
Abstract

A common genetic variation in the alpha-actinin-3 (ACTN3) gene causes a replacement of an arginine (R) with a premature stop codon (X) at amino-acid 577 (rs1815739). While the R allele has been found to be associated with power-oriented performance, the XX genotype may be linked with endurance ability. To test this hypothesis, we studied the distribution of ACTN3 genotypes in 155 Israeli athletes (age=35.9±12.2 years) classified by sport (endurance runners and sprinters) and in 240 sedentary individuals. The sprinters' allele frequencies (AF: R/X=0.7/0.3) and 577RR genotype distribution percentage (GD: RR=52%) differed markedly from those of the endurance athletes (AF: R/X=0.53/0.47, p=0.000007; GD: RR=18%, p=0.000009) and the control group (AF: R/X=0.55/0.45, p=0.0002; GD: RR=27.3%, p=0.000003). A comparison between the top-level and national-level sprinters revealed that the R allele occurs more often in the top-level sprinters. A significantly higher proportion of the XX genotype was observed in endurance athletes (34%) compared with controls (18%, p=0.02) and sprinters (13%, p=0.002). However, top-level and national level endurance athletes had similar allele and genotype frequencies. We conclude that the ACTN3 R allele is associated with top-level sprint performance and the X allele and XX genotypes may not be critical but rather additive to endurance performance.