,7 % n.a. 0 %

Would you want to know if an athlete/player had a genetic variation associated with sport performance? 77,3 % 0 % 22,7 % n.a. 0 %

Would you want to know if an athlete/player had a genetic variation associated with injury susceptibility? 81,8 % 0 % 18,2 % n.a. 0 %

Should genetic information help determine selection/employment in your sport? 50 % 18,2 % 27,3 % n.a. 4,5 %

Would genetic information be a valuable tool for talent identification in your sport? 54,5 % 0 % 40,9 % n.a. 4,5 %

Genetic information could reveal medical conditions and predisposition to some diseases. 54,5 % 13,6 % 31,8 % n.a. 0 %

Do you think your club/governing body should have this information about their athletes/players? 54,5 % 13,6 % 31,8 % n.a. 0 %

Do you think genetic testing should be tightly regulated by governing bodies? 40,9 % 4,5 % 54,5 % n.a. 0 %

N.R., no response, n.a., not applicable

Source: Survey made on sport support staff by the student.

**Current use** of genetic testing in sport in Europe is according to the survey (Table 2) about 20 % for the sport performance, injury susceptibility and nutrition optimization, clearly indicating that genetic testing is already happening in sport. These numbers are higher than in previously reported study (Varley et al., 2018), most probably the main reasons being the time that has passed between the two surveys (approximately 4 years) and that in the first study only UK athletes were included. This would suggest that with time the use of genetic testing is increasing and that in the future years it will probably become even more common practice in sport, especially with the price of genetic testing dropping and with the science advancing in determining the genetic traits that might influence sport performance. In addition, genetic testing is getting more accepted and appreciated in general public, mainly due to advances in medicine and healthcare, and presumably the same might be happening in sport. Similarly, around 20 % of the support staff also admitted their clubs/sport organizations have used genetic testing in sport. Survey also showed that ~60%
of support staff and 47.1% of athletes never heard about genetic testing at all, indicating that there should be some genetic education and counseling involved in sport. In line with this conclusion only 8.8% of athletes could recall the genes that were looked for, 17.6% were informed but cannot remember and 50.5% were not informed (Table 2). Taken together, it could be concluded that genetic testing in sport is already happening in Europe, and that with years its use will only increase.

**Beliefs of athletes and support staff about genetic testing.** Slightly less than 50% of both athletes (44.1%) and support staff (45.5%) thinks that genetic testing will be part of sport in future, around 40% is unsure and less than 10% (5.9% of athletes and 9.1% of support staff) believes that there is no place for genetic testing in sport in the future, and approximately the same percentage stated that genetic testing already is a part of sport (Tables 2 and 3). Vast majority of athletes (~76.5%) and support staff (~90%) surveyed believes that sport performance is due to combination of both genes and training, perfectly aligning with the beliefs of the UK survey, 79 and 97%, respectively (Varley et al., 2018). When athletes were asked if they would undergo genetic testing if asked by their coach or governing body, about two thirds of surveyed players would do it, and less than 10% would deny taking the test (Table 2). These percentages are slightly higher than the ones reported in Varley et al. (2018), indicating that athletes are becoming more willing to undertake the genetic testing if asked. In addition, more than 50% of surveyed support staff reported there is a place for genetic testing in sport (Table 3). Moreover, athletes would like to know if they have a genetic variation associated with sport performance (73.5%) and injury susceptibility (64.7%). Altogether, these results indicate that athletes and support staff in general believe there is a place for genetic testing in sport; and more importantly, vast majority of athletes would undertake it if asked by coaches or governing bodies.

**Ethical considerations.** Surprisingly, when asked should genetic testing help determine selection/employment in sport, ~20% of respondents think it should not (20.6% of athletes and 19% of support staff). These results are much lower than ones previously reported (34% for athletes and 61% for support staff; Varley et al., 2018), suggesting that athletes and support staff do not see major ethical concern about using genetic information for employment purposes or in other words, they do not see it as genetic discrimination. The most probable reason for this being that athletes are used to be "discriminated" based on their physical and health features as for example height in basketball. The difference with values reported by Varley et al. (2018) might be due to the fact that genetic testing per se has gained respect and acceptance in the last 4 years. Furthermore, majority thinks that genetic testing would be a valuable tool for talent identification, and only 23.5% of athletes would be uncomfortable with club/governing bodies knowing their genetic health conditions and predispositions to some diseases (Table 2 and 3). Lastly, there are
mixed opinions if genetic testing should be regulated by governing sport bodies. With support staff being more inclined for implementing regulations than athletes (Table 2 and 3). To summarize, athletes and support staff surveyed are less concerned about the ethical issues regarding sport genetic testing than expected and previously published (Varley et al., 2018).

- **Conclusions drawn from the survey on the athletes and support staff in Europe.** To my knowledge this is the second study (first being the one published by Varley et al., 2018) on the current use and opinions of the athletes and support staff on genetic testing in sport. Even though relatively small number of athletes and support staff responded to the survey, most probably represents the actual status of the genetic testing in sport in Europe. The main reason for this assumption are: (i) majority of the findings correlate with the one published before (Varley et al., 2018); (ii) one of the main differences found with Varley et al. (2018) is increase in the actual use of genetic testing in sport, from 10% among athletes up to 20%, which would be an expected growth in 4 years’ time between the two studies; (iii) in present study athletes and support staff is less concerned about ethical issues, which is also expected as at the beginning any new technology is taken cautiously and with time things get more relaxed. In conclusion, genetic testing does take place in sport in Europe and majority of the respondents think that genetics do play a role in sport performance, are willing to undertake genetic testing and are not as concerned about the ethical issues as previously reported (Varley et al., 2018).

### 2.3 Analysis of practices of genetic testing in sport

Survey done in this study and by Varley et al. (2018) clearly show that genetic testing is present in sport, and is gradually increasing with time. Several press reports further confirm presence of genetic testing in sport (The New York Times, 2005; The New York Times, 2009; The Mirror, 2016; South China Morning Press, 2018). Thus, the current practices and protocols of genetic testing in sport were examined.

#### 2.3.1 DTC companies offering sport genetic testing

During the last two decades several commercially available genetic tests have been put on the market, among them also the ones related to sport performance or exercise performance, injury risk and nutrition. In the Table 4 companies from all over the world are included, not just from the Europe, as samples can nowadays be easily sent through mail to whatever country. DTC companies offer customer friendly mail order of the testing, which consist of receiving a DNA kit by mail, following the instructions of the kit on how to obtain your DNA sample (usually scrubbing the inner side of the mouth), sending the kit back to the DTC company to process the sample normally in 2-6
weeks (depending on the testing requested) and afterwards the customer receives his/her report through e-mail.

DTC sport genetic testing varies from company to company in the number of the gene variants tested, costs and how the results are presented (Table 4). Companies that test only one or just few variants should not claim that they are doing sport genetic testing. On average, customer hardly understands all the genes and variants included in the report given, even though some companies provide some explanation on why exactly those genes were tested, but in general descriptive results are given as for example "you have a moderate ability to develop your muscle power" or "strength training is less beneficial to people with your genotype, as you are likely to gain fat mass; moderate training is recommended" (24Genetics). With results presented in such a way, it is almost impossible to use this information in order to increase sport performance or to personalize training habits. This is why some of the DTC companies offer genetic counselling in their premium packages and also training recommendations by previous athletes, trainers, sport nutrition experts and sport science advisor (like for example DNA fit or Genetic Performance companies, however, the price of these packages increase and can be up to 609€ (GP Elite Performance Pack, which includes two meetings with a sports nutritionist and two meetings with a sports science advisor)).

