Genome-wide association study identifies a novel association between a cardiovascular gene polymorphism and superior athletic performance

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Aims

1. To carry out the largest metabolomics study in elite athletes to identify metabolic signatures of endurance sports.
2. To carry out the largest GWAS in elite athletes to identify genetic predisposition to high endurance sports.
3. To discover novel genetic loci affecting metabolites in elite athletes compared to previously published loci in non-elite athletes.
4. To discover novel genetic loci associated with endurance metabolites.

Methods

Study design

Elite athletes are competing athletes at national and international sport events who have their samples sent for doping tests at ADQ and FMU.

A pilot study comparing the metabolic profiles of elite-level athletes from different sporting disciplines

Conclusions

GWAS

Modulator of cardiac remodeling

Metabolomics

Sulfotransferase

mGWAS

VOCs

mGWAS

Sulfate

mGWAS

SULT2A1 (Sulfotransferase) regulates DHEA, thereby reducing downstream activation of DHEA to active androgens

Novel mQTLs associated with endurance metabolites

List of genes in eQTL with rs10952373 in the blood including their function and associated diseases

Aims

Introduction

- The superior physical performance of elite athletes is a multifactorial trait (environmental and genetic factors) (1).
- There is ample evidence of genetic influence of multiple genetic variants with small effect size over several phenotypic traits (2). However, not reproducible!
- GWAS in athletes versus non-athletes have uncovered new loci, none of which has reached GWAS significance (3,4).
- Thus, there is no concrete evidence of genetic predisposition of athletic performance due to small sample size, small effect size and complex phenotype (5).
- The advancement of metabolomics tools including mass spectrometry technologies has offered a unique opportunity to complement genomics data with intermediate phenotypes.
- Identified metabolites show direct functional association with genetic variants with a greater effect size (6).
- The integration of genomics and metabolomics technologies has also allowed a better chance to reveal genetic predisposition of complex metabolic pathways (7,8).

Metabolomics

Phenotype

GWAS

INTRODUCTION

Health monitoring

Exercise

Talent(Genetics)

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