The current use, and opinions of elite athletes and support staff in relation to genetic testing in elite sport within the UK

AUTHORS: Ian Varley¹, Seema Patel², Alun G. Williams³, Philip J. Hennis⁴

¹ Sport, Health and Performance Enhancement Research Centre, Nottingham Trent University, Clifton Lane, Nottingham, NG11 8NS, UK
² Centre for Sports Law, Nottingham Law School, Nottingham Trent University, Goldsmith Street, Nottingham, NG1 5LP, UK
³ Sports Genomics Laboratory, Department of Exercise and Sport Science, Manchester Metropolitan University, Crewe Green Road, Crewe, CW1 5DU, UK
⁴ Centre for Health, Exercise & Active Living (HEAL), Department of Exercise and Sport Science, Manchester Metropolitan University, Crewe Green Road, Crewe, CW1 5DU, UK

ABSTRACT: The purpose of the study was to investigate the current use of genetic testing in UK elite sport and assess how genetic testing might be received by those employed in elite sport. Seventy-two elite athletes and 95 support staff at UK sports clubs and governing bodies completed an online survey of 11 questions concerning their experience of genetic testing and beliefs regarding the use of genetic testing in sport. Genetic testing related to sports performance and injury susceptibility is conducted in UK elite sport, albeit by a relatively small proportion of athletes (≤17%) and support staff (≤8%). Athletes and their support staff agree that genetics are important in determining elite status (≥79%) and appear willing to engage in genetic testing for individualising training to improve sport performance and reduce injury risk. Opinion was divided on whether genetic information should be used to identify talented athletes and influence selection, eligibility or employment status. Genetic testing for sports performance and injury susceptibility occurs in UK elite sport, however it is not commonly conducted. There is a belief that genetics is an important factor in determining an athlete and there is a willingness to engage in genetic testing for sports performance and injury susceptibility.

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INTRODUCTION

Current state of the genetics-performance literature and its relevance to practice

The extent to which genotype can influence sporting performance and how the identification of specific genetic variants can complement existing coaching and medical practices continues to be questioned [1, 2]. On the one hand, genetic factors account for a substantial portion of the variability of many physical performance and injury susceptibility traits across a population [3, 4] and hundreds of studies have identified associations between specific genetic loci and favourable athletic performance and injury risk [4, 5, 6]. However, although genotype-performance phenotype associations have been identified, the extent to which genetic variations influence a given trait has proved difficult to quantify [7]. Moreover, while genotype-phenotype associations have been shown, little convincing literature exists [8] demonstrating the ability of certain genes to predict training responses, athletic success or injury susceptibility.

Genetic testing in practise

Despite the lack of predictive research, the value of genetic testing in athletes should not be underestimated. It is thought that genetic information shown to be associated athletic prowess and injury risk can be translated into clinical settings and benefit the treatment of disease in the general population [9]. This could lead to better healthcare provisions, improved patient choice, increased research and effective treatment for disease in the future [11]. In light of this potential, genetic testing is becoming easily accessible to a worldwide population and genetic test services are now offered by many companies directly to the consumer (DTC) [2]. Some DTC companies target elite sport with marketing material and provide client-specific information supposedly related to their adaptability to training and injury susceptibility based on their genotype. However, little is known about the current use of genetic testing in elite sport and what information is available tends to be anecdotal [12].
1. Investigate the current extent of genetic testing in elite sport in the UK.
2. Assess the beliefs of elite athletes and support staff about genetic testing in sport.
3. Assess how genetic testing might be received by people employed in elite sport.

