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The frequency of, and attitudes towards, genetic testing amongst athletes and support staff

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Abstract

Recently, knowledge of the genetic influence on the attainment of elite athlete status, along with aspects such as training adaptations and injury risk, has grown. At present, there are various direct-to-consumer genetic tests targeted at athletes. Here, we aimed to determine to prevalence of, and attitudes towards, genetic testing in a group of athletes, coaches, and support staff. 243 participants (110 athletes and 133 support staff) took part in an internet-based survey. 51% of athletes had competed internationally, and 54% of support staff reported that their main contact time was with international athletes. The frequency of genetic testing was relatively low, with 10% of athletes and 11% of support staff stating they had utilised such a test. Overall, the majority of athletes and support staff felt that genetics played a role in the attainment of sporting success and training adaptation. The main barriers to undertaking genetic testing were a lack of awareness, high cost, and a lack of scientific evidence. The majority (73% of athletes and 64% of support staff) of participants utilising genetic tests found the information useful.

Key Words: elite athlete, genetics, talent identification, direct-to-consumer

1. Introduction

Over the last few decades, research has illustrated how genetic variation may influence the likelihood of attaining elite athlete status (Ahmetov, Egorova, Gabdrakhmanova, & Fedotovskaya, 2016; Bray et al., 2009), the magnitude of training-induced adaptive responses (Bouchard, 2012; Pickering, Kiely, Suraci, & Collins, 2018; Timmons, 2011), and injury risk (Goodlin, Roos, Roos, & Kim, 2015). Recent reviews suggest that at least 155 genetic markers are associated with elite athlete status and/or fitness phenotypes (Ahmetov et al., 2016; Bray et al., 2009; Bouchard, 2012). As a result of these findings, a number of companies now market direct-to-consumer genetic testing to athletes and fitness enthusiasts (Webborn et al., 2015). Whilst there are vast differences in the quality and claims of these companies, the current scientific consensus is that “genetic tests, based on current knowledge, have no role to play in talent identification or the individualised prescription of training to maximise performance” (Webborn et al., 2015). Similar sentiments have been echoed by the Australian Institute of Sport (Vlahovich, Fricker, Brown, & Hughes, 2017). Furthermore, the provision of genetic testing raises a number of potentially contentious ethical issues (Camporesi & McNamee, 2016). Nevertheless, some of the authors of these consensus statements remain hopeful that the evidence-base may soon support practical applications of genetic tests. Williams, Day, Lockey, Heffernan, and Erskine (2014), for example, predicted that training modifications based on genetic information, both to reduce injury risk and increase training adaptations, would soon be evidence-based, and that in the future talent identification processes could be informed by genetic information; some such innovations, in fact, are objectives of the Athlome Project Consortium (www.athlomeconsortium.org/about/). Indeed, some recent research has provided support for the contention that genetically guided training and nutritional advice for athletes may be

advantageous, but more research and replication are clearly required (Pickering et al., 2018; Jones et al., 2016; Pickering & Kiely, 2018).

Athletes and sporting teams tend to be early adopters of new technologies as they seek innovative and novel means to gain an edge over their competitors (McNamee, Coveney, Faulkner, & Gabe, 2018). In relation to genetic testing, this is no different; over ten years ago, the journal *Nature* reported that the Manly Sea Eagles, an Australian Rugby League team, had genetically tested a number of players in order to inform training programme design (Dennis, 2005). Since then, this practice has grown, with a number of sporting teams currently known to have used the results of genetic tests to better inform holistic athlete management and talent identification (Edwards, 2018; Singer, 2017; Miller, 2016). Indeed, it was announced in 2014 that Uzbekistan's National Olympic Committee was involved in a genetic testing programme to identify future elite athletes (Synovitz & Eshanova, 2014). As such, there is an evident mismatch between the general scientific consensus and current practice.

At present, the true prevalence of genetic testing in elite sport is largely unknown. Many organisations and/or clubs wish to retain confidentiality, potentially in part to retain an advantage over competitors, and potentially because such testing may be negatively interpreted by the public and media. Recently, Varley, Patel, Williams, and Hennis (2018) conducted an online survey of 72 elite athletes and 95 support staff based within the UK. Their results indicated that fewer than 17% of elite athletes had undergone genetic testing, although most athletes and coaches (79%) indicated that they were willing to engage in such tests. However, in that online survey, respondent numbers were limited, and the diversity of sports represented was low. In directly addressing this information deficit, the present study

was designed to a) determine the prevalence of genetic testing in sports, and b) advance our understanding of the relevant prevailing beliefs and opinions of athletes, sports coaches, sports scientists, and sports medicine providers, as to the utility of genetic testing in sports. In addition, secondary aims were to a) determine whether teams or individuals who had utilised genetic testing found the acquired information relevant and useful, and b) identify the perceived barriers amongst athletes and support staff towards genetic testing.

