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# Genomics as a practical tool in sport - have we reached the starting line?

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## Abstract

The genetic component of athletic performance approximates 50%, depending on which specific element of performance is considered. Limited genetic testing is already available commercially and genetic tests are likely to become powerful tools to improve sport performance in the future. Currently, however, selection of athletes for training squads or competition based on genomic data is premature. Larger volumes of longitudinal data within individual sports are needed to determine the efficacy of using genomic data in the management of elite athletes via manipulation of training load and diet based on personal genomic information.

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Since the 1990s, exercise genomics research has regularly involved investigation of DNA sequence variation in relation to exercise-related phenotypes. Although the pace of this research remains modest, progress is being made and significant advances are predicted for the near future (Pitsiladis et al., 2013).

The difference between success and failure in competitive sport is often determined by extremely small performance margins (Pyne et al., 2004) that can be influenced by physiology, genetics, psychological factors, etc. Just one of many possible examples is the 0.01 s margin of victory for Michael Phelps during the 100 m butterfly at the 2008 Olympic Games. There are numerous accounts of scientists, coaches and athletes adopting (or at least trialling, in a practical setting) innovative, permitted strategies in an attempt to improve performance. A 1% (or even 0.1%) improvement in performance could be the difference between winning and losing and the drive to improve performance by even those tiny fractions can be all-consuming for those people involved. Yet the consequences of failure to improve performance do not threaten health - even adverse responses rarely pose a risk to life or have serious health consequences. Therefore, it is arguable that the standard of evidence required before practical trials begin by athletes and coaches is lower than that required for newly-developed cancer drugs in a clinical setting, for example. Nevertheless, responsible scientists should act and base recommendations on evidence.

In exercise genomics, our impression from speaking with colleagues at international conferences and similar events, is that the scientific consensus appears to be that practical application to sport at this time is probably premature – and this impression is despite the standard of evidence comments we make above. Certainly, in terms of athlete selection for competition or training squads, more research is required before existing practices in sport could justifiably be changed due to genomic data. Firstly, researchers must recruit larger numbers of athletes for case-control study designs (Bouchard, 2011; Pitsiladis et al., 2013) and identify more genetic variants that contribute to the genetic component of sport performance. We are aware of major efforts in this regard involving athletes of recent African ancestry and other efforts taking place in eastern Europe (Russia and neighbouring countries), China, Australia, Brazil, Japan and the UK (UK athletes are a current focus of our own work), while several other research groups are making similar efforts. Thus, international collaboration seems logical to pool data and resources where feasible and appropriate. Secondly, researchers must conduct comprehensive research within a given sport. Grouping different sports (and even different specialities within a sport) to boost sample size theoretically brings important increases in statistical power but also other difficulties by diluting the phenotypic characteristics, selection and training practices of elite athlete groups.

Conducting comprehensive genomics research within a given sport would limit the problem of diluting phenotypic characteristics, selection and training practices, and requires co-operation from relevant stakeholders within at least one reasonably large nation (and preferably internationally), as well as careful consideration of the recent geographic ancestry of the athletes. The net effect of purifying the sport phenotype versus limiting sample size, both of which affect statistical power but in opposite directions, is unclear at this time. However, if a large sample size can be obtained where the phenotypes, selection and training practices are precisely defined and consistent, we believe that powerful, convincing data will ultimately be produced that influence the management of elite athletes. We have enticing, unpublished, preliminary genomic data from within the sport of rugby union that addresses some of these issues. We of course envisage genomic information complementing the existing phenotypic data collected and used

by sports coaches and supporting exercise scientists – genomic information will never be a replacement for such data, but an additional tool.

We predict the first evidence-based practical application of genomic information in the management of elite athletes will involve modification of training in an attempt to reduce the likelihood of injury when an athlete is predicted from his or her genome to be at higher than average risk. Secondly, personalisation of training programmes should become possible in sports where two or more physiological characteristics combine to produce athletic performance. For example, in many team sports where a combination of endurance and sprint ability are required, at a truly elite level (where physiological monitoring and strength and conditioning support are of the highest standard) it should become possible to carefully modify the relative emphasis during training of the development of those two physiological characteristics according to athlete genotype. Thirdly, and probably more distantly in the future, some selection processes in sport might become informed by genomic information, although even in those cases the genomic information will always be secondary to the more informative phenotypic information.

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