The Ethics of Genetic Testing and Research in Sport

A Position Statement from the Australian Institute of Sport

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British Journal of
Sports Medicine
An international peer-reviewed journal of sport and exercise medicine

Ethics of genetic testing and research in sport: a position statement from the Australian Institute of Sport

OPEN ACCESS

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Published Online First 29 November 2016

Genetic research and testing advances

- 1999
  - Human Genome Project
  - Human genome mapped for the first time
  - Took 13 years
  - Cost $US2.7 billion
- 2017
  - Whole Genome Sequencing
  - Takes 3 days
  - Costs <$US1000

Genetic research and testing advances

- Personalised medicine based on genetics permeating all medical fields
  - Which medication / chemotherapy will work for this individual?
  - Predisposition to diseases, injuries
- Doctors do genetic testing every day
  - BRCA1 & BRCA2 gene for breast / ovarian cancer
  - HLA B27 for inflammatory joint disease
  - CY282 gene for haemochromatosis
  - FBN1 for Marfan Syndrome

The AIS Position

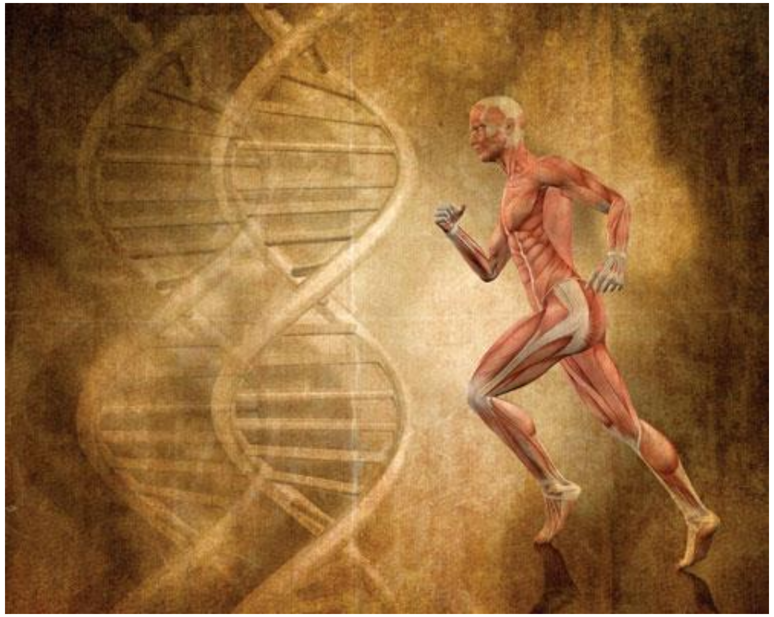
- Genetic testing for health related purposes will be ordered by a medical practitioner
- Genetic testing for health related purposes will be ordered in conjunction with genetic counselling

Genetic research advances

- Complex issues involved in genetic testing/research
  - Inadvertent discoveries
  - Ramifications for patient & patient’s family
  - Privacy and genetic information
  - Inappropriate use of genetic information (insurance companies, talent identification)
  - Issues with consenting to genetic research

Genetic research in sport

- Many projects underway worldwide
  - Genetics and injury
  - Genetics of power versus endurance
  - Studies into energy metabolism
- Currently the field is not well advanced
- Many ‘claims’ made in relation to genetics and aspects of sports performance do not have a good evidence base



The AIS Position

- Genetic testing as part of a research project, will only occur with the informed, written consent of participants
- The purposes for which genetic information will be used is clearly articulated to athletes
- The management of, and confidentiality pertaining to genetic testing results will be clearly articulated to the athlete, prior to the participation in research
- The results of genetic testing will remain confidential unless otherwise explicitly stated
- Participants in genetic research will be informed about the possibility of unintentional discoveries that could potentially impact on the participant’s health or the health of their relatives

The AIS Position

- Athletes have the right to decline a genetic test
- There will be no discrimination against athletes who decline genetic testing
- Genetic testing for the purpose of research in sport will not be conducted on athletes under the age of 18 years
- Athletes participating in genetic research have the right to withdraw from research at any time and/or have all of their material and/or results destroyed at any time during the process of testing or research
- Athletes participating in genetic research have the right to have their material and/or results sent to a third party.
- Genetic testing for research in athletes will involve the least invasive method of sample collection required to deliver the research outcomes

The AIS Position

- Clear guidelines must define the dissemination of genetic information before a research study or testing regimen is commenced

Talent identification

- Genetics and Talent ID began in the early 2000s
- Research supports an association between ACE gene, ACTN3 gene and human performance
- No predictive power

The AIS Position

- Genetic testing will not be used to include or exclude athletes from a high performance program
- Genetic testing will not be used as a method of talent identification

Injury risk

- Several genes have been implicated in increased risk of tendon, ligament and bone injuries
  - Collagen genes and tendon structure genes
  - Genes relating to tendon homeostasis
  - Genes relating to bone metabolism and growth
- Small numbers

The AIS Position

- Directing evidence-based interventions to reduce injury and improve health is a legitimate and valid use of genetic information

Genetic Testing – Direct to consumer (DTC)

- Rapid expansion of DTC genetic testing
- Offered to the public on a commercial basis – no input from medical practitioners
- Results are often accompanied with health/sport/nutrition advice
- Evidence supporting the 'advice' is often weak
- Over 20 companies world-wide – including those servicing Australia, some with a sport focus

AIS

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Our Tests

DNAeX sports specific genetic tests

DNAeX work exclusively with Partners and Exercise Professionals.

