

Genetic Testing and Sports Medicine Ethics

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Abstract

Sports medicine ethics is neither a well established branch of sports medicine nor of medical ethics. It is therefore important to raise to more general awareness some of the significant ethical implications of sports medicine practices. The field of genetics in sports is likewise in its infancy and raises significant ethical concerns. It is not yet clear how genetics will alter our understanding of human potential and performance in sports. While a number of professional medical bodies accept genetic interventions of a therapeutic nature, we argue that the use of genetic technologies to predict sports potential may well breach both the European bioethics convention and North American anti-discrimination legislation, which are designed to support important ethical ideals and the ongoing commitment of the physician to the welfare of their patient. We highlight further ethical problems associated with confidentiality and consent that may arise in genetic testing as opposed to more conventional methods of testing in sports medicine. We conclude that genetic testing in sport that is not strictly limited to the protection of the athlete against harm, should be viewed in a very sceptical light by sports medicine professionals.

With the particular exception of doping, the emerging field of sports medicine ethics has attracted relatively little ethical discussion compared with more established branches of medicine. Recently, commercial,^[1,2] professional and scientific discussions of genetics have raised the possibility of genetic testing for sports performance prediction^[3-7] in addition to preventative and therapeutic purposes.^[6-8] We note the powerful case that can be made for genetic testing regarding the identification of predisposition to hypertrophic cardiomyopathy.^[9-13] However, in contrast, we

raise key ethical issues that reveal a conflict between common employment practice and professional sport from within the more established fields of medical ethics and legislation. Genetic testing for predictive purposes such as talent identification or performance profiling is potentially in breach of the Council of Europe Bioethics Convention^[14] and the Genetic Information Non-Discrimination Act in the US.^[15] Given the economic and power asymmetry between professional sports franchises and individual professional athletes, we argue that the voluntary

consenting to genetic testing may be undermined and that duress or even coercion may be used to secure the data that can be acquired through genetic testing. Moreover, even if acceptance is secured under voluntary conditions it is necessary to consider the degree of comprehension and thus the 'informedness' of athletes of the consent. Finally, if genetic testing is to develop in sports medicine, we highlight the need for acceptable and available systems for genetic counselling before and after testing.^[7] We conclude that genetic testing in sport, which is not strictly limited to the protection of the athletes against harm, should be viewed in a very sceptical light by sports medicine professionals.

While sports ethics as a research field is in its infancy, sports medicine ethics can only be classified as neonatal. Very few articles exist that attempt to lay down the boundaries of what is a potentially important research field.^[16,17] The conflicts of interests between team doctors in relation to their employing sports franchises and other governing bodies such as the National Basketball Association or the National Football League and their long-term commitment to the health and well-being of their athlete patient are relatively well known^[18] but rarely scrutinized from an ethical point of view. The advent of genetic medicine in general and genetic testing in sports medicine brings new ethical issues to light that merit critical ethical scrutiny.

It is not yet clear precisely how genetics will alter our understanding of athletic potential and performance.^[19,20] Some of the claims made by sports ethicists and scientists regarding the potential for human enhancement seem to blur the lines between fact and science fiction.^[3,21] In addition to these problems, a fairly recent article neglects even to mention professional athletics and sports medicine in its review of ethical, legal and social implications of genetic medicine.^[22] Whether or not we accept the enhancement scenarios, some research has established some important genetic precursors to athletic development especially regarding muscular contraction and growth. However, little comment has been made concerning the preventive application of genetic testing in sports as well as of the ethical

implications for sports medicine. This article raises a number of interrelated ethical issues that affect sports medicine in relation to professional athletes.

1. Genetic Testing for Prevention or (Sports) Performance?

Recently both popular and scientific discussion has raised the possibility of genetic testing for sports performance prediction. The following is indicative: "Many of the variables that determine athletic performance are partially inherited (Spurway, 2007) and therefore one can foresee the use of genetic tests to predict performance."^[3] Beyond biomedical science, some bioethicists have also made similar claims as to its potential uses,^[4-7] while others, ourselves included, are more sceptical.^[8] The potential of sports genetics is often based on the claim that a single gene (*ACTN3*) is crucially related to sports performance potential.^[23] This is based on its expression in type II (fast twitch) muscle fibres, which are of importance to sports where speed is integral.