**MATERIALS AND METHODS**

Elite athletes and support staff at UK sports clubs, governing bodies and those with known affiliations to these associations were contacted via email and by word of mouth from pre-existing personal and professional contacts. During the survey recruitment stage, the authors did not contact pre-existing associates within elite sport, who may have already taken part in previous work by the authors research-
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This method of recruitment prevented bias and potential artificial exaggeration of the number of athletes that have taken part in genetic testing for research purposes. Individuals associated with a range of sports were approached in order to represent UK sport as a whole. Participants were asked to provide informed consent and complete a survey related to their views on genetic testing in elite sport. The internet-based survey (Bristol Online Survey, Bristol, United Kingdom) was completed anonymously and comprised of 11 multiple choice questions (Table I and II) related to genetic testing in sports performance and injury susceptibility. These questions can be broadly separated into the following topics: current use, beliefs, potential utility of genetic testing in sport. Respondents were also given the opportunity to elaborate on their answer. These qualitative data in the form of additional comments further supported and enriched the quantitative data gathered from the question.

Professional athletes were classified as elite due to their full time participation in sport; non-professional athletes were classified as elite if they regularly competed at international or national level. Support staff met the inclusion criteria if their role involved working in a support capacity with elite athletes. Prior to taking part in the study, each participant provided informed consent. Ethical approval

| TABLE II. Responses from elite athletes to questions related to the use of genetic testing in sport. |
|---------------------------------------------------------------|
| Elite athletes (n=72) | Response |
|---------------------------------------------------------------|
| Current use of genetic testing in sport | |
| Have you ever engaged in genetic testing in relation to sports performance? | Yes, n (%) | No, n (%) | Unsure, n (%) | No response (%) |
| Have you ever engaged in genetic testing in relation to injury susceptibility? | 10 (13.9) | 60 (83.3) | 2 (2.8) | 0 (0) |
| If you have engaged in genetic testing can you recall which genes were sampled for in relation to sports performance? | 3 (4.2) | 68 (94.4) | 1 (1.4) | 0 (0) |
| If you have engaged in genetic testing can you recall which genes were sampled for in relation to injury susceptibility? | Yes: 0 (0) | I was informed but can't remember: 8 (80) | I wasn't informed: 2 (20) | 0 (0) |
| Beliefs about genetic testing in sport | |
| Elite athletes are elite due to? | Genetics: 1 (1.4) | Training: 15 (20.8) | Combination of both: 56 (77.8) | 0 (0) |
| Potential for the use of genetic testing in sport | |
| If asked by your coach/governing body would you take a genetic test in order to gain information that may be related to sports performance? | 57 (79.2) | 8 (11.1) | 6 (8.3) | 1 (1.1) |
| If asked by your coach/governing body would you take a genetic test in order to gain information that may be related to injury susceptibility? | 55 (76.4) | 11 (15.3) | 6 (8.3) | 0 (0) |
| Would you want to know if you had a genetic variation that might be associated with sports performance? | 58 (80.6) | 5 (6.9) | 9 (12.5) | 0 (0) |
| Would you want to know if you had a genetic variation that might be associated with injury susceptibility? | 61 (84.7) | 6 (8.3) | 5 (6.9) | 0 (0) |
| Should genetic information be considered to determine selection/eligibility/employment in your sport? | 20 (27.8) | 34 (47.2) | 18 (25.0) | 0 (0) |
| Would genetic testing be a valuable tool in the talent identification process in your sport? | 48 (66.7) | 16 (22.2) | 8 (11.1) | 0 (0) |
was granted by the Nottingham Trent University Non-Invasive Ethical Review Committee and was in accordance with the ethical standards of the Helsinki Declaration.

Frequency based descriptive analysis was performed on the completed surveys.

RESULTS

In total, 95 support staff (70 male; 24 female; 1 didn’t provide this information) and 72 elite athletes (59 male; 13 female) completed the survey between December 2015 and July 2016. Support staff were employed in various sports including: football (n=21), swimming (17), rugby (6), athletics (5), cricket (5), figure skating (3), speed skating (2), hockey (2), gymnastics (2), table tennis (1), rowing (1), cycling (1) with some servicing multiple sports (29). The specific role occupied by those surveyed was diverse and included, but was not limited to: Coach (n=24), Sport scientist (18), Physiotherapist (17), Strength and condition coach (10), Doctor (7) and Nutritionist (7). Athletes surveyed completed in various sports: rugby (30), speed skating (12), volleyball (11), hockey (8), cricket (5), swimming (2), canoe slalom (2), athletics (1) and football (1). Support staff and athlete responses to the questions are displayed in Table I and II.

