

# ACE I/D Polymorphism Determination in Turkish Elite Wind-surfers.

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**I**nsertion/ deletion (I/D) genetic polymorphism of the angiotensin converting enzyme I gene (ACE) represents the first and the most investigated genetic indicator of athletic performance in humans. In this report, we aimed to analyze the distribution of ACE I/D polymorphism in elite Turkish wind- surfers. DNA samples from the surfers were isolated from peripheral blood and standard PCR application was carried out in order to genotype the polymorphism. Of the 8 surfers (5 males and 3 females), 3 had I/I genotype (2 males and 1 female), 4 had I/D (2 males and females) and 1 was D/D (female). Direct allelic count revealed the I allele as 10 and D allele as 6 in our study cohort. According to our results, I allele was counted more than D allele, which is in agreement with the previous studies, indicating the importance of I allele in predisposition to endurance activities. We conclude that determining *ACE* I/D polymorphism will be a good biomarker in the terms of predisposition to certain kind of sports. However, these results must be extended with further studies in order to have more precise information about the effect of *ACE* I/D polymorphism on human performance.

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## Introduction

Angiotensin converting enzyme gene (*ACE*) is the one of the most frequently studied gene in predisposition to sports activity and elite athlete status. *ACE* is localized on 17q23 and codes for the enzyme which is a zinc membrane-bound metalloproteinase. 287 base pair *Alu* repeat sequence in intron 16 creates two polymorphic variant of *ACE*, presence of the repeats (insertion, I) or the absence (deletion, D) of the repeats (Cieszczyk et al., 2010). According to the presence or absence of the repeats, *ACE* genotype has 3 variants, I/I and D/D homozygotes and I/D heterozygotes. Most of the studies reported that D allele is associated with increase in blood pressure due to the elevated blood or tissue ACE enzyme levels, whereas I allele has the reverse effects. Additionally, I allele is associated with the increase in the proportion of type I muscle fibers and increase in muscle endurance (Zhang et al., 2003).

To date, most of the case- control studies suggested that individuals with *ACE* D/D genotype are much more successful in speed- strength disciplines, like short distance running, long jump, high jump, discus throw or short distance swimmers (Woods et al., 2003; Costa & Slocombe, 2012). On the other hand, individuals with I/I genotype has a lower ACE serum concentration and have more success in endurance related disciplines like medium and long distance running, race walking and skiing (Holdys et al., 2001).

In this report, we performed solely *ACE* genotype distribution in elite Turkish wind-surfers, who represented National Team and achieved gold medals in the last five years, and tried to find out the allelic distribution of *ACE* gene in elite wind- surfers.

## Materials and Methods

A total of 8 National Team and medal winner wind- surfer (5 males and 3 females) volunteers were enrolled in the present study. 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. All participants were of Turkish ancestry. DNA isolation from 200  $\mu$ l venous blood was carried out by using High Pure PCR Template Preparation Kit (Roche Diagnostics, Mannheim, Germany).

*ACE* genotypes were determined by PCR amplification of genomic DNA with sense primer 5'-CTGGAGACCACTCCCATCCTTTCT-3' and antisense primer 5'-GATGTGCCATCACATTCGTCAGT-3'. We did not have any difficulties in determining genotypes by using these primers and we did not do

the second PCR analysis in order to confirm the results. 50 µl PCR mixture contained 50-100 ng genomic DNA, 1 mM of each primer, 50 mM KCl, 1 mM dNTP, 1.5 mM MgCl<sub>2</sub>, 10 mM Tris-HCl (pH 8.0), and 1U *Taq* DNA polymerase. The first cycle had a denaturation at 94°C for 5 min, annealing at 58°C for 1 min, and extension at 72°C for 2 min. After the first cycle, we had 30 cycles with the conditions as: denaturation at 94°C for 1 min, annealing at 58°C for 1 min, and extension at 72°C for 2 min, followed by a final elongation at 94°C for 1 min, annealing at 58°C for 1 min, and extension at 72°C for 7 min. Amplicons were separated by 2% agarose gel electrophoresis and visualized under ultraviolet light after ethidium bromide staining. The amplicons are 190 bp fragments in the presence of a deletion (D) allele, and a 490 bp fragment in the absence of a deletion (I) allele (Figure 1).

