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Against genetic tests for athletic talent: the primacy of the phenotype

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Abstract

New insights into the genetics of sport performance lead to new areas of application. One area is the use of genetic tests to identify athletic talent. Athletic performances involve a high number of complex phenotypical traits. Based on the ACCE model (review of analytic and clinical validity, clinical utility, and ethical, legal and social implications) a critique is offered of the lack of validity and predictive power of genetic tests for talent. Based on the ideal of children's right to an open future a moral argument is given against such tests on children and young athletes. A possible role of genetic tests in sport is proposed in terms of identifying predisposition for injury. If meeting ACCE requirements such tests could improve individualized injury prevention and increase athlete health. More generally, limitations of science are discussed in the identification of talent and in the understanding of complex human performance phenotypes. An alternative approach to talent identification is proposed in terms of ethically sensitive, systematic and evidence-based holistic observation over time of relevant phenotypical traits by experienced observers. Talent identification in sport should be based on the primacy of the phenotype.

Key points

- Genetic tests for athletic talent have insufficient scientific basis and low predictive power and challenge the ethical ideal of children's right to an open future.
- A possible role for genetic tests in sport is identifying predisposition for injury.
- In talent identification the superior approach seems to be ethically sensitive, systematic and evidence-based holistic observation over time of relevant phenotypical traits by experienced observers.

Against genetic tests for athletic talent: the primacy of the phenotype

1. Introduction

Research on associations between genetic predispositions and sport performance is breaking new ground and has a series of actual and potential applications (1-3). As with most biomedical innovations insights can be used both in constructive and more problematic ways (4, 5). A controversial topic is the potential use of invasive gene therapy techniques to enhance athletic performance. There is relatively strong consensus in the sporting community on the non-acceptability of so-called gene doping. World Anti-doping Agency (WADA) lists gene doping as a banned method (6). Another possibility upon which there is less consensus is the non-invasive use of genetic insights to identify athletic talent (7-10). In what follows a critical discussion is presented of the use of genetic talent tests. It is concluded that such tests are scientifically unsound and morally problematic. More generally, the discussion illustrates some of the limitations of science when it comes to understanding highly complex human phenotypes like those involved in athletic expertise. An alternative approach to talent identification (TI) is proposed in terms of ethically sensitive, systematic and evidence-based holistic observation over time of relevant phenotypical traits by experienced observers. TI should be based on the primacy of the phenotype.

2. Talent, genetic tests, and perfectionism

Competitive sport is linked to a diversity of individual and social values, among them experiential values such as joy, excitement and mastery; social values such as team building and integration; and improved health. The social logic of sport competitions however is less open to interpretation. The structural goal of competitions is to measure, compare, and finally rank competitors according to rule-defined athletic performance

(11). In formally organized and professional sport, success depends upon performance and progress. Hence, TI is considered crucial.

This has in part pragmatic reasons. Sport systems aspire to cost efficiency and avoiding spending resources on athletes with marginal chances of success. There are also moral arguments supporting systematic TI and development. From an Aristotelian perfectionist point of view individuals have an obligation to explore and develop their innate potential to the largest possible extent, that is, to realize their 'human nature' (12). Philosopher John Rawls' expresses the idea in more detail with what he calls the Aristotelian principle (13):

Other things equal, human beings enjoy the exercise of their realized capacities (their innate or trained abilities), and this enjoyment increases the more the capacity is realized, or the greater its complexity.

Perfectionism aims at human flourishing and development of a diversity of human talent. With perfectionism as a premise it can be argued that competent and morally sound TI and development in sport (as in other areas of life) should be strived for as valuable in itself. What are the criteria for competence and moral soundness in this respect?

On the competence side, sport science offers knowledge of many kinds ranging from physiological and anthropometric measures to psychology and socio-cultural analyses (14, 15, 16). One common assumption is that insights into the genetics of physical performance, for instance into associations between genes and basic bio-motor qualities such as endurance, strength and power, will increase the quality of these schemes.

On the more radical side, there are discussions on the potential of genetics in pre-natal testing and selection of germ line cells and embryos (4, 17). There is talk of a 'genetic making of champions'. These wide-ranging perspectives will not be discussed here. More realistic and topical approaches build on insights into the genetic basis of relevant performance phenotypes. An example of the latter is the 2003 publication on associations between variants of the ACTN3 gene and composition of fast and slow twitch muscle fibers (18). In less than one year commercial companies followed up with

simple direct-to-consumer solutions marketed as athletic talent tests predicting predispositions for endurance and/or power events (10). Do such tests stand up to critical scrutiny?

Building on relevant research and clinical experience, the Office for Public Health Genomics at US Centers for Disease Control and Prevention (CDC) has supported development of a procedure for critical reviews of DNA (and related) testing for disorders with a genetic component. In the so-called ACCE (Analytic validity, Clinical validity, Clinical utility, and Ethical, legal and social implications) model,¹ genetic tests are assessed according to analytic and clinical validity on sensitivity (how often is the test positive when a mutation is present?), specificity (how often is the test negative when a mutation is not present?), and prevalence. Moreover, new tests are examined in terms of clinical utility and practical implementation (is the test applicable and useful?), and of whether they are ethically, legally and socially acceptable.