Current use of genetic testing in sport

The use of genetic testing within elite sport is rare in the UK with most support staff not aware of their club or organisation having ever implemented genetic testing in relation to sports performance (92%) or injury susceptibility (91%). Similarly, the majority of athletes surveyed had never engaged in genetic testing for sports performance (83.3%) or injury susceptibility (94%). The responses from the survey show that genetic testing is being conducted in elite sport within the UK for sports performance (Support staff 2%; Elite athletes 14%) and injury susceptibility (Support staff 5%; Elite athletes 4%). However, only 2 of the 5 support staff who had reported genetic testing for injury could recall the genes tested, while no athletes could recall the genes tested for performance or injury susceptibility (Table I). Collagen-related genes (presumably because of some prior evidence of association with tendon or ligament injury risk [17]) were reported by the respondents that could recall the specific genes analysed.

Opinions about genetic testing in elite sport

The overwhelming majority of the support staff surveyed believed that genetics and environmental factors have a role in athletes becoming elite (97%), which was supported by the additional qualitative comments. One participant commented that, “Natural talent is the key but quality training and commitment is needed”. Most of the athletes surveyed also believed that genetics and environmental factors played a role in becoming an elite athlete (79%) with one athlete expressing that, “Genetics play a big part, but nothing substitutes hard work”.

The majority of support staff reported that there is a place for genetic testing in their sport to inform about sports performance (Yes: 63%; No or unsure: 37%) and injury susceptibility (Yes: 72%; No or unsure 26%). The comments reflect practical reasons for the positive response, with genetic information considered to potentially be a “key element in signing-on medicals”, beneficial for “reducing injury risk” and such information could “have a massive financial impact in reducing medical bills and paying wages for injured players”. However, other comments were more cautious, stating that, “I don’t feel this is a black or white issue” and it should be used for the “purposes of research, but not for any selection/exclusion purposes”.

Potential employment of genetic testing in sport

The majority of support staff reported wanting to know if an athlete working with them had a genetic variation that might be associated with sports performance (61%) or injury susceptibility (78%). Similarly, most athletes would like to know if they had a genetic variation that was associated with sports performance (81%) or injury susceptibility (85%). Three common factors appear to influence this opinion. Firstly, comments centred around the use of genetic testing for research purposes, “from a research perspective, to identify associations and inform modelling and computational analysis to elucidate the role of genetics in trainability/injury/performance”. Secondly, curiosity seemed to drive this positive response. For example, one athlete responded “it would be good to know what my strengths and weaknesses are”. The third factor concerned the use of genetic information to enhance performance through individualised training and injury prevention, for instance one athlete commented, “it would help with prehab trying to avoid the injury”. Although the responses were largely positive, some support staff were cautious about genetic testing which is reflected in comments such as, “As long as it was relatively non-invasive, ethical and safe” and not used to “exclude athletes”.

The majority of support staff surveyed thought that genetic information should not be used to determine athlete selection, eligibility or employment (64%). The comments imply that this is driven by ethical concerns about discrimination: “Wouldn’t this be discrimination?” and “Arguably immoral/unethical…”. At the same time, it was opined that “genetic information can be used as a tool within the process but not as a determining factor”. Furthermore, testing may “help to determine which events their genetics are best suited? Eg Sprint or distance”. Athletes’ opinions were also divided on the use of genetic information to determine athlete selection, eligibility or employment status, which is reflected by one comment stating, “I am split between the ethics of the situation”. Almost a third of athletes surveyed (28%) thought that genetic information should be used, while 47% disagreed and 25% were unsure. The divided opinion among athletes is reinforced by the spectrum of comments, ranging from positive support that, “genetic advantages should be considered when selecting athletes for sports” to more equality-
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minded responses, for example, “everybody should be treated equal regardless of their genetic information”.