Table 4: Companies providing DTC sport genetic testing

<table>
<thead>
<tr>
<th>Company</th>
<th>Website</th>
<th>Traits tested</th>
<th>Cost</th>
</tr>
</thead>
<tbody>
<tr>
<td>Asper Biotech</td>
<td>asperbio.com</td>
<td>2</td>
<td>80€</td>
</tr>
<tr>
<td>Athletigen</td>
<td>athletigen.com</td>
<td>30, 25, 40</td>
<td>Nutrition report – 210$</td>
</tr>
<tr>
<td>DNA fit</td>
<td>dnafit.com</td>
<td>44, 26, WES</td>
<td>Health Fit – 199€, Diet Fit – 149€, Circle Premium – 499€</td>
</tr>
<tr>
<td>GenePlanet</td>
<td>geneplanet.com</td>
<td>29, 58</td>
<td>Nutrifit – 249€, Nutrifit Premium – 399€</td>
</tr>
<tr>
<td>Genetic Performance</td>
<td>geneticperformance.com</td>
<td>4, 10</td>
<td>GP Foundation Starter – 99€, GP Pursuit Plan – 199€</td>
</tr>
<tr>
<td>Gknowmix</td>
<td>gknowmix.com</td>
<td>5</td>
<td>Sport Injury Gene Screen – n.s.</td>
</tr>
<tr>
<td>My DNApedia</td>
<td>mydnapedia.com</td>
<td>10, 8</td>
<td>MyDNApedia Fitness – 99€, MyDNApedia Nutrition – 99€</td>
</tr>
<tr>
<td>Molecular Testing Labs Fitness</td>
<td>mtlfitness.com</td>
<td>n.s.</td>
<td>Molecular Fitness – 249$</td>
</tr>
<tr>
<td>MyRISQ</td>
<td>myrisq.com</td>
<td>8</td>
<td>Sport Injury Risq – n.s.</td>
</tr>
<tr>
<td>Nutragene</td>
<td>nutragene.com</td>
<td>n.s.</td>
<td>n.s.</td>
</tr>
<tr>
<td>Pathway Genomics</td>
<td>pathaw.com</td>
<td>23, 20, 41</td>
<td>Fit iQ – 129$, Sport iQ – 99$, Pathway FIT – 299$</td>
</tr>
<tr>
<td>S.M.genomics</td>
<td>smgenomics.com</td>
<td>13</td>
<td>Sports Injury Genetic test – n.s.</td>
</tr>
<tr>
<td>Company</td>
<td>Website</td>
<td>Price</td>
<td>Services</td>
</tr>
<tr>
<td>------------------------</td>
<td>----------------------------------</td>
<td>----------------</td>
<td>-----------------------------------------------</td>
</tr>
<tr>
<td>Sports Gene</td>
<td>sportsgene.ee</td>
<td>n.s. 8</td>
<td>Athletic abilities – 95€</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Weight Management – 95€</td>
</tr>
<tr>
<td>ThemakingsofMe</td>
<td>themakingsofme.com</td>
<td>n.s.</td>
<td>Speed vs. Endurance – 79$</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Response to Diet – 79$</td>
</tr>
<tr>
<td>23andMe</td>
<td>23andme.com</td>
<td>1</td>
<td>Health and Ancestry – 199$</td>
</tr>
<tr>
<td>24Genetics</td>
<td>24genetics.com</td>
<td>&gt;100</td>
<td>DNA Sport test – 199€</td>
</tr>
<tr>
<td></td>
<td></td>
<td>&gt;100</td>
<td>Nutrigenetics test – 199€</td>
</tr>
<tr>
<td>Dante Labs</td>
<td>dantelabs.com</td>
<td>whole genome</td>
<td>Premium Whole Genome Seq. – 599€</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Whole Genome for Advanced Analysis – 849€</td>
</tr>
<tr>
<td>Veritas Genetics</td>
<td>veritasgenetics.com</td>
<td>whole genome</td>
<td>My Genome – 599$</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

n.s., not specified; WES, whole exome seq.; Seq., sequencing

Source: Websites of the DTC companies listed in the table.

Some of the genetic testing companies are cautious and try not to exaggerate the sport, injury or nutrition genetic testing beyond the scientifically proven findings, however, this is not true for each and every company, especially for the ones that make claims on single or few genetic markers. Thus, before making decisions about personalized nutrition, exercise and sport participation; customers should receive some genetic counselling to put genetic information into proper context, including limitations and its usefulness (Williams et al., 2016). In professional sport, educated nutritionists, geneticists, medical doctors should be involved in the transferring obtained genetic information into actual practice.

In addition, when choosing a DTC company several things beyond the costs should be taken into consideration:

- Genetic variants that are being assessed as there is literally millions of different variants that can be tested. Companies that do not give information on genetic variants tested most probably test genetic variants that are not scientifically well documented to have a role in sport performance, injury susceptibility or nutrition optimization. The argument that it is commercial secret should not be admitted.

- How the results of the genetic test are being interpreted and presented.

- Data protection – how customers personal data and genetic data is being protected. Majority of companies state that they process data in compliance with national or regional data protection laws. Also it should be carefully checked which third parties might have access to the data.

- Genetic testing companies should comply high level quality standards and have appropriate certificates.
• What happens to the DNA sample after processing and receiving of the results? Not all companies state what they do with the DNA and definitely not all companies destroy the sample. For example, DNAfit destroys the sample, while Gene Planet has it stored and it is destroyed just if the customers specifically request it (information found on the companies’ webpages, Table 4).

• References of the genetic testing company should be checked like satisfaction of the clients and in the case of genetic testing in sport, if the company has previous experience with professional athletes. For example DNAfit has a partnership with the Egyptian Football Association and works with several Premier League clubs (SportsPro, 2018).

The price of genetic testing itself is constantly decreasing, and even more important, the cost of whole genome sequencing (WGS) has dropped significantly in the last decade from approximately 10,000 $ in 2011 to around 1,000 $ in 2019 (National Human Genome Institute, 2019) and some companies in the US are offering it already for 599$ (Table 4); and the price will definitely decrease even more in coming years, some predictions say that will drop to astonishing 100$ (Forbes, 2017).

Why is this important? Genetic testing companies normally sequence just the part of the DNA, the part that encompasses the information around the variation of the genes of interest, in general for the testing that these companies do they read just about few % of the human DNA. Whole genome sequencing reads the whole human genome (our whole genetic information), meaning that in the near future we will have our whole genetic information in the digital format. With science advancing in which gene variants have to do with sport performance, injury risks, etc., customer will not have to send the DNA sample again to the genetic testing company and pay again for sequencing of the newly found variants, but instead genetic information of the whole genome sequencing (which has to be done just once) will be checked. Of course, for that a good bioinformatician or big data analyst would be needed, or by then algorithms and apps will be available, where we will just upload the whole genome sequence and the app itself would find and read the gene we are interested in. In sport this would mean sequencing the genome of the athletes and have it stored safely in special databases, and when the new genetic variants associated with sport performance are identified, the genetic data would be retrieved from database and examined if the athletes have the newly identified genetic variants.

2.3.2 Case studies of nations, leagues and clubs using genetic testing in sport

Use of genetic testing is already part of sport, however, the reports on who and how it is using it are scarce and vague. Having in mind that it is new technology, sport industry on all levels is cautious and not very keen on talking about it. Especially since the science itself is quite ambiguous on how it should be properly used (at the moment), and with no clear regulation from sport governing bodies and several important ethical questions raised, it is obvious that nobody wants to get in the
spotlight. In the last decade there were few reports of nations, leagues or clubs that have admitted using or banning the genetic testing. In this study few of them were looked at more in detail, even though for the most of the cases the majority of the data were not available.

- **Egypt.** Egypt Football Association (EFA) and Egyptian Olympic Committee (EOC) have signed the deal with the UK genetic testing company DNAFit (SportsPro, 2018; Sport Industry Group, 2018). EFA used the genetic testing to prepare for the FIFA World Cup in Russia 2018, and they incorporated DNA testing into its players’ programmes to improve and maintain performance (DNAFit, 2017). Furthermore, EFA stated: "The DNAFit test results can be harnessed to optimize dietary and training programmes as well as assess a player’s risk to injury. An independent clinical study (Jones et al., 2016) showed that sports people whose training programmes are matched to their genetic make-up can achieve an athletic performance that is improved almost three times more than those on mismatched training programmes." DNAFit analyzed Egypt’s players abroad at their clubs, and the rest of the team was tested in Cairo by DNAFit’s local company, DNAFit Egypt (DNAFit, 2017). EOC will use genetic testing to prepare for the Olympic Games in Tokyo 2020 (Sport Industry Group, 2018). Mr. Sharif Al Erian, Secretary General of the EOC said: "This is an exciting partnership that will go a long way to ensuring our athletes have the best possible preparation for future competitions and the upcoming 2020 Tokyo Olympics. We are delighted that the Egyptian Federation of Modern Pentathlon has embraced the potential of genetics, and urge other member federations to explore for themselves." (Inside the Games, 2018). Egyptian athletes were tested according to DNAFit code of practice, which includes mandatory consent of the customer, no testing for talent identification or predicting sporting ability, destroying the sample after the testing, accepting their terms of service and privacy policy (DNAFit webpage, Table 4). However, once the DNAFit made the genetic testing on the athletes, it is not known how this information was exactly used (supposedly DNAFit collaborated with Egyptian sports science department to integrate the insights from DNA with training regimes and nutrition plans), who had or still has the access to it, how privacy and confidentiality once results have been given to the players and governing bodies itself was guaranteed. Moreover, at the moment it is impossible to measure if Egyptian athletes have or will benefit from genetic testing done on them.