2. Material and Methods

2.1 Experimental approach to the problem

This study utilised an online questionnaire to determine the prevalence of, and attitudes towards, genetic testing amongst athletes and coaches.

2.2 Participants

243 individuals gave consent to take part in this study and completed the survey, comprising of 110 current or former sporting participants (45.3%) and 133 members of support staff (54.7%). The majority of respondents (76.5% of athletes and 84% of support staff) were male. The most common sport, for both athletes (66%) and support staff (40%), was athletics (Track & Field). Table 1 below lists the sports with the frequency of respondents.

Primary Sport	Athlete	Support Staff
Athletics (Track & Field)	66%	40%
Football	3%	13%
Rugby (League/Union)	2%	15%
American Football	3%	2%
Basketball	0%	4%
Swimming	1%	2%
Racquet Sports	0%	2%
Winter Olympic Sports	7%	3%
Other	18%	19%

Table 1. Frequency of different sports within this survey sample

Sports in the “other” category included rowing (4% of athletes; 2% support staff), combat sports (4% of athletes; 3% support staff), volleyball (1% of athletes & support staff), field hockey (1% of athletes & support staff), cycling (4% of athletes & support staff), and triathlon (1% of athletes).

18% of the athletes taking part in this survey had competed at the Olympic Games or World Championships, and a total of 51% had represented their country. A further 22% had competed at the highest level within their country, such as the national championships or top league. The vast majority (78%) were from the UK and Ireland; 9% were from the US, and 6% from other European Countries.

Within the support staff cohort, 18% most frequently worked with Olympic or World Championships competitors, with 36% of respondents working with international athletes, and a further 30% working with athletes who had competed at the highest level within their

country. Most of the support staff (53%) were sports coaches, 18% were strength and conditioning coaches, 12% were sports scientists, and 5% were physiotherapists. Most (62.5%) were from the UK and Ireland, with a further 11% from other European countries, 7.5% from the US, and 5% each from Australia & New Zealand and North America (excluding the US).

2.3 Procedures

Prior to the commencement of data collection, ethical approval was granted by the University of Central Lancashire Ethics Board in accordance with the Declaration of Helsinki. Athletes and support staff were recruited via social media accounts from both authors. Potential participants were provided with a link, which directed to the survey home page; this page contained both the participant information and informed consent forms. Participants were asked to provide informed consent, and complete an online survey related to both their views and use of genetic testing within sport. The survey was comprised of 42 questions, with participants directed to specific questions based on their previous answers. The majority of questions were multiple choice, although two required a written answer. The questions broadly fit into four parts: 1) demographic data, 2) beliefs about the effects of genetics in sport, 3) prevalence of genetic testing in sport, and 4) the utility of genetic testing in sport. Following the completion of the questionnaire, frequency-based descriptive analysis was carried out.

3. Results

3.1 Beliefs around the impact of genetics on sporting phenotypes

Participants were asked about their opinion as to the relative contribution of genetics to various sporting phenotypes. These results are shown in table 2.

Table 2. Athlete and Support Staff opinions as to the impact of genetics on sporting phenotypes.

		Athletes	Support Staff
What impact do you think an individual's genetic make-up has on their chances of being an elite sportsperson?	None	2%	1%
	Minimal (<25%)	6%	8%
	Somewhat (25-75%)	59%	69%
	Almost Entirely (75%+)	33%	22%
What impact do you think an individual's genetic make-up has on their sporting/fitness improvements following exercise?	None	3%	2%
	Minimal (<25%)	9%	7%
	Somewhat (25-75%)	59%	70%
	Almost Entirely (75%+)	29%	21%
What impact do you think an individual's genetic make-up has on their nutrition requirements?	None	2%	3%
	Minimal (<25%)	23%	27%
	Somewhat (25-75%)	57%	59%
	Almost Entirely (75%+)	18%	11%

3.2 Prevalence of genetic testing within sport

10% (n = 11) of the athletes had utilised a genetic test that was targeted at sports performance, and 11% (n = 14) of support staff respondents stated that they had utilised genetic testing within their organisation.