The ACCE model is developed for assessing genetic tests for disorders but is also relevant to a discussion of genetic tests for athletic talent. A first task is to define athletic talent and describe genetic tests associated with relevant phenotypes. When it comes to analytic and clinical validity, the basic question concerns the strength of the association between genetic information provided by the test and phenotypes. How sensitive and specific is the test? Questions of clinical utility, or what in the context of TI in sport is better labeled practical utility, are related to the predictive strength of the test and its applicability. Is it really useful? Does it provide sufficient information? Genetic testing for talent is also a morally contested field, in particular if performed on children and young athletes. Critical questions concern the meaning and value of children's sport and more generally the ethics of children's upbringing. In what follows a discussion of TI in sport is presented based on the ACCE model.

3. Scientific validity and practical utility

¹ The ACCE model defines a procedure for genetic test assessment with 44 targeted questions. For background information and a complete overview of the 44 questions, see Sanderson et al. (19) and the CDC website (20).

How can athletic talent be defined, and are there valid and useful tests measuring genetic predispositions for athletic performance?

In most sports, successful performance depends upon deliberate practice for many years under competent guidance. In studies by Ericsson and others it is estimated that to reach expertise in performance areas such as music and chess one needs about 10000 hours of practice before the age of 20 (21, 22, 23). Similar theses have been put forward when it comes to sport (24).

Training however is not enough. As pinpointed by Ericsson and Lehman there are individual and qualitative differences in experts' organization of knowledge that are not really well understood (25). Differences between proficient performers and experts are often explained by differences in talent.

The concept of talent is much discussed, and TI and talent development are considered complex processes (14, 15, 16). Somewhat simplistically, 'talent' can be defined as an individual's potential to develop performance in a particular field of expertise. Moreover, talent is often linked to genetic predispositions; the idea that individuals are naturally endowed and gifted. Some individuals are 'born' with a larger or lesser degree of talent. To rephrase exercise physiologists Astrand and Rodahl (26), '...if you want to become a world-class athlete you must choose your parents wisely.'

Empirically speaking, drawing clear-cut distinctions between genetic predispositions and environmental impact is complicated if not impossible. Any serious attempt on explaining and identifying talent has to account for an immense number of interacting factors. Still, in a discussion of the potential of genetic tests a theoretical *analytic* distinction between genetic predispositions (talent) and environmental factors serves to clarify the argument (11).

How, more specifically, can genetic impact on athletic performance be categorized? Informed answers have to relate to performance requirements found in various sport disciplines (27). On the 100-meter sprint race, genetic predispositions for developing explosive power and speed are crucial. These are basic bio-motor abilities and can be associated relatively clearly with genetic predispositions (3, 18). In this context one could think that genetic tests for identifying talent would be of significant value. In ball games such as European football, there is need for endurance and speed

but also for advanced technical and tactical skills. The genetic basis of performance is far more complex. Skill development depends upon social interaction and learning over many years and cannot be easily traced genetically.

Informed answers also have to relate to the complexity and multi-factorial nature of core phenotypic traits of athletic performance (3, 27). Genetic predispositions for performance are defined in the so-called 'natural lottery', that is, by chance. From the moment of conception and onwards, their phenotypic expression depends upon an immense complexity of gene-gene-environment interactions. Some of these interactions are more or less outside of control of the individual. A person with strong predispositions for power, speed or endurance is born next to a good athletics facility and comes under the influence of an excellent coach. This person performs well in athletics. Typically, we see this as a matter of having good luck (28). Other persons with the same or even better predispositions may be born into less fortunate environments, or may suffer malnutrition or injury. If their ambition is to perform well in sport, typically we characterize them as being unfortunate and having bad luck.

Other interactions are within control of the individual. No individual can develop talent towards expertise without systematic and hard training. Elite athletes are characterized by a series of strong psychological qualities, among them 'adaptive perfectionism' and will power (29). They are dedicated. Typically great performances are admired as the results of hard work. Good performances are clear outcomes of merit.

Factors of chance, luck and merit work together in complex ways from the moment of conception to the moment of performance and are impossible to fully control and manipulate. In this context identifying and testing for genetic talent is challenging if not impossible. In an article with the telling title 'If you don't know where you are going, you might wind up someplace else', Greenbaum (30) pinpoints the crucial invalidity and lack of practical utility and predictive power of genetic tests for athletic talent. At least at this point in time such tests do not meet even basic requirements of the ACCE model. Use of genetic tests in TI is unsound on scientific grounds and therefore also without practical utility in sport.

4. The ethics of genetic testing for athletic talent

The last part of the ACCE framework requests consideration on ethical, legal and social issues. Obviously, lack of scientific validity makes it unethical to use genetic talent tests. Hypothetically, however, if in the future such tests meet validity requirements on certain aspects of talent in some sports, should they be included in TI programs?