- **China.** In August 2018 Chinese Ministry of Science and Technology announced that they will establish a laboratory standard for the selection of athletes by genetic markers, and that athletes with the potential to represent the country in the Olympic Winter Games in Beijing 2020 will have to undergo whole genome sequencing. The large-scale genetic profiling, screening and analysis work will be carried out by the General Administration of Sport of China, the Ministry of Education and the Chinese Academy of Sciences (South China Morning Post, 2018). The news
immediately got attention all over the world and also quite a lot of criticism. What is the difference between the Chinese and Egyptian decision to do genetic testing on their athletes, that one is being so criticized and the other practically went unnoticed? The main and important distinction is that China wants to use genetic testing for athlete selection, which at the moment is not scientifically proven, and moreover, it goes against the non-genetic discrimination ethics. On the other hand, Egypt is not using genetic testing for selection, but for improvement of training regimes and nutrition. Nonetheless, there are other concerns about the Chinese decision. First, there will be no consent of the athlete as they will have to undergo the genetic testing if they want to go to the Olympics. Secondly, they will or already are sequencing the whole genome, the whole genetic information of the athletes and the main question is, who will have this information, will the government access the information whenever they feel like it, which genes will be looked at, how the privacy and the confidentiality will be guaranteed in the country with known violation of human rights and ethics. The exact procedure of the testing, the budget, the code of practice, etc., was not disclosed and was impossible to find any data about it.

- **NFL.** Genetic testing is banned in the NFL under the current Collective Bargaining Agreement between the players and the owners (South China Morning Post, 2019). In US the three major leagues, NBA, NFL and MBL set the rules by the Collective Bargaining Agreement. It is not known why exactly NFL included genetic testing in the agreement, but it most probably has to do with the loose general regulation of the anti-doping rules in the league, and it could be that genetic testing at the time of the negotiating the agreement might have been seen more as the threat to the players than anything else (in 2011).

- **FC Barcelona.** One of the few clubs that has admitted using genetic testing, even though indirectly, is FC Barcelona (Mirror, 2016). There are reports that few Premier League Clubs collaborate with DNAFit, however, the names of the clubs were never given (Daily Mail, 2014). FC Barcelona is allegedly the first club in the world to try the method on professional football players, including Messi, Neymar and Suarez. They are using it for injury prevention and the whole process is overseen by club’s doctor Ricard Pruna, who has been taking consented player’s mouth swab to study 45 different genes in collaboration with Swedish doctor Matilda Lundbland, who said: "It is very sci-fi. Doctor Ricard and I are looking at the genes in saliva and finding ways to individualize fitness programmes" (MailOnline, 2016; Mirror, 2016). Ricard Pruna was contacted through official mail at FC Barcelona to be interviewed about how FC Barcelona is doing genetic testing and how he sees possible regulation of it in sport, but unfortunately, he never responded to the mail. In 2017 FC Barcelona quietly launched Barcelona Innovation Hub, which was created to help invent the future of football and where science and sport intersect (Financial Times, 2019). Supposedly FC Barcelona let’s Hub’s specialists research freely and genetic testing
is part of the Innovation Hub. Barcelona’s club president Bartomeu said: "We have the best laboratory in the world – 2500 men and women athletes from eight years old to 30, in different sports" (Financial Times, 2019). This statement, in my opinion, is clearly saying that they are doing scientific research on all of their athletes. Actually, they are not just doing it, but also publishing some of the results in peer-reviewed scientific journals. For example, when the name Ricard Pruna, the doctor responsible for genetic testing, was put in the pubmed search (one of the biggest and mostly used scientific journal libraries), there were 21 scientific papers with him as an author, majority of them on genetic testing, susceptibility to muscular injury, and nutrition. In some studies Ricard Pruna and collaborators used DTC company S.M.Genomics (Table 4) for genetic testing and in others laboratories of the University of Barcelona were used (Pruna et al., 2013; Artells et al., 2016). S.M.Genomics was contacted through mail for the interview on genetic testing, but again, no response was received. Majority of the studies were approved by Ethics Committee at Universitat de Barcelona (registry no. IBR00003099) or by the Ethics Committee of the Hospital Clinic in Barcelona (registry no. 2012/7117), however, what this really means is that these two Ethics Committees approved the study itself, the design and use of human material for the study, but it has nothing to do with the ethical dilemmas about the use of genetic testing in sport discussed in the chapter 1.4.2. Moreover, in all studies the players were informed about the procedure of the study and gave their signed consent (Pruna et al., 2013; Artells et al., 2016; Clos et al., 2018). To summarize, FC Barcelona is doing genetic testing as scientific research on their athletes and publishes findings of which gene variants might be associated with injury risk in scientific journals, however, they do not publish how they optimize trainings according to data obtained (which they do by themsleves) nor if they use this data also for talent identification, which was suggested in the UEFA Research Grant Final Report 2012 in the section Benefits to UEFA with exact words: "Measures of injury prevention and methods of talent selection can be improved." (Pruna, 2013). Moreover, there is no information on data protection, confidentiality, if all the subjects involved in the study are informed about their specific results, what they do with the DNA sample (is it destroyed or stored) and what happens to the genetic information and sample of the players who leave FC Barcelona, like for example Neymar.

The case studies revealed that nations, leagues or clubs are differently doing genetic testing and using different DTC companies or laboratories. Also there is no consensus nor data which gene variants were tested and which protocols were developed to implement genetic testing results into practice (i.e. training, nutrition optimization). It was impossible to find guidelines, frameworks or any regulations that were followed by Egypt, China or FC Barcelona. The major issues of all cases is data protection, confidentiality, who is the owner of the genetic information obtained and ethical concerns. Major European leagues like Turkish Airlines EuroLeague (Ignacio Muro, Euroleague’s medical
doctor: "EuroLeague has not an official position about genetic testing"), UEFA (even though it did fund Ricard Pruna’s research on genetic markers for injury susceptibility (Pruna, 2013)) or any other (to my knowledge) do not have official statements nor positions on genetic testing. This lack of guidance and regulation by sport governing bodies basically allows the federations/clubs to do whatever they want regarding the genetic testing in sport.

2.4 Costs and benefits of implementing genetic testing in sport

Genetic testing being already a part of the sport as the literature review and field analysis have revealed, the next question was, what are the actual costs and benefits of it; i.e. how much would the genetic testing cost a given club and how would its implementation affect the revenues. Since there is no data yet on the actual benefits or drawbacks of genetic testing in sport, it is impossible to predict the actual impact on revenues, nevertheless, some estimations will be given.

2.4.1 Budget for the genetic testing on clubs level

The budget needed for genetic testing will depend on the DTC or laboratory contracted, the number of players to test, etc. The budget for the football and basketball club that never did the genetic testing would be around 60,000 € for the football club and slightly less, around 37,000 € for the basketball club (Table 5). If more or less players need to be tested, the budget would vary slightly as it is calculated only for the first squad. In my opinion, the clubs should sequence directly to whole genome, as the price has decreased reasonably and there are several advantages over directly testing selected gene variations as already discussed in section 2.3.1. There will most probably be no additional costs for the medical doctor to collect the samples as all sport clubs have several medical doctors on the paycheck and the same is true for nutritionist. However, most probably majority of the clubs do not have employed yet the bioinformatician, who can analyze the genome sequencing data and give the report on which genetic variants are present in athletes. The budget for freelancing bioinformatician is probably slightly over-exaggerated (the price per hour estimation is from personal communication with few of them), however, at least in the first season of genetic testing implementation the bioinformatician will have to spend quite some hours for making the profiles for all the players. Most of the clubs probably also do not have people who know how to integrate genetic information into practice as FC Barcelona has (nutrition optimization, training optimizations, etc.), so they will definitely need genetic counseling or they will have to educate existing staff, namely medical doctors and nutritionists. And lastly, whole genetic information in the digital format are big data files that will have to be handled and protected by IT support people (that majority of the clubs has IT staff on the paycheck) and some additional software, tools and programs will have to be installed/bought. The budget in Table 5 is for the first year of implementation as the second year the
costs will be lower for several reasons: (i) not the whole team will have to be sequenced, just the new players (and if they do not already have their genetic data); (ii) probably the bioinformaticion will do less hours as there will be less reports to do and newly identified genetic variations connected to sport performance, injury susceptibility or nutrition optimization will be very few if any; (iii) similarly the hours of genetic counseling and (iv) IT support and tools will be reduced.

**Table 5: Budget for the genetic testing on clubs level.**

<table>
<thead>
<tr>
<th></th>
<th>Football</th>
<th>Basketball</th>
</tr>
</thead>
<tbody>
<tr>
<td>Whole genome sequencing</td>
<td>14.376€ (24x599€)</td>
<td>7.188€ (12x599€)</td>
</tr>
<tr>
<td>Medical doctor to take the DNA samples</td>
<td>0€</td>
<td>0€</td>
</tr>
<tr>
<td>Bioinformaticion</td>
<td>20.000€ (200h x 100€/h)</td>
<td>10.000€ (100h x 100€/h)</td>
</tr>
<tr>
<td>Genetic counseling</td>
<td>0-10.000€</td>
<td>0-10.000€</td>
</tr>
<tr>
<td>IT support and tools</td>
<td>15.000€</td>
<td>10.000€</td>
</tr>
<tr>
<td><strong>TOTAL</strong></td>
<td>~60.000€</td>
<td>~37.000€</td>
</tr>
</tbody>
</table>

Source: DTC companies listed in Table 4 and student’s personal communication and knowledge.