3.3 Attitudes towards genetic testing

The 90% of athletes and 88% of support staff respondents who had not utilised genetic testing were asked for their reasons for having not done so. These results are reported in table 3.

Table 3. Most common reasons cited for not utilising genetic testing (respondents able to select multiple answer options)

	Athlete	Support Staff
Too expensive	31%	41%
Unaware that genetic testing was possible	49%	25%
Insufficient evidence for its use	21%	39%
Concerns around data protection	1%	6%
Concerns about negative press coverage	0%	2%
Ethical Considerations	4%	19%

Of those who had not utilised genetic testing, 10% of athletes and 5% of support staff envisioned doing so within the next year, 26% of athletes and 28% of support staff within the next 5 years, and 11% of athletes and 29% of support staff within the next 10 years. 53% of athletes and 38% of support staff believed they would never utilise genetic testing. Table 4 (below) provides the most frequent responses to the question “what would cause you to use genetic testing?”

Table 4. Responses to the question “what would cause you to use genetic testing?” (respondents able to select multiple answer options)

	Athletes	Support Staff
Publication of peer-reviewed case studies	29%	63%
Greater number of intervention studies	14%	36%

Publication of randomised controlled trials	26%	46%
More athletes/teams using it	48%	18%
Players requesting it	N/A	25%
Direct approach from genetic testing company	N/A	18%
Increased awareness of the product	35%	N/A
Lower Price	48%	36%

3.4 Experience of genetic testing

Of the athletes who had utilised genetic testing, the most common reason cited (n = 4) was to inform training programme design, followed by general interest (n = 2), a desire to identify the most appropriate sport to compete in (n = 1), and to acquire insights potentially informing injury prevention and nutritional practices (n = 1). Eight of the eleven (73%) athletes who had undertaken a genetic test reported that the information they received was useful. Of the three who did not find the information useful, the main reason (n = 3) was that the information provided was considered too generic, and not adequately targeted at sports people. Most athletes (73%; n = 8) reported that the results of their genetic test were easy to understand, with 8 receiving after-testing follow-ups from the testing company to provide them with additional information. The majority of athletes (n = 8) who had undertaken genetic testing reported they had made changes based on the results of the test.

Similarly, of the 14 support staff members that had utilised genetic testing within their organisation, 50% (n = 7) had done so primarily to inform training programme design, 21% (n = 3) for injury prevention, and 14% (n = 2) to guide nutritional interventions. Interestingly, 7% had done so as a screen for disease risk. No support staff had utilised genetic testing for talent identification purposes. 64% (n = 9) of support staff who had used a genetic test found

the information provided useful; of those who didn't, the main reason ($n = 4$) was that the results were too generic. Most (86%; $n = 12$) found the information provided easy to understand, and 64% ($n = 9$) received follow up information from the testing company/institution. 64% ($n = 9$) of support staff who utilised a genetic test within their organisation made changes based on the test results, with 100% of respondents stating they made changes to their athletes training programmes, 86% ($n = 12$) to their diet, 71% ($n = 10$) to their recovery, and 42% ($n = 6$) to their lifestyle.

All of the genetic testing reported by athletes in this study was conducted by commercial companies (brands/names not specified). Conversely, for support staff respondents, 14% ($n = 2$) of the genetic testing had been carried out by a university or academic institution, with the remaining 86% ($n = 12$) coming from commercial companies.

4. Discussion

This study, which surveyed high level athletes and support staff from across the globe, suggests that genetic testing in elite sport remains infrequent and sporadic, with only 10% of athletes and 11% of support staff who responded to this survey stating they had utilised genetic testing within their practice. This prevalence of genetic testing in athletes is similar to previously published research (Varley et al., 2018) although the reported use by sporting organisations is much higher, standing at 11%, compared to 2% in Varley and colleagues (2018).

Overall, the survey respondents believed that genetics has a sizeable (>25%) impact on an individual's potential to be an elite athlete (92% of athletes and 91% of support staff).

