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Are genome-wide association study identified single-nucleotide polymorphisms associated with sprint athletic status? A replication study with 3 different cohorts

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Abstract

Purpose: To replicate previous genome-wide association study identified sprint-related polymorphisms in 3 different cohorts of top-level sprinters and to further validate the obtained results in functional studies. Methods: A total of 240 Japanese, 290 Russians, and 593 Brazilians were evaluated in a case-control approach. Of these, 267 were top-level sprint/power athletes. In addition, the relationship between selected polymorphisms and muscle fiber composition was evaluated in 203 Japanese and 287 Finnish individuals. Results: The G allele of the rs3213537 polymorphism was overrepresented in Japanese (odds ratio [OR]: 2.07, P = .024) and Russian (OR: 1.93, P = .027) sprinters compared with endurance athletes and was associated with an increased proportion of fast-twitch muscle fibers in Japanese (P = .02) and Finnish (P = .041) individuals. A meta-analysis of the data from 4 athlete cohorts confirmed that the presence of the G/G genotype rather than the G/A+A/A genotypes increased the OR of being a sprinter compared with controls (OR: 1.49, P = .01), endurance athletes (OR: 1.79, P = .001), or controls + endurance athletes (OR: 1.58, P = .002). Furthermore, male sprinters with the G/G genotype were found to have significantly faster personal times in the 100-m dash than those with G/A+A/A genotypes (10.50 [0.26] vs 10.76 [0.31], P = .014). Conclusion: The rs3213537 polymorphism found in the CPNE5 gene was identified as a highly replicable variant associated with sprinting ability and the increased proportion of fast-twitch muscle fibers, in which the homozygous genotype for the major allele (ie, the G/G genotype) is preferable for performance.

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Keywords

Athletes, Copine-V, Genetics, Sprint performance, Synaptic plasticity

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