In the case when the nation decides to implement the genetic testing like China did, the budget increases proportionally to the number of athletes that will be tested. Unfortunately, China never revealed the budget they have for the genetic testing of its athletes.

### 2.4.2 Reduction of cost of injuries

Every year millions of euros are being lost on the salaries of the injured players. In the Table 6 predictions about how much money could have been saved in major European football leagues (Premier League – UK; La Liga – Spain, Serie A – Italy; Bundesliga – Germany; and Ligue 1 – France) for the season 2017/2018, if the genetic testing for injury susceptibilities and risk would decrease the rate and time of injuries by only 10%. The cost of injuries in the Premier League in the season 2017/2018 was 210 million £ (246,3 million € on a currency exchange rate on 5.1.2020) for the salaries of injured players (JLT Specialty Limited, 2019). The average annual reported player’s salaries for each league (Sport Intelligence, 2018) were used to calculate the cost of injuries in other European football leagues (Table 6) under assumption that in all leagues the rate and time of injuries was the same as in the Premier League. In this prediction were not included the medical costs of injuries, the impact of injuries on teams performance, the impact of injuries on ticket and merchandise sales and other revenues that could be affected by injuries of players, especially if they are the best players of the club. The prediction was calculated for average of salaries and how much the average club in a given league would save (Table 6); obviously the more club spends on its squad salaries, more savings could be expected from injury prevention programme based on genetic testing. In addition, clubs could also save money on transfer fees and actual salaries of the players, whose genetic
information predicts high injury and long recovery time risk. Moreover, the assumption was made that genetic testing would prevent only 10% of injuries, but reality could be that it would prevent much more. The data at the moment is not available, the only (or one of them) club that could make better estimation on this is FC Barcelona, however, it was not possible to get this information.

Table 6: Prediction of savings on salaries for injured players in season 2018/2019.

<table>
<thead>
<tr>
<th>League</th>
<th>Average player's salary</th>
<th>Cost of injuries in the league</th>
<th>Savings (10%) per league</th>
<th>Number of clubs</th>
<th>Savings (10%) per club</th>
</tr>
</thead>
<tbody>
<tr>
<td>Premier League</td>
<td>3.4 million €</td>
<td>246.3 million €</td>
<td>24.6 million €</td>
<td>20</td>
<td>1.23 million €</td>
</tr>
<tr>
<td>La Liga</td>
<td>2.5 million €</td>
<td>181.1 million €</td>
<td>18.1 million €</td>
<td>20</td>
<td>0.905 million €</td>
</tr>
<tr>
<td>Serie A</td>
<td>1.7 million €</td>
<td>123.2 million €</td>
<td>12.3 million €</td>
<td>20</td>
<td>0.615 million €</td>
</tr>
<tr>
<td>Bundesliga</td>
<td>1.6 million €</td>
<td>115.9 million €</td>
<td>11.6 million €</td>
<td>20</td>
<td>0.64 million €</td>
</tr>
<tr>
<td>Ligue 1</td>
<td>1.1 million €</td>
<td>79.7 million €</td>
<td>8.0 million €</td>
<td>20</td>
<td>0.4 million €</td>
</tr>
<tr>
<td>TOTAL</td>
<td>746.2 million €</td>
<td>74.6 million €</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Source: JLT Specialty Limited, 2019; Sport Intelligence, 2018.

For the basketball, more precisely Turkish Airlines EuroLeague it was not possible to get the injury statistics or the percentage of time the players did not play due to injuries. Dr. Ignacio Muro, medical doctor at EuroLeague, said they at the moment do not publish injury statistics, but they do share the data within the league with all the doctors (personal communication through mail). In order to predict the savings on salaries for injured players in EuroLeague, first the average total budget for the season 2019/2020 was calculated based on published budgets of the clubs (TalkBasket, 2019); 417.3 million € for all 18 clubs, which gives on average 23.18 million € per club (with FC having the highest budget of 41 million €, and Crvena Zvezda the lowest with 8.3 million €; TalkBasket, 2019). Salaries for the players are around 60% of the budget in general in EuroLeague (MBA in Sport Business first case study data), giving an average of 13.9 million € per club for players' wages, and with percentage of injuries in basketball hypothetically around 10-15% (Harkins, 2013), would mean that approximately 1.39 to 2.085 million € is the cost of injuries per club in EuroLeague (just on injured players salaries). Assuming again that genetic testing would reduce injury rates only 10%, this would mean around 139-208.5 thousand € of savings per club. The same is true here as said for the football clubs, all the other benefits associated with less injury of the players are not included in this prediction.

2.4.3 Cost-efficiency analysis of implementation of genetic testing on the clubs level

Due to several limitations of doing a cost-efficiency analysis on the leagues level as for example the data on the tickets revenues on average for the whole league is difficult to find, or under
or over estimation of the benefits due to averaging the budgets, etc.; the cost/efficiency analysis was done on one club, in this case Real Madrid. The reasons for choosing Real Madrid are: (i) supposedly they are not doing genetic testing at the moment or there are no reports about it (FC Barcelona for example could not have been chosen precisely because they are already implementing it); (ii) Real Madrid is one of the best European and World club; (iii) Real Madrid has two sport section, football and basketball. The predictions made in sections 2.4.1 and 2.4.2 will be taken into consideration here and corrected accordingly.

**Table 7:** Cost-efficiency analysis of implementing genetic testing for the Real Madrid club.

<table>
<thead>
<tr>
<th></th>
<th>Football</th>
<th>Basketball</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cost of implementing genetic testing</td>
<td>-0.06 million €</td>
<td>-0.037 million €</td>
</tr>
<tr>
<td>Benefits due to reduction of injuries</td>
<td>3.45 million €</td>
<td>0.24 million €</td>
</tr>
<tr>
<td>Benefits due to decrease in talent identification costs</td>
<td>n.d.</td>
<td>n.d.</td>
</tr>
<tr>
<td>Benefits due to nutrition optimization of the players</td>
<td>n.d.</td>
<td>n.d.</td>
</tr>
<tr>
<td>Benefits due to increase in merchandise sales</td>
<td>n.d.</td>
<td>n.d.</td>
</tr>
<tr>
<td>Benefits due to increase in ticket sales</td>
<td>1.43 million €</td>
<td>shared with football</td>
</tr>
<tr>
<td><strong>TOTAL</strong></td>
<td>4.82 million €</td>
<td>0.203 million €</td>
</tr>
</tbody>
</table>

n.d., not determined

Source: Statista, 2019; TalkBasket, 2019; Real Madrid, 2019; Table 5 and 6 of this study.

In the table 7 the cost and benefits of implementing the genetic testing into club Real Madrid are calculated. The costs of implementations are the same as in the Table 5, while the benefits due to reduction in injuries has been adjusted from the Table 6 according to actual average salaries of Real Madrid football players, which is 3.8 times higher than reported for La Liga (Statista, 2019), and to the actual budget for Real Madrid basketball team, which is 40 million € (TalkBasket, 2019). The ticket revenue for 2017/2018 was 143 million for both sports (Real Madrid, 2019), and the assumption was made that genetic testing would improve the ticket sales by 1%. At the moment it was impossible to predict the benefits of genetic testing on talent identification, nutrition optimization and merchandise sales, this is why it was left as not determined. Altogether, quite cautious and unambitious cost-efficiency analysis showed that implementation of genetic testing into Real Madrid club would save them at least 5 million € per year (taking into consideration just the first teams in football and basketball). Obviously, big clubs with high player’s salaries would benefit more from the implementation of genetic testing than smaller clubs. The possible drawback or risk could be that genetic testing would turned out not to reduce injury rates or improve any of the categories mentioned in Table 7 (due to not implementing it properly into training regimes or that simply does not work as
expected), in this hypothetical case, the financial loss would be the cost of the implementation, which for the Real Madrid is around 0.1 million €.