Opinion was almost evenly divided amongst support staff on the use of genetic testing for talent identification (yes = 48%; no or unsure = 51%), which is evidenced by the diverse range of comments. Whilst it was expressed that, “better genetics testing could produce and make better athletes,” other participants noted that it could be, “valuable but not the only method.” and that, “it may be detrimental if coaches forget the historical uninformed principles and miss talent not identified through genetic testing”.

Conversely, a large proportion of athletes thought genetic tests would be a valuable tool in the talent identification process (67%), perceiving this to be “in the best interest of the sport and athlete to know this to ensure success...” and some commented on its use to decide an athlete’s optimal role within a team or sport ‘it could determine your position’.

DISCUSSION
Main findings
To the authors’ knowledge this is the first study to quantify the use of genetic testing in elite sport, albeit in a relatively small number of elite athletes and support staff. The present study shows that despite anecdotal evidence of the use of genetic testing, the prevalence of genetic testing within elite sport in the UK is low. There is a belief amongst elite athletes (~80%) and their support staff (nearly 100%) that genetics is important in determining athletic status and that genetic profiling could be useful for predicting sporting performance and injury susceptibility. This belief is consistent with published data in the field [18, 19, 19, 21]. There were mixed responses to how and why genetic testing might be used in a sporting context. Comments made indicated varied levels of knowledge, awareness and understanding about the role of an individual’s genotype and its association with performance and injury susceptibility.

Current use of genetic testing in sport
In the present study, genetic testing was shown to occur in elite sport within the UK, although it is not common. This finding supports anecdotal testimonies suggesting that genetic testing has been used in sport within the UK and worldwide. In 2008, an English football club approached a scientist about “the possibility of screening players to discover whether they have a genetic predisposition to athletic excellence” [12]. The DTC company 23andMe reportedly conducted genetic analyses of National Football League players to investigate how genes impact upon athletic performance [22]. In advance of the 2016 Olympics in Rio, there were claims by another DTC company that it was working with elite UK athletes during their competition preparations [23]. Despite the number of respondents undergoing genetic testing being low, the fact that our study and anecdotal claims such as those we mention have identified that some athletes and support staff are using it in attempts to inform practice raises questions about how genetic information is being used. In the USA, genetic testing in Major League Baseball is conducted to verify parentage with the hope of ensuring an athletes’ age is not falsified [24, 25], although there is speculation that genetic information is being retained to determine a player’s susceptibility to disease and injury [26]. Genetic testing for risk of disease and injury in athletes without appropriate consent raises a number of moral and ethical issues. It is important that genetic testing is conducted in accordance with good practice and an appropriate ethical and legal framework to ensure the safeguarding of athletes and their families.

Opinions about genetic testing in elite sport
Elite athletes and their support staff in the present study overwhelmingly agreed with the scientific consensus that both genetics and environment play a role in becoming an elite athlete [18, 19, 20, 21]. This highlights that both athletes and support staff are generally informed in that regard and have appropriate awareness of the factors that are involved in becoming an elite athlete, despite popular literature [28, 29] downplaying the role of genetics in becoming elite in mass participation sports. However, that narrative in the popular literature might be responsible for the belief of ~20% of athletes that success in sport is entirely due to training. It is understandable why that belief might be a useful one for athletes, from a self-efficacy perspective, but that does not make it true.

Potential employment of genetic testing in sport
There were largely positive responses to the questions related to whether athletes would be keen to know about any genetic variations they had, and whether support staff would want to know their athletes’ genetic information. This shows a willingness to engage in and undergo genetic testing for genes related to sport performance and injury susceptibility. The inclination to access genetic information is probably somewhat at odds with the general view of the scientific community. The use of genetic testing in an applied sport setting would present some theoretical challenges. Despite continued studies in the area, and some genetic variations appearing to be reasonably strong candidates for influencing sports performance (e.g. ACE II associated endurance athletes, ACTN3 RR associated with power athletes) [7] the exact genes of interest and the impact of individual SNPs on sport performance remains far from conclusive [1, 2]. Moreover, published literature has not demonstrated effective practical applications of genetic testing for sport performance or injury susceptibility. The inherent difficulties in assessing gene-environment interactions, the almost certainly polygenic nature of athletic prowess, and the lack of understanding about gene-gene interactions and gene hierarchy [30] suggest very limited applied use in assessing current athletic excellence. The potential for more substantial and evidence-based future applications nevertheless remains.