There are even commercially available test kits^[1,2] for the most eager of sports parents or youth sports coaches with talent identification (and its economic and social benefits) in mind. The discovery of *ACTN3* apparently "marks the beginning of a new era."^[3] What is to be made of such a grand claim? The first is that perhaps the attraction of citing such an important genetic contributor is so great that some are prepared to leap precipitously to claim that without *ACTN3* there is no quick muscle contraction. However, a recent single case report of a Spanish double-Olympic, world-class long jumper has shown that his achievements were notable because of a deficient *ACTN3* gene.^[24] We would be mistaken if we tried to reduce complex traits such as muscle power and speed down to a single gene.^[25] Therefore, a precautionary approach might be wiser until the evidence is supported more widely about the function of *ACTN3*. However, a recent analysis of commercial genetic profiling for health risks and interventions suggests:

“Although genomic profiling may have potential to enhance the effectiveness and efficiency of preventive interventions, to date the scientific evidence for most associations between genetic variants and disease risk is insufficient to support useful applications.”^[26]

Given that genomic medicine and technology are advancing so rapidly, it is worth considering their potential ethical impact in advance of actual medical applications in athletics.^[6] Although in several cases healthcare practitioners are powerless to prevent or treat certain conditions after the realization of a (genotype) positive test result, genetic testing for athletes might enable physicians to prevent individuals who are not aware of their health condition from dying a death triggered by sports, as in the case of hypertrophic cardiomyopathy.^[9-13]

Currently, the health-related use of genetic testing in sport (i.e. in preparticipation examinations) is not a standard procedure. It is recommended by some cardiologists in borderline cases (ambiguous ECG/echo, borderline wall thickness). Pigozzi and Rizzo state that “if certain diagnosis is not possible but the suspect of disease is high, the most definitive evidence for the presence of hypertrophic cardiomyopathy (HCM) comes from DNA analysis.”^[27] If, however, a family member is diagnosed with hypertrophic cardiomyopathy, then, despite lack of symptoms, a genetic screening of the entire family should be considered.^[28] On the other hand, genetic testing is *mandatory* when definitive diagnosis for genotype-related risk stratification and therapy is required, as can be the case in athletes with long QT syndrome (LQTS).^[11] Regarding LQTS, the Heart Rhythm UK Familial Sudden Death Syndromes Statement Development Group sounds caution by stating that “genetic testing is not recommended for diagnosis of uncertain or ‘borderline’ congenital LQTS outside the setting of expert clinical and detailed family assessment.”^[29] Not all diseases are monocausal, which obviously reduces the predictive quality of such tests, not to mention the high costs as a consequence of testing a whole range of genes that are suspected to be linked to a certain disease. These obstacles (among others) lead to the con-

clusion that genetic tests are probably not becoming a standard in preparticipation screenings, at least not in the near future.

2. Genetic Testing in Sports and the Legal Regulation of Genetic Testing in Employment

Suppose that athletic predestination is reliably predictable by new genetic sports medicine at some point in the near future. What will follow from this? What is scientifically possible and what is ethically permissible do not always go hand in hand. One very significant barrier to this use of genetics already exists in Europe in the form of the Council of Europe Bioethics Convention where Article 12 regarding predictive genetic tests states:

“Tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counselling.”^[14]

Here, the idea of someone undergoing genetic testing in order to establish some kind of performance profile would itself go against the strict therapeutic or preventative rationale of the Council of Europe Convention. Also, in the US the Genetic Information Non-Discrimination Act of 2008 Section 202a explicitly prohibits the use of genetic information in employment decisions.^[15]

There are further obstacles to genetic testing for sports performance arising from this Convention that raise ethical issues to be addressed beyond European confines, which we address below. For the moment, it is important to note one important difference between genetic testing in public health and in professional sports. However, as noted above, where healthcare practitioners are incapable of therapeutic interventions, the testing is still not futile given the possibility of identifying susceptibility to HCM.^[9-13] Equally, this may cause problems since genetic data revealed about a given athlete may be disclosed to public bodies such as the World Anti-Doping

