

PRELIMINARY FINDINGS OF α -ACTININ-3 GENE DISTRIBUTION IN ELITE TURKISH WIND SURFERS

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ABSTRACT

A common polymorphism in the α -actinin-3 (*ACTN3* R577X) gene represents one of the most widely examined variations in terms of performance and genetic predisposition to certain sports. The aim of the present study was to examine the *ACTN3* R577X polymorphism in elite Turkish wind surfers. The genotyping procedure was carried out by polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP). Five male and three female wind surfers, eight elite wind surfers in total, were enrolled in the study. Five of the surfers had RX, two had XX and one had RR genotypes. Previous findings indicated that the X allele was the endurance allele. Our findings were in agreement with the previous reports. Seven of our subjects had at least one copy of the X allele that was considered to have a tendency to prolong endurance. Our preliminary results must be supported with further studies in greater numbers of subjects to clarify the effect of gene polymorphism.

Keywords: α -Actinin-3 (*ACTN3*) gene, Polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP), Performance, Polymorphism, Sports genetics.

INTRODUCTION

α -Actinin-3 (*ACTN3*) is one of the most examined and discussed structural gene in sports science. This gene codes for the major and important structural proteins that are located within sarcomeric Z lines, there they anchor actin filaments, maintain myofibrillar arrays and have roles in signaling functions [1,2]. The mononucleotide transition at position 1747 (C>) in the *ACTN3* coding sequence leads to the change of amino acid arginine (R) to a stop codon (X) in codon 577 (R577X), producing a short form of protein. The short form of the protein does not contribute to any known disease, and over 1 billion people are considered to have the mutant genotype [3].

Yang et al. [4] first reported the evidence of *ACTN3* influence on elite athletic performance. They associated the *ACTN3* R allele and RR genotype with top-level and power-orientated athletic performance and also hypothesized that total deficiency of the *ACTN3* protein may confer some beneficial effect on endurance performance. The presence of the R allele in high power muscle contraction was also reported by Delmonico *et al.* [5].

In this report, we analyzed the *ACTN3* genotype distribution in elite Turkish wind surfers, who represented the National Team and won gold medals in the last 3 years. We tried to find out the association between allelic types of the *ACNT3* gene in elite wind surfers.

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MATERIALS AND METHODS

Eight National Team wind surfers and medal winners, were enrolled in an *ACTN3* genotyping procedure. The study procedure was in accordance with the principles of the Declaration of Helsinki II and all subjects provided written informed consent prior to enrollment.

DNA isolation was carried out by using high pure polymerase chain reaction (PCR) Template Preparation Kit (Roche Diagnostics, Mannheim, Germany). The region of interest of the gene was amplified by using the following primers: forward, 5'-CTG TTG CCT GTG GTA AGT GGG-3' and reverse, 5'-TGG TCA CAG TAT GCA GGA GGG-3'. Polymerase chain reaction was performed by initial denaturation at 95°C for 5 min., followed by 35 cycles of denaturation at 95°C for 30 seconds, annealing and extension at 72°C for 1 min., and a final extension for 7 min. at 72°C. The 290 bp length amplicons were digested by *DdeI* (New England Biolabs, Beverly, MA, USA) in a condition recommended by manufacturer. The wild-type allele, 577R, showed fragments of 205 and 85 bp on 10.0% polyacrylamide gel electrophoresis (PAGE), whereas the variant allele, 577X, showed fragments of 108, 97 and 85 bp. Visualization of the digested fragments was performed by staining with ethidium bromide.

RESULTS

Genotype determination was provided by restriction fragment length polymorphism (RFLP) patterns that the amplicons displayed on PAGE (Figure 1). Five of the surfers were heterozygous for the RX genotype for the examined polymorphism. Two of them were homozygous for the XX genotype for the same polymorphism and only one was homozygous for the wild-type RR genotype. When we examined the genders according to their genotypes, four of the males had the XY genotype and only one had the XX genotype. Of the three females, one had XX, one had XY and one had the RR genotype. Genders and genotypes of the subjects are summarized in Table 1.