Research on twins indicates 66% of sport performance is genetic. Olympians and elite international athletes have been screened to identify the subtle variations in genes that are involved in performance. Certain muscle genes indicate a genetic predisposition for speed and power. Variations in other genes such are more likely to be found in the genetic profiles of elite endurance athletes.

Exercise genomics allows individual athletes to exploit genetic advantages to maximise performance and overcome potential genetic weaknesses. Genetic testing can identify individuals with optimal physiology and an enhanced capacity to respond to training. Event selection can be optimized, and training methods and nutrition are personalized to an individual's DNA.

DNAeX genetic tests are made up of many gene markers and crack the unique genetic code of elite athletes. DNAeX Fit-Test is the most advanced and accurate sport gene product in the world.

So what does all of that mean in plain English?

Refer an Athlete

Want to refer an athlete, but don't want to become a partner? Simply Contact Us and we will arrange everything for you.

Find a Partner

DTC – DNAex example reports

What this means for you

HIGH RISK
Injury
'Be Careful'

Meniscus Tear

NORMAL
Injury
Risk

Genes Tested

COL1A1 Result	GG	IL6 Result	GG
COL5A1 Result	TT	IL6R Result	CA
CRP Result	GG	TNF Result	GG
GDF Result	CT		

DTC – DNAex example reports

Int J Sports Med. 2014 Jun;35(7):625-8. doi: 10.1055/s-0033-1355417. Epub 2013 Nov 13.

The GDF5 SNP is associated with meniscus injury and function recovery in male Chinese soldiers.

Ge W[1], Mu J[1], Huang C[2].

Author information

Abstract

Genetic factor have previously been shown to play an important role in sports injuries and recovery. GDF5 Single-Nucleotide Polymorphism rs143383 has been recently reported to be associated with fracture susceptibility. Furthermore, the effect of GDF5 during the recovery processes of trauma is increased. In the present study, we aimed to evaluate whether this SNP was associated with susceptibility to the meniscus injury and postoperative recovery in Chinese male soldiers. GDF5 SNP was genotyped in 135 male soldiers with meniscus injury and 400 healthy male controls. Moreover, the function recovery of the soldiers suffering from the meniscal repair was also assessed. Our data showed that the GDF5 TT genotype (60.0 vs. 47.25%; P=0.010) and T allele (76.3 vs. 68.75%; P=0.019) were significantly over-represented in the meniscus injury group compared with the control group. We found that the TC (P<0.05), CC (P<0.05) and C carriers (P<0.05) genotype exhibited significantly higher Lysholm Scores than the TT genotype at 1 month postoperative. In addition, the CC (P<0.05) genotype also demonstrated significantly higher Lysholm Scores than the TT genotype 2 months postoperative. Taken together, our results revealed that the GDF5 SNP was associated with susceptibility to the meniscus injury and postoperative function recovery in Chinese male soldiers.

PMID: 24227118 [PubMed - indexed for MEDLINE]

DTC – DNAex example reports

Consume
LESS
Refined Carbs

Genes Tested

ACE Result	ID	ADRB2 Result	GA
PPARG Result	PRO-PRO	TCF7L2 Result	CT

DTC – DNAex example reports

J Nutrigenet Nutrigenomics. 2009;2(4-5):235-42. doi: 10.1159/000276991. Epub 2010 Mar 31.

Dopamine D2 receptor genotype (C957T) and habitual consumption of sugars in a free-living population of men and women.

Eny KM[1], Corey PN, El-Sohemy A.

Author information

Further

- Polyr
- Huma
- Gene
- Allelic
- Dopa
- Gene

n Study.

Abstract

BACKGROUND/AIMS: The dopamine D2 receptor (DRD2) has been implicated in modulating the rewarding effects of foods high in sugar. The purpose of this study was to determine whether a variation in the DRD2 gene affects habitual consumption of sugars in a free-living population.

METHODS: Caucasian men (n = 96) and women (n = 217) 20-29 years of age completed a 1-month food frequency questionnaire and were genotyped for the C957T polymorphism in the DRD2 gene. Analyses of covariance with post-hoc Tukey tests were used to compare nutrient intakes between genotypes adjusting for potential confounders.

RESULTS: Among men, consumption of sucrose was 60 +/- 6, 48 +/- 4, and 39 +/- 5 g/day for those with the CC, CT and TT genotypes, respectively, with a significant difference between the homozygotes (p = 0.03), suggesting an additive mode of inheritance. Among women, sucrose consumption was 42 +/- 4, 53 +/- 2, and 44 +/- 4 g/day for the CC, CT and TT genotypes, respectively, with CC and CT differing significantly (p = 0.02), suggesting a partial heterosis mode of inheritance. No differences were observed for protein or fat.