These attitudes correspond to the findings of published research exploring the genetic influence on sporting phenotypes. For example, De Moor and colleagues (2007) reported that heritable factors explained approximately 66% of the variation in elite athlete status between individuals within their cohort. Furthermore, a number of single nucleotide polymorphisms have been identified which may increase an individual's chance of attaining elite athlete status (Ahmetov et al., 2016), although the research in this area remains equivocal (Rankinen et al., 2016). However, at present, this information does not appear to be all that useful in identifying potential elite athletes, leading to the prevalent scientific consensus that genetic testing should not be used as a talent identification tool (Webborn et al., 2015; Vlahovich et al., 2017). This viewpoint appears to be mirrored in the practice of support staff, none of whom had utilised genetic testing as a talent screen. One athlete did, however, report using their genetic results as a means of identifying which sport they should compete in.

Additionally, 88% of athletes and 93% of support staff respondents believed that genetics has a sizeable (>25%) impact on an individual's improvements following a training programme. Again, this perspective is mirrored in the research literature; individual SNPs, such as *ACTN3* and *PPARGC1A*, appear to modify the magnitude of post-training adaptations (Pickering & Kiely, 2017; Ring-Dimitriou et al., 2014). More recently, studies have started to explore the potential utility of Total Genotype Scores in explaining the variation in training response (Pickering et al., 2018; Jones et al., 2016; He et al., 2018; Moraes et al., 2018) and potentially in maximising adaptations to exercise (Jones et al., 2016). Of the athletes within this study who indicated they had undertaken genetic testing, 44% stated it was to inform training programme design, as did 50% of support staff.

Interestingly, fewer athletes (74%) and support staff (70%) believed that genetics had a sizeable (>25%) impact on an individual's nutrient requirements. This is somewhat surprising, given that the field of nutrigenetics is well established, with a number of studies demonstrating how SNPs in genes such as *MTHFR* (Ashfield-Watt et al., 2002) and *SOD2* (Li et al., 2005) potentially modify micronutrient status and requirements, although this may be outside the scope of practice of many involved within elite sport. Consequently, only 11% of athletes, and 15% of support staff, utilised genetic testing to gain insights into nutritional requirements.

Although the vast majority of both athletes and support staff surveyed believed that genetics had a substantial influence on a number of sporting phenotypes, the overall uptake of testing was somewhat low (~10%). Consequently, we explored potential reasons for this disparity. 49% of athletes stated they were unaware that genetic testing was available, suggesting that one of the main drivers for the low uptake of genetic testing is a lack of awareness. A greater number of support staff were aware that genetic testing was available, possibly due to recent publications in the scientific literature on the subject (e.g. Webb et al. [2015]; Jones et al. [2016]). However, potentially due to the conclusions of two recent consensus statements (Webb et al., 2015; Vlahovich et al., 2017), a large proportion (39%) of support staff believed that there was insufficient evidence supporting the use of genetic testing within elite sport. Cost was also an issue, with 31% of athletes and 41% of support staff stating that one of the reasons they had not utilised genetic testing was that it was too expensive. Whilst, historically, genetic testing has been costly (Hayden, 2014), in recent years technological improvements and increased sales volumes have reduced prices, such that a genetic test today typically costs £100-£200. Neither athletes nor support staff appeared especially concerned around data protection or negative press coverage, with few citing these

as reasons they had not undertaken genetic testing. However, 19% of support staff stated that ethical considerations, such as the perceived use of genetic information for talent identification, was one of the reasons they had not utilised genetic testing.

Support staff generally noted a need for increased scientific evidence before they would consider utilising genetic information in the future. Conversely, athletes were less concerned about this, instead stating that, if more athletes and sports teams began using genetic testing, they too would consider it. Both athletes (48%) and support staff (36%) stated that a reduction in price would lead them to consider genetic testing, and 25% of support staff would consider a genetic test should a player request it. This latter finding is interesting, as it demonstrates that many practitioners understand the value of player buy-in and potential expectancy effects surrounding the use of genetic information, thereby mirroring the findings of a recent survey of Premier League medical staff regarding the use of Platelet Rich Plasma injections (McNamee et al., 2018).

When genetic testing had been used, athletes (73%; $n = 8$) and support staff (64%; $n = 9$) perceived it to be useful. The main reasons cited for a lack of utility were that the information was either too generic, or not targeted specifically towards sports people. Most athletes who had undertaken a genetic test stated they had made changes to either their training or lifestyle based on the results of the test, demonstrating a perceived utility of the genetic information. The vast majority of genetic testing reported by participants in this survey was carried out by commercial companies, as opposed to academic institutions. Most athletes (73%; $n = 8$) and support staff (64%; $n = 9$) received follow up information from the testing company, giving them the opportunity to ask additional questions and clarify any misunderstandings.