DISCUSSION

Yang *et al.* [4] initiated the discussion about *ACTN3* influence on elite athletic performance and was soon followed by others [1,3,6]. To date, sev-

Table 1. Genders and genotypes of the wind surfers (M: male, F: female).

Wind Surfer	Gender	Genotype
1	M	XR
2	M	XR
3	F	RR
4	M	XX
5	M	XR
6	F	XX
7	F	XR
8	M	XR

eral polymorphism determination studies have been done by many researchers in order to find the suitable genotype in different sports. Some of these studies suggest the RR and RX genotypes as having advantages in power sports and correlated positively with strength/power athletes [6-8]. In contrast, Norman *et al.* [9] suggested that individuals with the XX genotype who had no wild-type *ACTN3* expression, may benefit endurance performance due to evolving type I muscle fibers. Additionally, a shift in the metabolic path-way, promoting aerobic performance, was reported in *ACTN3* knockout mice [10]. There are also other studies showing the association between the *ACTN3* XX genotype and endurance athletic capacity [4,11,12]. In our study group, seven of the surfers had at least one X allele, five of them were heterozygous

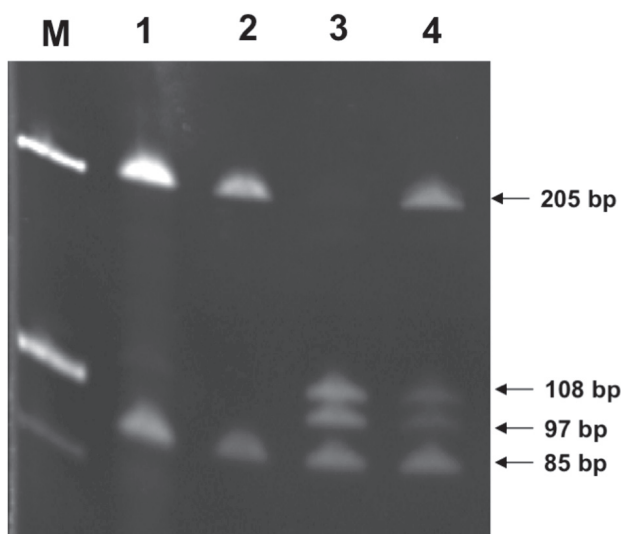


Figure 1. RFLP results of *ACTN3* gene in PAGE analysis; M is the 100 bp marker, Lane 1 and 2 are RR genotypes whereas lane 3 is XX and lane 4 is RX genotypes.

and two were homozygous, and these results are in agreement with the latter studies. On the other hand, there are some other reports in which the *ACTN3* XX genotype is not associated with endurance sports [13-15]. In our cohort, only one surfer had the RR genotype, which was in agreement with these studies.

To the best of our knowledge, there are no reports that examined the *ACTN3* polymorphism in surfers and also in Turkish wind surfers. Sanlısoy *et al.* [16] examined the R577X polymorphism in the elite athletes of the Aegean region in Turkey, and reported a significant difference between elite athletes and sedentary individuals.

The main limitation of the study was the low number of elite Turkish wind surfers. In this report, we analyzed eight National Team medal winner wind surfers. This limited number of elite surfers represents the developing nature of professional wind surfing. The interest in wind surfing is growing and within a few years, the number of the individuals competing in wind surfing events is expected to increase.

In conclusion, the X allele of the *ACTN3* plays an important role in the eight elite Turkish wind surfers. More-over, it is difficult to predict an individual's sporting ability based solely on only one type of polymorphism. In future, with the improvement in molecular science and by the addition of related genetic variations, genetic testing will play more important roles for deciding the proper type of sports. However, due to the limited number of examined subjects, we are unable to comment on the effect of *ACTN3* on Turkish wind surfers. We hope this preliminary finding will contribute to the literature about the effect of the *ACTN3* gene in surfing and will be supported by further studies.

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