TI in sport deals almost without exception with children and young athletes. A predominant Western moral ideal for children's upbringing is the right to an open future (31, 32). Children should be brought up not by 'guardians' but by 'gardeners' (33). Children ought to be exposed to many possibilities in life so as to find activities and areas of expertise in which they can experience meaning and mastery and flourish. From a perfectionist perspective developing ones abilities and talents and contributing to similar developments in others are of value in itself. To the largest possible extent children and youth ought to be exposed to an all round education.

Therefore the use of genetic tests for TI in children and young athletes is problematic. Even if validity of these procedures improves, the complexity of developing human performance phenotypes will nevertheless make their predictive power limited. Moreover, even if such tests become part of a more extensive TI regimen, test results are easily misinterpreted and their results over-emphasized. Typically, testing takes place at early stages of athletic development before clear phenotypic traits are developed. The risk of making selection mistakes is significant. An additional critical point is that even if a selected child has a significant athletic talent, the same child may have several and even greater talents worthwhile pursuing; talent for music, or for mathematics, or dance. The critique of limiting children's possible choices in life is a general critique of early TI and selection (to which genetic tests seem to contribute), and not only of genetic talent tests (32).

5. Athletic talent, injury risk, and the primacy of the phenotype

The limitations of genetic talent tests do not mean that insights into the genetics of human performance are of low relevance in sport. On the contrary, even if genetic tests are unsuitable in TI they may fill other significant roles. Several authors reflect upon

tests not for 'performance genes' but for genetic predispositions for injury (3, 16, 30). If research demonstrates clear associations between certain genetic predispositions for injury, for instance for tendinopathy or concussion, and if tests can meet the requirements of the ACCE-model (including requirements on confidentiality and sound procedures for incidental findings), tests should be considered. Applied sport genetics can improve athlete health.

Interestingly, genetic tests to map injury risk are based on a different logic than tests for athletic talent. As Camporesi and McNamee observe, norms for medical ethics are not easily translated into sport medicine (34). Competitive sport is a perfectionist practice cultivating performance. The goal of medicine is to prevent and cure illness and disease. Within a preventive, medical scheme it makes sense to speak of the primacy of the genotype. The point is to map a genetic predisposition to prevent its phenotypic expression. In TI the aim is to select persons with genetic predisposition for phenotypical traits of significance to athletic success. The aim is not mapping the genotype but developing the phenotype. In this context genetic tests have proven to be problematic and of limited value.

As mentioned in the introduction, competent and ethically responsible TI can be justified both on pragmatic and ethical grounds. In their thorough review of conventional TI research, Breitbach et al (16) point towards a series of weaknesses; cross-sectional design prevents insights into athlete development over time, there are no clear criteria for optimal timing of the tests, test items vary, and the immense complexity of interactions between the many factors of athletic performance development seems impossible to capture. Conventional TI faces significant validity and reliability problems.

Janvier and Farlow (35) talk of strong beliefs in genetic talent tests as 'arrogance-based medicine'. It seems pertinent to warn of similar 'arrogance-based' sport science. In successful TI, scientific insights have a complementary function in providing evidence-based information and a systematic and critical perspective. Science is necessary but by no means sufficient. A core recommendation for future TI research is multi-disciplinary longitudinal design with the possibility of following young athletes over time (16). This seems close to traditional best practice among expert coaches. As any experienced coach can report, talent is expressed over time in a complex mix of physical abilities and

psychological qualities developing in close interaction with social-psychological and socio-cultural contexts (15, 27). If a child enjoys running and runs fast, this child is a running talent. If a child has good control over the ball, understands team tactics and has a strong passion and motivation to learn and improve, this child is a ball game talent.

Experienced coaches knowing their sport have what Polanyi calls 'tacit knowledge'. They are experts in the execution of their practice and combine a systematic and critical approach with the ancient Greek ideal of *phronesis*; practical wisdom and good judgment (36). Good coaches are 'enlightened generalists' (37). In TI therefore the less reductionist and probably most accurate approach seems to be systematic and evidence-based holistic observation over time of relevant phenotypical traits by experienced observers combined with ethical sensitivity of children's right to an open future.

6. Conclusion

Competent and ethically sensitive TI can be justified on both pragmatic and ethical grounds. The requirement on competence however does not include genetic tests. As of today identifying and testing for genetic talent is difficult if not impossible. Hence, ACCE-requirements of analytic and clinical validity of genetic talent tests cannot be assessed, and the usefulness of tests as predictors of future sport success is negligible. Moreover, their interpretation and use tend to challenge an ethical principle of children's right to an open future.

Genetic tests for injury risk on the other hand may have a future role to play in TI and in training in general. Such tests follow the logic of medicine. The primacy is on the genotype with the aim of preventing development of non-desired phenotypes. If meeting the requirements of the ACCE model, genetic tests can improve individualized injury prevention and athlete health.

In TI, science contributes with evidence-based insights and a systematic and critical perspective. This however is a complementary role. The superior approach seems to be ethically sensitive evidence-based holistic observation over time of relevant phenotypical traits by experienced observers. The principle to be held high is that of the

primacy of the phenotype.

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