ACTN3 R577X POLYMORPHISM IN MARATHON ATHLETES

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INTRODUCTION

The effect of the ACTN3 genotype has mainly been studied in elite athletes, based on the hypothesis that its influence on muscle function would be most readily observable at the extremes of human performance. The X allele tends to be overrepresented in those humans with an ‘extreme endurance phenotype’, i.e. elite endurance athletes (1-2). The aim of this study was to examine the genotype distribution of the R577X polymorphism (rs1815739) in α-actinin-3 (ACTN3) gene among marathon athletes.

METHODS

Participants of the study were 173 men athletes with best personal time <3h in marathon (mean age=43.06 years). The control group consisted of 216 non-athletes male young adults (mean age=20.73 years). Genotyping was performed by polymerase chain reaction. We used the chi-square test to determine whether the genotypic frequencies of the ACTN3 R577X genotypes differed between groups and logistic regression to calculate the odds ratio for being a marathoner based on the aforementioned genotypes. The SPSS 18.0 program was used for all statistical analyses.

RESULTS

Genotype allele frequencies were similar between marathon athletes and control groups (P>0.05). We did not find an association between the ACTN3 R577X polymorphism and the likelihood of being an athlete marathon using the dominant (RR vs. RX+XX) and the recessive model (RR+RX vs. XX).

DISCUSSION

The ACTN3 R577X polymorphism is not associated with marathon athlete status, at least in the cohort we studied. These results were theoretically unexpected, given the role of α-actinin-3 on skeletal muscle phenotypes, particularly muscle endurance and the importance of this phenotype for marathon performance.

REFERENCES