**In summary,** the field analysis showed that genetic testing is already taking place in sport in Europe based on results obtained from the survey on athletes and support staff. The survey also revealed that athletes are curious about possibility of having genetic variants related with sport performance and injury susceptibility, and that are less concerned about ethical issues raised by use of genetic testing in sport than previously reported by Varley et al. (2018). Analysis of DTC companies offering sport genetic testing showed no consensus on what genetic variants should be tested, there is quite a disparity in number of genes tested, costs of the tests, explanation and genetic counseling offered, and which exact procedures were followed. For someone who is not an expert in the field is hard to digest all the scientific data and to smartly decide which company to choose. Furthermore, case study analysis on different nations or club that are already implementing genetic testing indicate again no consensus on which genetic variants to use, which procedures or guidelines were followed, nor give any data on how the transfer of genetic data into sport practices was done. In addition, there is still a debate if genetic testing should be allowed for talent identification as seen from the China case. And finally, cost-efficiency analysis showed that if genetic testing in sport would help reduce injuries by 10% that alone would save a lot of money in different leagues and clubs. A more detailed cost-efficiency analysis was made for the club Real Madrid, where also possible benefit for increase in ticket sales revenues was included, and the data predicted about 5 million € more in the club’s cash-register.
III. RECOMMENDATIONS FOR IMPLEMENTATION OF SPORT GENETIC TESTING INTO SPORT INDUSTRY

3.1 Recommended approach towards implementation of genetic testing into sport industry

In this study the possible implementation of the genetic testing in sport was analyzed. Even though using genetic testing to improve competitiveness is becoming a trend, there is at the moment no consensus in the sport industry whether it should be done or not. This study clearly showed lack of the regulation and guidelines for genetic testing in sport on the governing sport bodies level. Based on the findings from literature review and field analysis; and also based on my personal scientific background (M.Sc. in Microbiology; PhD in Biomedicine and more than 15 years of scientific investigation in leading world laboratories) actions are proposed that could be considered by sporting community when implementing genetic testing in sport.

Proposed approach. Implementation of genetic testing in sport on all levels (international, national, leagues and clubs) by setting the minimal regulation guidelines that have to be followed in order to safeguard the athlete’s wellbeing and rights. This section concentrates on key considerations regarding genetic testing in sport that have been identified by literature and field analysis, and should be resolved by governing sport bodies. Moreover, clear action plans for implementation of genetic testing in sport on all levels will be given.

The key considerations of implementing the genetic testing into sport that have to be addressed by sport industry are:

1. Ethical considerations such as athlete’s right to open future
2. Legal considerations as for example genetic information non-discrimination acts
3. Consensus about genetic traits to be tested (especially regarding sport performance)
4. Standardization of the genetic testing procedures and protocols in sport
5. Guidelines on how genetic information is translated into training and nutrition regimes (or specialists that have to be employed in order to properly integrate genetic information into sport practices)
6. Genetic counseling
7. Data protection and confidentiality
In the chapter 1.3 stakeholders involved in sport genetic testing were identified and this study is providing more in detail the guidelines on how to implement genetic testing for each of listed stakeholders:

- Nations
- Federations/leagues
- Clubs
- Players/athletes

### 3.2 Implementation and regulation of genetic testing on the level of sport governing bodies

Sport is unique as it has enormous global power, impacts many areas of social activity and promotes economic growth. In the quest to be the best, athletes and clubs are constantly trying new training methods, new materials that would increase the speed, and often they are also among the first to try the new technologies. With science advancing in understanding which genes are responsible for sport performance, injury susceptibility and personalized nutrition, sportsmen are already thinking in how to use it in order to improve their competitiveness, not always in a proper way and in a benefit of the athletes or sport itself. WADA immediately recognized the danger of new technologies and banned gene doping. However, the new scientific advances can be taken from another point of view and used for genetic testing of athletes for different genetic traits related to sport. As said before, reading genetic information of an individual is not prohibited by WADA, however, there are several ethical and legal issues about genetic testing in sport, the most concerning among others being the talent identification based on genetic traits, that at the moment not even science can confirm to be the real predictors of elite sport performance, and the genetic discrimination. Even though the survey among athletes and support staff showed they have no problems of taking genetic testing if asked by their clubs or sport governing bodies and they were in general less concerned about ethical issues associated with it as expected (Table 3 and 4), it is known that elite athletes are often blinded by their eagerness to be better than the rest, that they would do whatever it takes to get there. This is why sport governing bodies should set the guidelines for regulation of genetic testing in sport to ensure the rights of the athletes.

The first line of guidelines for the regulation of genetic testing in sport should come from the highest or better said most recognized sport governing body, which is International Olympic Committee (IOC), together with WADA, as genetic testing in sport and gene doping are closely related and measures/recommendations proposed in genetic testing can actually benefit WADA’s fight against gene doping. Since WADA is already considering to request genetic information of the
athletes for gene doping purposes (Niiler, 2018), it makes more sense to start implementing the use of athlete's genetic information at the level of IOC/WADA. All the European’s National Olympic Committees are part of IOC and they generally adopt their guidelines and practices. The proposed recommendations to be implemented by the IOC (and WADA), and thereafter adopted by European (and other NOCs) are:

1. Given the fact that we already are in so called genomic era and that the costs of the whole genomic sequencing have dropped significantly and will continue to decrease, the first recommendation is to **create the Athlete Genetic Passport** (the name was given in concordance with the biological passport) as a mean to define athlete’s genetic information profile in similar way as the Athlete Biological Passport was created in 2009 (WADA, 2009). One of the main objectives of introducing the Athlete Genetic Passport is:

   - To provide a sample of athlete’s genetic information for WADA to have a reference to which to compare the genetic information obtained in the gene anti-doping strategies. The exact gene anti-gene doping strategies are not fully developed yet, but they will have to have a reference genetic information to which to compare the sample. Even though, anti-gene doping strategies are not in the scope of this master thesis, it would be faster and more efficient to in part combine the two processes. In addition, the Athlete Genetic Passport could help clear some cases of suspected doping due to rare mutations that improve red blood count or increase the level of testosterone in female athletes.

   - To provide valuable information that can be used for determining athlete’s genetic variants related to sport performance, injury susceptibility, nutrition optimization (however, this will not be done directly by IOC or WADA – further explained later on) or any other that might be proven to be important for athlete wellbeing in the future.

   - To safeguard (as a backup system) the athlete’s genetic profiles in the case of being lost (by athlete itself or clubs, national governing bodies (hardware getting burned for example) – the point here is to have a real and early in athlete’s life taken sample for the whole genome sequencing before any possible manipulation of genetic information might have been done as for example genome editing by the CRISPR technology, because if hypothetically the sample would have been taken after the genome manipulation, it would have been considered as the genetic information that athlete was born with since the genome editing is almost impossible to detect at the moment and there is no reference genetic information to compare with), misuse, misconduct, falsification of the information, etc.
• To have a library of athlete’s genetic information that in the future might be used to determine the genetic variants and traits related to elite sport performance, injury susceptibility, nutrition optimization, etc. The data could be used anonymously and with the consent of the athletes by the Gene doping and Laboratory Expert Groups already formed at WADA or by any other science research group that IOC or NOC (National Olympic Committee) select based on the scientific criteria. It is not new and unusual that the genetic information of the athlete’s is used for the scientific research by the sport entity as for example FC Barcelona is doing exactly that.

• To harmonize the sport genetic testing and gene anti-doping strategies.

2. In order to obtain the Athlete Genetic Passport, **whole genome sequencing laboratories or appropriate DTC companies have to be accredited** and approved as such by the IOC or NOC for sequencing the genomes of the athletes for their genetic passport and have to comply the preset standards and quality controls. Field analysis showed huge discrepancies in the way DTC companies are conducting their work (Table 4), especially which gene variants they test, it was not possible to find all the information about the accreditation, quality controls they have, etc. It is of great importance to have approved and accredited laboratories, which will provide accurate sequencing, since the Athlete Genetic Passport will be athlete’s reference genetic information for gene anti-doping strategies and the gene variant screenings for sport performance, injury susceptibility and nutrition optimization regimes. As explained in field analysis, whole genome sequencing is the preferred method and is the one to use for the Athlete Genetic Passport. In the protocol for the whole genome sequencing it has to be incorporated that the DNA sample of the athlete is not stored by the accredited laboratory after the results have been sent to the customer. IOC/WADA and NOC have to decide if the DNA sample is stored by themselves or destroyed.

3. A debate on which gene variants should be used for genetic screening is still ongoing and as seen from field analysis, different DTC companies use different gene variants or different number of gene variants to assess sport performance, injury susceptibility and nutrition optimization, this is why **appropriate genetic testing committee or expert group will be formed by IOC**, which would integrate leading scientist on the field (or the already existing Gene Doping Expert Group of WADA should integrate this function into its work plan) and the role of the committee or expert group will be to give recommendations and guidelines on which gene variants to test in:

• Sport performance: here the committee or expert group will give the list of gene variants that without any doubt and substantially supported by scientific research have an impact on sport performance. At the moment the scientific community has not reached the consensus on the
gene variants that would clearly predict sport performance and there is quite some skepticism about it, especially if it should be used for talent identification.