CONCLUSIONS: These findings suggest that genetic variation in DRD2 influences food selection and may explain some of the interindividual differences in sugar consumption.

DTC – DNAex example reports

Endurance Power	POWER and ENDURANCE ATHLETE	
Aerobic Potential	MEDIUM VO2 Max Potential	

Genes Tested

ADRB2 Result	GA	ACE Result	ID
ACTN3 Result	CC	AGT Result	Met/Met
COL5A1 Result	TT	BDKRB2 Result	DD
IL6 Result	GG	CRP Result	GG
PPARA Result	GG	NRF Result	GA
TRHR Result	AA	PPARGC1A Result	GG
VDR Result	TT	VEGF Result	GG

Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement

Nick Webborn,[1] Alun Williams,[2] Mike McNamee,[3] Claude Bouchard,[4] Yannis Pitsiladis,[5] Ildus Ahmetov,[6] Euan Ashley,[7] Nuala Byrne,[8] Silvia Camporesi,[9] Malcolm Collins,[10] Paul Dijkstra,[11] Nir Eynon,[12] Noriyuki Fuku,[13] Fleur C Garton,[14] Nils Hoppe,[15] Søren Holm,[16] Jane Kaye,[17] Vassilis Klissouras,[18] Alejandro Lucia,[19] Kamiel Maase,[20] Colin Moran,[21] Kathryn N North,[14] Fabio Pigozzi,[22] Guan Wang[5]

While further evidence will undoubtedly emerge around the genetics of sport performance in the future, the data are currently very limited. The *ACTN3* genotype is the most commonly tested by DTC companies. However, even for this genotype, its contribution to the degree of inter-individual variability in sprinting performance is trivial. Consequently, in the current state of knowledge, no child or young athlete should be exposed to DTC genetic testing to define or alter training or for talent identification aimed at selecting gifted children or adolescents. Large scale collaborative projects, such as the Athlome Project, may help to develop a stronger scientific foundation on these issues in the future but, currently, there is no place for DTC testing for predicting sports performance and talent identification.

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The AIS Position

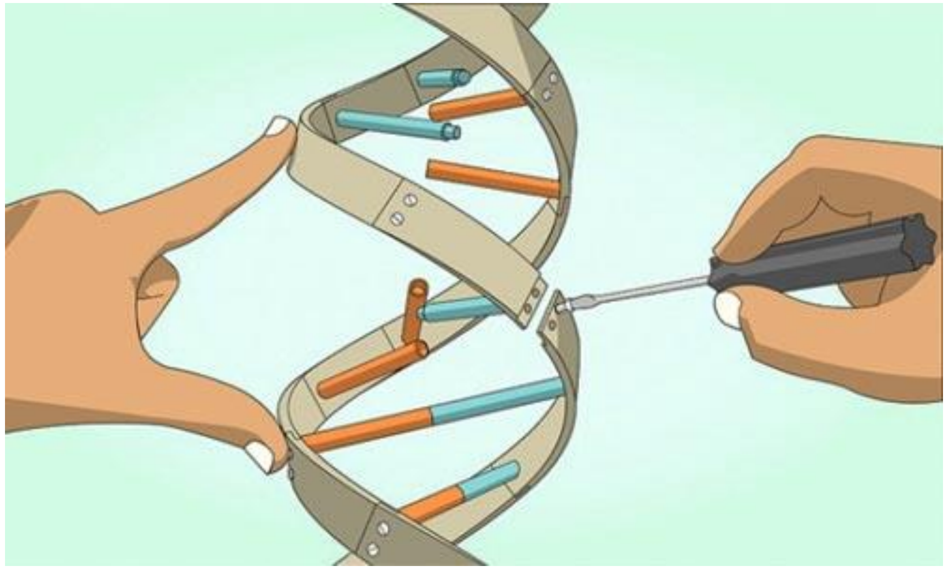
- Direct to consumer genetic testing in relation to sports performance is strongly discouraged
- Should individuals choose to undergo direct-to-consumer genetic testing they should be discouraged from acting on advice from the commercial company without seeking clarification from a medical practitioner

What does the future hold?

- Recent developments in gene editing:
  - CRISPR-Cas9
  - Cut and paste style of genetic manipulation
  - Cheap and ‘easy’
  - Rumoured to be used in some countries already
  - Athletes seen as potential ‘early adopters’ of this technique

The AIS Position

- Genetic manipulation will not be used for performance enhancement



Take home messages

- Currently, genetic testing has zero predictive power for sporting performance
- Much more work is needed in relation to genetic profiling for susceptibility to injury – bigger numbers and more ethnicities
- The AIS has published a position statement in relation to genetic testing of athletes and inclusion of athletes in genetic research

Position Statement

Vlahovich N, Fricker PA, Brown MA, Hughes DC. *Ethics of genetic testing and research in sport: a position statement from the Australian Institute of Sport* Br J Sports Med 2017;51: 5–11.