Whilst not hugely prevalent as of yet, the results of this current survey, alongside previous work (Varley et al., 2018), illustrates there is a potential appetite for genetic information within elite sport. This comes against the backdrop of consensus statements cautioning against the use of genetic tests within sport (Webborn et al., 2015; Williams et al., 2014). Accordingly, future research may wish to examine some of the many ethical issues surrounding the use of genetic testing within sport, along with the development of best practice guidelines. For example, is it ever ethical to test those under-18, who, in theory, cannot provide informed consent? Can a sporting organisation recommend or even require genetic testing of its athletes, and at what point would such organizational ‘pressure’ constitute coercion? Who owns the genetic data, and where and how is it stored? What happens to this data if a player leaves a club/organisation, or retires? What provisions, if any, are made for the discovery of potentially disease-causing or disease-associated variants within an athlete’s genetic data? How would this discovery affect relatives of the athlete, who may also require genetic screening for the particular disease variant? What are the additional healthcare burdens and costs associated with this? At present, there are no guidelines assisting practitioners in answering these questions, or even guiding them towards an informed decision. As such, the development of such guidelines represents a potential opportunity to enhance future practice.

4.1 Limitations

Whilst the results of this survey are novel and interesting, with the potential to impact research and practice, there are some limitations. Firstly, the survey was shared by the authors on social media, which may have affected the individuals targeted, as their followers

may have an interest in genetic testing. However, the extent of athletes stating they had undertaken genetic testing in this study (10%) was actually lower than in previous research (Varley et al., 2018), suggesting this had not skewed the results. Additionally, the vast majority (~80%) of respondents were male; whilst we see no reason why females would be more or less likely to undertake a genetic test, or hold different attitudes towards such a test, we cannot rule out this possibility. Furthermore, there is considerable variation between various genetic testing companies in terms of product quality, validity, and reliability; the companies utilised by participants was not collected in this present study, which may have yielded additional insights.

The survey was also undertaken anonymously; as a result, it is possible that the support staff survey respondents were from the same club or organisation, thereby artificially inflating the apparent prevalence of genetic testing based on the results of these respondents. The survey collected descriptive data, and did not seek to understand the motivations of respondents undertaking genetic testing; future research should explore this area in more depth. Finally, the survey questions had not been validated; the use of validated questionnaires is an important consideration for future research. Future research should also seek to address the various limitations identified by aiming to recruit a higher number of female athletes—the lack of female support staff participants may (sadly) mirror actual female representation within high-level sport (Kane & LaVoi, 2017)—and verify that only a single support staff responds from each club or organisation. Future studies should also aim to recruit an increased number of participants, and explore the attitudes towards genetic testing within regions that were not adequately covered here. That said, the present study recruited a number of Olympic/World Championship athletes (18%), with over half the sample being international athletes. Additionally, of the support staff polled, 36% stated that

their most frequent contact was with athletes of an international standard, demonstrating that the cohort was of a high standard, a potential strength of this study.

5. Conclusion

Despite the fact that the majority of athletes and support staff polled in this survey believed that genetics exert a sizeable influence on a number of sporting-related traits, the overall uptake of genetic testing within this cohort, in which more than half of the athletes polled had represented their country, was low, at around 10%. The reasons for this are varied, but include a lack of awareness of availability, cost, and a perceived lack of scientific evidence substantiating the use of such tests. Despite concerns from researchers in this field (Webborn et al., 2015), it appears that the vast majority of those who have utilised genetic testing within sport are not doing so as a talent identification tool, and instead are doing so as a method to inform training programme design. Whilst there is some evidence supporting the use of genetic information in this way (Jones et al., 2016; Pickering & Kiely, 2018), additional research is required to:

- (i) Build an evidence-based framework for the use of genetic information within sport;
- (ii) Development of best-practice guidelines relating to the testing of athletes by sporting organisations;
- (iii) Address any and all ethical considerations, including how individual athlete data can best be shared, stored, and/or protected by clubs and sporting organisations.

This study demonstrates that ~10% of high-level athletes have utilised genetic testing as part of their athletic preparation process, and many more believe they will do so within the

next five years. This suggests that coaches may need to better understand the relative impact of genetic information within sport, be able to critically review any genetic information presented, and understand some of the ethical challenges genetic testing may present, in order to best serve and protect their athletes.

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