- **Injury susceptibility**: the expert group will provide the list of genes that have been scientifically proven to have a role in injury susceptibility, injury recovery rate, etc. However, here it will be optional, it will be just the recommendations which genes to check in the Athlete Genetic Passport, but not obligatory as for sport performance. Federations and clubs could decide by themselves and also based on their proper research and experience which genes to include in this category and how to integrate it into the training regimes.

- **Nutrition optimization**: similarly as in the injury susceptibility, only the recommended list of genes will be provided, however, federations and clubs will decide based on their experience and research which to check in order to improve athlete’s nutrition regimes.

This expert group will set the minimal guidelines of which gene variants have been scientifically proven to have an impact on above mention genetic testing categories in sport and will also help the federations and clubs that have no proper staff incorporated to do this by themselves (as for example FC Barcelona has). In addition, this will also help the Gene Doping Expert Group of WADA to create a list of possible genes that could be targeted by gene doping strategies and technologies.

4. The Ethics Committee of IOC, that is a guardian of the ethical principles, will integrate the ethics on genetic testing in sport in its agenda and **provide ethics guidelines in relation to genetic testing in sport**. Even though survey on athletes showed they are not particularly concerned about ethical issues (Table 2 and 3), sport governing bodies should provide minimal regulation in order to safeguard athlete’s interests. The ethics guidelines should include minimal regulation on use of genetic testing in sport as for example:

- **Open athlete’s future**: prohibition of identifying future sport talent solely on genetic testing on gene variants supposedly identifying future elite athletes. However, genetic testing could be an additional tool in talent identification, but never should be used to exclude from the possible sport career (like for example in youth selections).

- **Genetic discrimination**: currently known legal international and regional acts on genetic information non-discrimination should be revised and adopted or redefined for the sport industry (non-discrimination in sport employment, insurance, etc.).

- **Ethics of genetic testing data or genetic information on transfers and medical examinations**: ethical questions like should club have genetic information about the players injury
susceptibilities and sport performance before signing the contract with him or after signing the player just to optimize his/her training regimes, should player’s transfer fees and salaries be subjected to reduction due to non-favoring genetic information on sport performance and injury susceptibility, should medical examinations include genetic testing for different disease and health predispositions, etc.

5. **Collection and management of Athlete Genetic Passport** should be done by NOCs for its athletes and at time when they start to compete on the international level (i.e. start to participate in European’s championships, European’s leagues like EuroLeague or UEFA competitions, etc.). Collection is done as follows:

- The sample should be taken by the doctor in the presence of the NOC delegate or appropriate national federation league delegate (like for example national basketball or football federation) in order to ensure the proper athlete’s identity. If by any chance athlete already has whole genome genetic information, the protocol by which was obtained has to be revised and if it does not coincide with the ones set by the IOC and NOC, then it should be repeated.

- The sample is afterwards sent to accredited laboratory for whole genome sequencing and the results are sent back to the NOC and the athlete itself.

- NOC is responsible for sending the data to IOC/WADA (if they request the genetic information, in any case NOC has to ensure the proper management of Athlete Genetic Passport – explained below).

- There are at least three copies of the Athlete Genetic Passport, one at IOC/WADA (if requested), one at NOC, and the player also has one copy. The national league, federation or athlete’s club can request the Athlete Genetic Passport, however, the athlete itself has to give signed consent for it.

- All the procedures of the sample collection and processing have to be standardized and follow exact protocols, which have to be written and specified in detail:
  - Sample collection procedure.
  - Transportation requirements.
  - Sample reception and timing of the analysis.
  - Instrument check (that the sequencing machine is working properly and in accordance with the manufacturer’s recommendations).
  - External quality assessment of the technology should be implemented.
  - Sample analysis procedure.
- Reporting of results protocol (how the results are presented and sent).

The management of Athlete Genetic Passport will be on the level of IOC/WADA incorporated into already existing management of Athlete Biological Passport and it will represent and additional file in the Athlete Passport Management Unit (WADA, 2019). In the case of NOC, national federations and/or regional leagues management of the Athlete Genetic Passport has to be incorporated in accordance with one already existing by the IOC/WADA:

- The federations/leagues that already have the management of Athlete Biological Passport or equivalent will add the file of Athlete Genetic Passport to the Management Unit.
- The federations/leagues that do not have this management already in place will create such a unit.
- Governing bodies can access the Athlete Genetic Passport when there is a suspicious of genetic doping (WADA), or when an athlete’s federation/club or athlete itself request the genetic data (see point 6).
- The Athlete Genetic Passport should be administered and managed through online secure database management tool that for WADA is the Anti-Doping Administration and Management System (ADAMS; WADA, 2019), and other sport governing bodies should incorporate similar online secure database management tools.
- IOC/WADA and other sport governing bodies should incorporate appropriate personnel for the Athlete Genetic Passport Management (i.e. bioinformaticians, data analysts, IT experts).
- All the procedures at the sport governing bodies and clubs level have to be standardized and follow the same protocols in order to allow sharing and mutual recognition of the Athlete Genetic Passport between different sport organizations.
- Proper data protection and confidentiality measures should be implemented on all levels from sport governing bodies like IOC/WADA till the club level in order to ensure that just authorized personnel has access to the Athletes Genetic Passport and that data is not disclosed to third parties.

6. The Athlete Genetic Passport use and sharing should be properly specified. All the athlete genetic information, also the specific genetic variants identified should be collected under one file of the Athletic Genetic Passport per athlete. The Athlete Genetic Passport could be shared with appropriate agencies if there is a suspicion of gene doping or editing (probably carried out by WADA itself; how this is done is not in the scope of this thesis), with other governing bodies that athlete is part of and with athlete signed consent (national federations, leagues, etc.), with the
athlete club and with the athlete signed consent (if the club does not have this information already). The files have to be shared via online secured mailing system. The Athlete Genetic Passport should not be shared with the insurance companies, sport sponsors, journalist and public. The athlete himself or herself could share his or her genetic information with whomever he thinks is appropriate, however, he/she should receive genetic education and/or counseling (see point 7) before and after the genetic testing in order to understand the possible pros and cons of disclosing his/her genetic information publicly.

7. **Education and genetic counseling.** From the survey conducted it was seen that athletes are not familiar with the genetic testing in sport as only 27.6% of athletes and 31.8% of support staff heard of it and the rest was not sure or never heard of it (Table 2 and 3). Sport governing bodies should implement education programs on genetic testing in sport for their stakeholders including players, coaches, doctors and other staff. The education programs could include e-learning, course for medical support staff, etc. In addition genetic counseling by experts should be encouraged and incorporated into the guidelines, especially at the level of federations and clubs.

8. **Costs of genetic testing.** The costs of whole genome sequencing should be covered by national governing bodies for their athletes (like for example China will do for theirs as presented in the case study) or by the athlete’s clubs as they are the ones who will benefit the most from the genetic testing in sport, especially if all the predictions about genetic testing will be indeed fulfilled.

As seen from this chapter, the first level of recommendations for genetic testing in sport is implementation and integration of it into well defined Athlete Biological Passport. This has the following advantages:

- It simplifies the implementation of the genetic testing into sport as it follows the pre-set rules for the Athlete Biological Passport.
- It serves several purposes – determination of genetic variants related to sport performance, injury susceptibility and nutrient optimization; and it serves as the reference genetic information of the athlete in the fight against the gene doping and gene editing.
- It could be easily adopted or implemented by NOCs, federations, leagues.
- It reduced the costs of the implementation as basic infrastructure is already present.

**3.3 Implementation of genetic testing at the national sport federations and leagues level**

Once the genetic testing in sport is approved and the protocols and guidelines are harmonized by the IOC/WADA, the NOC and different international and national sport federations could easily adopt the majority of the measures proposed to the IOC/WADA. Normally NOC and international
federations adopt IOC/WADA’s proposals, however, this is not always the truth for the national sport federations and even less for the independent leagues like for example UEFA and EuroLeague. As seen in the case study of Egypt in the field analysis Egyptian Olympic Committee (EOC) signed the deal with the DNAFit genetic testing company, but just the Federation of the Modern Pentathlon decided to test their athletes, even though EOC was encouraging other federations to do the same, there are no reports at the moment that it really happened (Sport Industry Group, 2018; Inside the Games, 2018). Moreover, there was quite a dispute going on several years ago, when FIFA and UEFA rejected the WADA rules for "whereabouts" of the payers; "whereabouts" is information provided, normally by individual elite athletes, about their location at a given time in order for WADA to conduct the out-of competition doping controls (Law in Sport, 2017), however at the end the agreement was reached. If some leagues will not adopt the IOC/WADA protocols and guidelines about genetic testing in sport, the recommendation is that they give a position statement about the genetic testing in sport (related to their sport of course), where they should clarify at least the following issues:

- Federations/leagues position on genetic testing in sport – do they approve it or not, do they leave free hands to the clubs about the genetic testing in sport (for example, since there is no regulations about genetic testing in sport at Spanish La Liga and in UEFA, FC Barcelona actually can do whatever wants and does not have to report about it to no one – it is important to specify here that they are most probably doing it in ethically and scientifically proper way).
- Position on ethically sensitive issues like for example talent identification based only on genetic testing for sport performance.
- Position on legal issues as genetic discrimination in sport employment and insurance, etc.

