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**Exome-wide association study of elite Jamaican and African-American sprint athletes**

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Elite human athletic performance is a multifactorial trait and previous candidate gene studies have been inconclusive.

**Purpose:** Two exome-wide association studies (EWAS) of elite Jamaican and African-American sprint athletes and respective matched controls were performed to identify common genetic variants.

**Methods:** 95 Jamaican sprint athletes and 102 Jamaican controls, 108 African-American sprint athletes were genotyped using the Illumina® HumanExome BeadChips. Standard EWAS quality control (QC) and population stratification correction were applied to the genotype data. Genetic associations were evaluated by logistic regression/standard allelic association analysis. Meta-analyses were performed for SNPs with association P-value  $< 5 \times 10^{-5}$  across the two sprint EWAS sample sets using a fixed-effects model. New significance level was re-defined based on the number of extra meta-analysis tests carried out.

**Results:** After QC, 96,698 autosomal non-synonymous SNPs in 88 Jamaican sprint athletes and 87 Jamaican controls, *ca.* 153,807 autosomal non-synonymous SNPs in 79 African-American sprint athletes and 391 African-American controls were available for analysis. The genomic inflation factor values were calculated for Jamaican and African-American EWAS sample sets, respectively. Various SNPs showed association with P-value  $< 5 \times 10^{-5}$  in the respective cohorts. Several SNPs remained significant after meta-analyses.

**Conclusion:** Two putative loci for elite sprint performance across Jamaican and African-American printers have been discovered using an exome-wide association approach followed by meta-analyses. Further validation of these signals requires replication before functional dissection can be carried out.