## Results

Of the 8 wind- surfers, 3 surfers had the I/I genotype, 4 the I/D genotype and only 1 had the D/D genotype. When we examine the genotype results according to their genders, 2 of the 5 males had I/I genotype, again 2 had the I/D genotype and only 1 had the D/D genotype. In female subjects, one had the I/I genotype and 2 had I/D genotype. The number of alleles were six and four for the I and D alleles, respectively, in males and four and two for the I and D alleles, respectively, in females. The overall study group had ten I and six D alleles for the examined polymorphism.

## Discussion

*ACE* has been widely studied in many populations in the terms of predisposition to sports activities. Most of the reports, to date, suggest the D allele as a factor for the increased power for anaerobic dominated activities due to the higher *ACE* activity. Montgomery et al. (1997) reported the individuals with D/D genotype had a significantly increased left ventricular mass in response to physical training. These findings were confirmed with further studies, Nagashima et al. (2000) showed the association of D allele in 308 Caucasian soldiers and, Fatini et al. (2000) showed similar results in 28 professional football players. There are also other reports indicating the association of D alleles with sprint-based activities, Amir et al. (2007) in 121 Israeli sprinters, Woods et al. (2001) in short distance swimmers and Cam et al. (2005) in Turkish sprinters.

*ACE* I allele, on the other hand, is mostly associated with endurance-based activities, due to the lower *ACE* activity, predominantly seen in aerobic pathways. Researchers have shown the I allele is associated with high response to physical training and improved muscle efficiency (Myerson et al., 1999). There are sev-

eral reports demonstrating the association of I allele with endurance activities; Montgomery et al. (1998) in high altitude mountaineers, Alvarez et al. (2000) in Spanish athletes, Collins et al. (2004) in 447 triathlon athletes and Gayagay et al. (1998) in 64 Olympic rowers.

As of our best knowledge, this report is the first in wind- surfers in the terms of *ACE* determination. Wind- surfing requires a high endurance capacity, in which the surfer deals with the weather conditions and race physiology, which can extend up to hours. Recently, Ulucan et al. (2013) examined the *ACTN3* allele distribution in the same study cohort, and reported the X allele, known as endurance allele, was higher than the R allele. In this report, we examine 8 national team surfers in the terms of *ACE* polymorphism, all had at least one medal in national levels. Our results are in agreement with the others, indicating the presence of I allele in endurance activities. In the present study, we had 3 *ACE* I/I genotypes, 2 were male and one was female. When we count the alleles, again I allele was predominating the surfers examined in the study. In our study group, only one had *ACE* D/D genotype. 7 of the 8 surfers had at least one I allele, which may force them to be successful in wind- surfing.

There are enough information relating to *ACE* polymorphism and the use of this polymorphism for identification of elite individuals in different kinds of sports in humans. But there are no information about the *ACE* polymorphism and Turkish wind- surfers. According to our results, we can suggest that determining *ACE* polymorphism in wind- surfing is an important genetic biomarker and should be considered in a genetic testing panel that can be designed to identify elite individuals. But this study should be replicated in other populations in order to have more precise information about the effect of this polymorphism.

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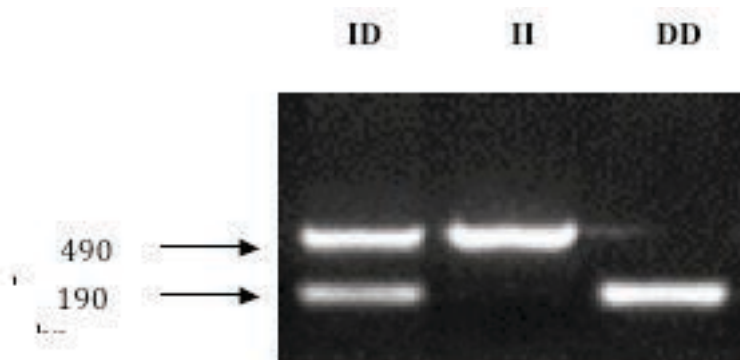
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*Figure 1.* Agarose gel images of ACE polymorphisms