If the genetic testing in sport becomes widely used as it seems from the survey on athletes and support staff, and if the benefits of the genetic testing in sport reach their predicted potential, it is obvious that all the federations and leagues will have to adopt the strategies for its implementation. Of note, at the moment there is no clear evidence that the gene doping or gene editing is really happening in sport, nevertheless, the measures are being put in place to prevent it. On the contrary, in the case of genetic testing in sport, it is already known that is already happening in sport, clearly calling for the regulation from the sport governing bodies in order to protect the rights of the athletes.

3.4 Implementation of genetic testing at the club’s level

The implementation on the club’s level is probably the most important one, as there is where most of the genetic testing in sport will probably take place. The main reason being is the benefit in saving money, especially if the genetic testing in injury susceptibility will help reduce the medical
bills and paying salaries to injured players – this will have the massive financial impact on club’s savings as showed by cost-efficiency analysis in the field analysis part (Table 6 and 7). Moreover, the cost-efficiency analysis was quite cautious and did not include all the parameters on which genetic testing in sport could have a positive impact, predominantly as there was no available data to do predictions from. Taken all this together, the recommendation for the clubs (independently if sport governing bodies will regulate genetic testing in their sport) is to implement the genetic testing into their daily life, especially for injury susceptibility and nutrient optimizations. And this is even more recommendable for the clubs with high player’s salaries as they will benefit the most from the implementation of the genetic testing (Table 6 and 7), and the costs related to genetic testing are quite reasonable and affordable (Table 5). If IOC/WADA have already implemented the recommendations proposed in the section 3.1, then club could adopt the majority of the measures proposed there, however, if there is no regulations, measures and guidelines from any sport governing body at the time of implementation of the genetic testing, then the following plan of actions is proposed:

1. **Creation of the Athlete Genetic Passport** as previously said, but now at the level of the club. This means that proper Athlete Genetic Passport Management Unit will have to be created at the club or the file could be added to the already ongoing medical and health data management of the players. In that file the player’s genetic information and all the gene variants checked will be saved. The club will have to ensure proper data protection and confidentiality measures.

2. **Choosing the appropriate DTC genetic testing company or laboratory** that does whole genome sequencing as this technology is better than separately testing different gene variants (previously explained why, Table 4). In addition, it should be checked that the company is accredited, has standard and quality controls; i.e. that does its job properly and adequately.

3. **Preparation of different protocols as**: sample collection protocols, sample transport to the chosen genetic testing company protocols, preparation of the consent of the player’s document, management of the Athlete Genetic Passport protocols, etc.

4. **Employment or education of existing personnel** to define which gene variants will be looked at, once the whole genome sequencing has been done by the genetic testing company. Moreover, bioinformatician and/or data analyst should be contracted to extract the information from the Athlete Genetic Passport (i.e. to find in the whole genome sequence of the players the gene variants that were decided to look at).

5. **Translation of identified players gene variants into practice**; i.e. based on the gene variants found in different players personalized trainings and nutrition regimes should be made. Genetic counseling might be needed here and also personnel that knows how to combine and optimize genetics and training/nutrition regimes.
6. **Set minimal ethic guidelines** that goes with the clubs philosophy and is accepted by the players, like for example non-genetic discrimination, talent identification will not be determined solely on genetics, etc. Since there is no regulation by the governing bodies it would be hard to know if this proposed action has been put in practice.

7. **Educate and inform players about the genetic testing** that has been conducted on them, including which gene variants were tested, why and what does it mean. Also, players should have their copy of their Athlete Genetic Passport.

8. Another important question is what happens with the Athlete Genetic Passport when the athlete leaves the club. In my opinion, **the club should first give the copy of the file to the athlete and destroy the file it has on him/her**. However, this is a little bit controversial as the club paid money for all this information and will most probably be resilient to destroy it. In addition, the information might be an advantage in the case the player goes to the club that will play against them in some of the competition(s).

### 3.5 Implementation of genetic testing from the players point of view

Athletes are the objects of the genetic testing and they should be properly informed about all the procedures and testing that is done on them. They should also be informed why some gene variants were checked and how this influences his/her sport career. The survey showed that athletes are willing to take the genetic testing if asked by the coach or governing body, clearly showing that they believe in the good attentions of their superiors. However, player or player’s unions should also take some actions in order to protect themselves from improper use of genetic testing. The following actions are recommended for the players:

1. Ask for clear information on genetic testing that will be conducted.
2. Get informed about the genetic testing in general and also the genes that will be looked at and with what purpose.
3. In case of any doubt ask for genetic counseling, if not provided, or do not sign the consent.
4. Ask for your copy of the Athlete Genetic Passport.
5. When leaving the club, ask that your file is erased from the club’s database.
6. Ensure that ethical and legal issues are clarified and met by the club.

In case of individual sports like tennis, swimming, etc., the genetic testing should be implemented and done on national federation’s level as for example the Egyptian Federation of Modern Pentathlon did it under the EOC approval (Inside the Games, 2018).
3.6 Discussion

Benefits. First, this study managed to analyze the current status of genetic testing in sport and identified the level of occurrence of genetic testing in sport in Europe. Secondly, the study analyzed DTC companies selling genetic testing in sport and several cases of use of genetic testing in sport, which enabled identification of key areas of consideration for implementation of genetic testing in sport. Thirdly, the study has provided the guidance for other scholars and sport governing bodies or clubs that would consider implementing genetic testing in sport. Next, the study predicted positive cost-efficiency analysis, which favors implementation of the genetic testing into sport industry. And lastly, the study provided the guidelines and plan of actions for implementation of genetic testing in sport on different levels, including sport governing bodies and clubs.

Risks. This study identified the key areas of considerations before implementing genetic testing into sport. One of the main concerns are ethical and legal issues, especially talent identification based exclusively on genetics and genetic discrimination. Even with banning talent identification and genetic discrimination, it will be hard to properly ensure that this is really not happening in sport, where winning is the ultimate goal. Moreover, some are considering to use the genetics as tool for talent identification already on kids, which at the moment is inappropriate and absolutely not supported by the scientific data. Furthermore, there are also rumors that some would like to use genetic testing already on the level of embryos in order to select the ones that have better possibilities to become future sport stars.

There is also a risk that players/athlete reject the use of genetic testing in sport by the collective bargaining agreement as it happened in the NFL. However, given the fact that genetic testing is already happening in sport, and that athletes are willing to take it if asked by the governing bodies, it is quite unlikely to happen.

Despite measures proposed to protect genetic information of athletes, there is a serious risk of its disclosure. This is why sport governing bodies and clubs should constantly update their data protection measures and check who has the access to this sensitive data.

Another possible risk is that genetic testing in sport does not meet the expectations of improving the sport talent identification, reducing injuries and optimizing nutrition regimes, however, based on literature review and field analysis done, it is unlikely to happen.

Relevance and originality. This is the first study, to my knowledge, that analyzed how genetic testing should be implemented into sport industry and provides guidance for different sport governing bodies and club’s on how to implement genetic testing into their organizations. The topic is relevantly new and there is limited data available about it, especially since the technology is hard to understand by non-scientific people, and the study clearly explains the science behind the genetic
testing in sport to the general public, and additionally explains the drawback and benefits of it by a person that understands science and sport. In addition, the study made a survey on elite athletes and their view on genetic testing, which is the second study of this type, however, the first one was only targeting the elite UK athletes, while in this survey the responses of the athletes all over the Europe were collected.

**Implications.** The study provides the insight in the current status of genetic testing in sport, and identified its occurrence. In addition, an advanced understanding of the impact of genetic testing in sport industry was presented and key considerations of genetic testing in sport were identified. Based on these considerations proposed guidelines and actions were recommended that would aid the sport governing bodies and clubs in how to efficiently and properly implement the genetic testing into their sport.
CONCLUSIONS

Sport industry is constantly in a search for the technologies that would improve sport performance in one or another way. One of the latest finding in genetics suggests the existence of gene variants that could predict the sport performance, injury susceptibility risk and nutrition optimization regimes. Shortly afterwards, the first rumors about the use of genetic testing in sport were reported and ongoing debate on if it should be used in sport or not started. Also different ethical and legal issues were raised. The study aimed at providing the guidelines for the implementation of the genetic testing into sport industry. Literature review identified the stakeholders involved in the sport genetic testing and the current use of genetic testing in sport, but failed to assess the frameworks and procedures of genetic testing in sport as there were no report on them, suggesting that genetic testing in sport is not regulated by any sport governing body.