Selection and employment
Athletes and support staff surveyed had mixed views on whether genetic testing should be used for selection and employment. The
suggestion of some respondents that genetic testing might be useful for selection and employment in sport may outline a lack of awareness or a disregard of the ethical and legal issues that currently surround this area. Genetic testing being used in this manner could lead to the potential misuse of genetic information, and therefore result in the genetic discrimination of an athlete which could have legal implications. On the other hand, sport is discriminatory in many respects, with athletes routinely selected for employment according to physical dimensions and performance capability – traits that have strong genetic components and which would not be considered suitable grounds for selection in most other forms of employment. Internally, whilst discussions have taken place within sport governing bodies and sport organisations about the potential for genetic testing [16], little guidance currently exists about the best way to ensure that sport practices are ethical, fair and consistent with equality rights. Currently, specific legislation, such as the Genetic Information Non-discrimination Act (GINA) in the USA seeks to protect and advise against individuals being excluded from employment and insurance on the basis of their genetic information. The compatibility of GINA with sport is not clear [22]. Nevertheless, the use of genetic information to make determinations about selection and employment is potentially incompatible with equality principles and fundamental human rights. In Australia, a comprehensive inquiry into the protection of human genetic information, led by the Australian Law Reform Commission, considered the use of genetic testing/information/therapy in sport [31]. It was recommended that policies and guidelines are developed by sports authorities on the use of genetic information in sport.

Talent identification
Opposing responses related to the usefulness of genetic testing in the talent identification process were received by elite athletes and support staff. The diversity of responses to this question may highlight the stark differences in awareness in relation to the role of genetic testing within elite sport. As previously mentioned, there are some associations between genetic characteristics and aspects of sporting prowess, but no literature shows an ability to identify a talented athlete based on genetic characteristics with anything even approaching convincing sensitivity and specificity [2, 32]. As only modest odds ratios (1.35 \textit{ACE} \textit{V}D; 1.21 \textit{ACTN3 R577X}) exist between the most prominently analysed genetic variations and sports performance [7], it is inevitable that both false negatives and false positives would be reported when trying to predict future sporting prowess [33]. The use of genetic testing for talent identification is not only questionable on scientific grounds, but also raises moral and ethical issues such as restricting child development/progression and their right to an open future [13]. At present, there is no predictive benefit for athletic performance from the identification of single nucleotide polymorphisms [2]. Furthermore, studies investigating the genotype-phenotype associations in athletic populations are often underpowered due to relatively small sample sizes [5] and have limited relevance to populations beyond the ethnicity of the studied cohort.

Limitations
Although our sample has canvassed opinion from athletes and their support staff across a wide range of sports in the UK, it is acknowledged that the sample size is relatively small, the majority of respondents were male and the views received are not those of the entire athletic community. That said, we are satisfied that the responses collected offer a good perspective of elite athlete and support staff opinions within the UK due to the number of surveys completed, and as some of the support staff are responsible for multiple sports they have governance over a large number of athletes.

CONCLUSIONS
In conclusion, genetic testing takes place within UK sport, elite athletes and their support staff believe that genetics is an important factor in sport performance and have an appetite to engage in genetic testing. Genetic testing has the potential to be implemented widely in UK sport and it remains to be seen if the increase in availability of DTC genetic testing [2] will increase its utilisation. However, it is important that research investigating genetic associations with sporting traits improves in order to provide of the necessary evidence base on which to base such genetic testing. A greater degree of education is encouraged to improve awareness of the potential application of genetic testing, its significant current limitations and the wider legal and ethical implications of genetic testing for talent identification and selection.

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