

## 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  $\alpha$ -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  $\alpha$ -actinin-3 on skeletal muscle phenotypes, particularly muscle endurance and the importance of this phenotype for marathon performance.

### REFERENCES

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