The survey on the athletes and support staff revealed that genetic testing is already taking place in sport (in Europe) and to higher extent as previously reported by Varley et al. (2018), the main reason for this being that the other study was done approximately four years earlier and only in the UK. The survey showed that athletes were keen on taking the genetic testing if asked by the coach/governing bodies and also curious about knowing if they had the gene variants associated with sport performance, injury susceptibility and nutrition optimization. Moreover, they were not as concerned about the ethical issues as expected and previously reported (Varley et al., 2018). The only limitation of this study is the small number of the responses, however, it has to be said that it is difficult to survey the elite athletes; and that the conclusions made from the survey go in line in majority of the answers with the previously published study (Varley et al., 2018).

The analysis of practices of DTC companies that offer sport genetic testing showed several inconsistencies and differences among the companies, including the number of genetic variants that are tested, the report or results that are given to the customer, the cost of the sport genetic testing, the explanation of the genetic testing itself, the information was not complete for all the DTC companies, etc., which raised serious doubts about which company should be selected for the genetic testing in sport. Based on my scientific background the whole genome sequencing is recommended, basically as it sequences the whole athlete’s genetic information and afterwards the proper personnel analyses the selected gene variants. However, this represents some challenges as for example employing data analyst or bioinformatician, data protection and confidentiality, and how to translate the genetic information into real sport practices.

The case study of different nations, leagues and clubs revealed that each of them (Egypt, China, and FC Barcelona) uses different companies, probably different set of gene variants are being looked at (there was no information about this actually), and different regimes of training and
nutrition prepared based on the genetic analysis done (also no information available about it). In the case of NFL, the genetic testing in sport is prohibited at the moment by the collective bargaining agreement. Based on the case studies done, it was concluded that there is no consensus nor guidelines available for the genetic testing in sport, not even by the nations or clubs that are already doing it, again indicating a need for the regulation by the sport governing bodies.

Next, the cost-efficiency analysis was done of implementing the genetic testing in sport, which indicated that if the genetic testing would only impact the injury rates and time of injury of the players by reducing them by 10% that it would return the costs of investment (of implementing the genetic testing into the club), clearly implying that it should be implemented, especially by the clubs that have players with high salaries like for example Real Madrid. The drawback of this analysis was that there were no actual numbers on how much would genetic testing really impact on injury rates and time, or other variables that could improve the cost-efficiency analysis and give more realistic view, however, this did not change the conclusion that the clubs in general would benefit from implementing the genetic testing into their sport practices and protocols.

Based on the literature and field analysis done, the strategy of implementing the genetic testing on all level was proposed. Specific guidelines were made for the IOC/WADA as they are one of the main sport governing bodies, from which other NOC, international and national sport federations, league and clubs adopt the guidelines, propositions and plan actions. The concept of the Athlete Genetic Passport was proposed in line with WADA’s Athlete Biological Passport, which can be easily implemented into already existing IOC/WADA infrastructure, guidelines, and protocols (detailed plan of action how to do it was provided). In addition, WADA can use the Athlete Genetic Passport for its gene anti-doping measures. The same guidelines and plan of actions could be followed by any sport governing body (in the case IOV/WADA do not approve it). In addition, the guidelines for leagues and federations that do not want to adopt the measures proposed to IOC/WADA were given. And lastly, also the plan of action on the club’s level was proposed in the case that there is no regulation (as at the moment) and the club would like to implement the genetic testing. In my opinion, any sport governing body can easily implement genetic testing in sport following the proposed guidelines.

This study is the first one of the kind, there is no similar study to my knowledge, which gives it additional merit as it was difficult to find all the necessary information, especially to contact the people from the sport industry (some unfortunately did not respond). Moreover, the study clearly identifies the key considerations to have in mind when implementing the genetic testing in sport and gives clear indications on how to do it.
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ANNEXES

GLOSSARY OF TERMS

ACE – Angiotensin-converting enzyme; an enzyme involved in catalyzing the conversion of angiotensin I into a physiologically active peptide angiotensin II. Angiotensin II is a potent vasopressor and aldosterone-stimulating peptide that controls blood pressure and fluid-electrolyte balance (Genecards, Available at: https://www.genecards.org/cgi-bin/carddisp.pl?gene=ACE). Some variants of this gene have been related with improvements in performance and exercise duration in a variety of populations.

ACTN3 – α-actinin-3; which is present in fast-twitch muscle fibers. These types of skeletal muscle are responsible for generating force at high velocity. Gene variants of ACTN3 can affect power and endurance ability (Genomic Express, Available at: https://www.genomicexpress.com/inherited/49-inherited-traits/83-inherited-traits-athletic-performance-actn3-information).

Allele – An allele is one of two or more versions of a gene. An individual inherits two alleles for each gene, one from each parent. If the two alleles are the same, the individual is homozygous for that gene. If the alleles are different, the individual is heterozygous (NIH, Available at: https://www.genome.gov/genetics-glossary/Allele).

BRCA1 – mutations in this gene predispose to the breast and ovarian cancer and it is used for genetic screening for these two cancers in health care.

Chromosome – A chromosome is an organized package of DNA found in the nucleus of the cell. Different organisms have different numbers of chromosomes. Humans have 23 pairs of chromosomes; 22 pairs of numbered chromosomes, called autosomes, and one pair of sex chromosomes, X and Y. Each parent contributes one chromosome to each pair so that offspring get half of their chromosomes from their mother and half from their father (NIH, Available at: https://www.genome.gov/genetics-glossary/Chromosome).

Chromosomal disorder – changes in the number or structure of entire chromosomes.

CRISPR technology – A technology used for genome editing, i.e. for changing individuals (or organism’s) DNA sequence.

DNA – constitutes the molecules inside cells that carry genetic information and pass it from one generation to the next.

Epigenetic regulation – Epigenetic processes regulate gene expression by modulating the frequency, rate, or extent of gene expression in a way that does not entail a change in the DNA sequence (Reactome, Available at: https://reactome.org/content/detail/R-HSA-212165).
Gene – A DNA sequence that encodes the information for a gene product that is RNA or protein. Gene is a functional unit of heredity (NIH, available at: https://ghr.nlm.nih.gov/primer/basics/gene).

Gene expression – Process by which the information encoded in a gene is used to direct the assembly of a protein molecule. The cell reads the sequence of the gene in groups of three bases. Each group of three bases (codon) corresponds to one of 20 different amino acids used to build the protein (NIH, Available at: https://www.genome.gov/genetics-glossary/Gene-Expression).

Gene mutation – A gene mutation is a permanent alteration in the DNA sequence of a gene, in a way that the sequence differs from what is found in most people (NIH, available at: https://ghr.nlm.nih.gov/primer/mutationsanddisorders/genemutation).

Gene variation – Acceptable differences in the DNA sequence that normally occur within the population; i.e. between individuals.

Genetic loci – Specific, fixed position on a chromosome where a particular gene or genetic marker is located (Wikipedia, Available at: https://en.wikipedia.org/wiki/Locus_(genetics)).


Genetic trait – A trait is a specific characteristic of an organism. Traits can be determined by genes or the environment, or more commonly by interactions between them. The genetic contribution to a trait is called the genotype. The outward expression of the genotype is called the phenotype (NIH, Available at: https://www.genome.gov/genetics-glossary/Genotype).

Genome – Complete genetic material of the organism.

Genotype – Full hereditary information of an organism, all the genetic information.

Myostatin – Protein found almost exclusively in muscles used for movement (skeletal muscles), where it is active both before and after birth. This protein normally restrains muscle growth, ensuring that muscles do not grow too large.

Phenotype – An organism's actual observed properties, such as morphology, development, or behavior. This distinction is fundamental in the study of inheritance of traits and their evolution (Wikipedia, available at: https://en.wikipedia.org/wiki/Genotype%E2%80%93phenotype_distinction).

Sickle Cell Trait – People who inherit one sickle cell gene and one normal gene have sickle cell trait. It affects the blood cells, causing blood cells to be 'sickle' shaped.

SNP – Single nucleotide polymorphisms, acceptable variances in the DNA, not to be mistaken with the gene mutation.
Transcriptional regulation – Regulation of the conversion of DNA into RNA (transcription); i.e. activity of gene (or its transcription) can be switched on or off.

WADA – World anti-doping agency.

WES – Whole Exome sequencing; pieces of DNA that hat provide instructions for making proteins are called exons. Together, all the exons in a genome are known as the exome, and the method of sequencing them is known as whole exome sequencing. This method allows variations in the protein-coding region of any gene to be identified, rather than in only a select few genes. Because most known mutations that cause disease occur in exons, whole exome sequencing is thought to be an efficient method to identify possible disease-causing mutations (NIH, Available at: https://ghr.nlm.nih.gov/primer/testing/sequencing).