Agency and in so doing the privacy of data of relatives will be denied.^[6,7] Nevertheless, just because it may be impossible to cure a patient of a given condition, it does not follow that genetic testing is redundant. However “DNA-based diagnostic tests that can definitively distinguish genetic heart diseases from athlete’s heart”^[13] could genuinely save lives. Moreover, genetic testing for *APOE4* in addition to traditional screening has been used voluntarily in Australia with boxers who are vulnerable to early onset of Alzheimer’s disease if they have the gene.^[4,30,31] Our point is not against genetic testing in sports *per se*, but merely against the questionable validity of genetic prediction of sports performance and the expansion of its role beyond traditional preventative and therapeutic aims. While it is clear that the boundary between the traditional curative goals of medicine and the more novel enhancement aims of sports medicine is not absolute, the distinction still remains a useful one.

3. Problems of Confidentiality and Consent in Professional Sports Medicine

In highly paid professional sports, what are the possible scenarios in offering a test for a sports-related risk factor? Can the asymmetry of power between franchise and player be made worse by genetic knowledge? If consent procedures are properly followed, the ability to volunteer might still be in question: how does the sports physician present the case for and against, under what circumstances and with what preconditions? Even if this process was followed in an ethically acceptable manner, what steps should follow from different test results. Testing itself does not guarantee objective and unequivocal prognoses: nor does the genetic counselling, which would need to follow.^[7,32] Human interpretation and valuation of risk factors are still necessary. How are the test results presented, by whom and under what conditions? What does a certain risk mean in one context, compared with another? All these issues will require serious public professional debate if genetic testing is to gain a foothold in sports medicine.

Moreover, while respect for patient autonomy is often regarded as the crucible of medical ethics,^[33] concerns arise from the very nature of genetic data presented under the heading ‘genetic exceptionalism’.^[34] For example, Yesley^[35] disputes the claims to the uniqueness of genetic data as opposed to other forms of traditional screening and testing. Given, however, the complexity of issues surrounding genetic data, what confidence is justified regarding professional athletes’ capacity to grasp fully the decision to be genetically tested? A further problem arises regarding confidentiality: with whom will the genetic data be shared? Will existing pressures on team doctors to divulge athlete/patient information^[16] be exacerbated? Often the physiotherapist or team doctor find themselves caught in a conflict of interest. They serve both the athlete/patient and the franchise/client who pay their wages. The data they have regarding the health and performance status are highly sensitive but in great demand from the employing franchise, the coach and potential commercial suitors.

Genetic testing in employment for anything other than health risk related to the specific job is generally frowned upon. Why should professional sports differ? Article 12 of the European Convention prohibits the use of predictive tests for non-health-related reasons, even with the assent of the patient. Predictive genetic testing as part of pre-employment medical examinations is forbidden whenever it does not serve the health-related interests of the individual. This means that in particular circumstances, when the working environment could have prejudicial consequences on the health of an individual because of a genetic predisposition, predictive genetic testing may be offered without prejudice with the aim of improving working conditions. Which genetic anomalies are deleterious to given athletes in specific sports? Should sports employers be allowed to hire and fire based on unexpressed genetic abnormalities? How should the athlete’s right not to know other deleterious conditions be respected? There is a justifiable requirement to carry out performance tests to judge, for example, the effort of the athlete, or their adherence and commitment to a specified training

regimen in order to decide whether to offer or extend a professional contract. This does not, however, justify a 'fishing expedition' of potentially wide-ranging personal genetic data.

4. Conclusions

We have attempted to raise critical questions regarding the potential of genetic testing in sports medicine without rejecting it *tout court*. We recognize that if genetic testing is carried out for enhancement purposes it may have the unintended, but desirable, consequence of highlighting potentially harmful diseases or conditions that may be exacerbated by high-level athletic activity.

Nevertheless, we have cast doubt on the possibility of sports performance prediction, but have also raised key ethical issues where there is a clash between common employment practice and sport and mainstream medical ethics and law. Given the economic asymmetry between the commercial sports franchise and the individual professional athlete, we have shown that genetic testing in sport that is not strictly limited to the protection of the athlete against harm should be viewed in a very sceptical light by sports medicine professionals.

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