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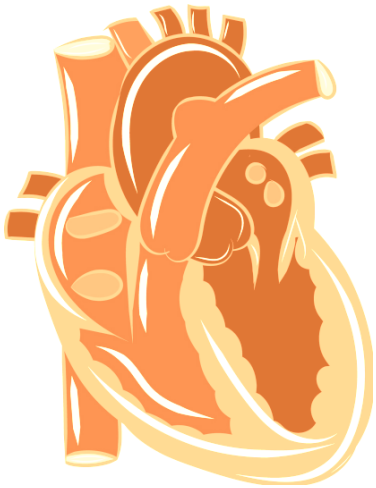
ANGIOTENSIN I CONVERTING ENZYME (ACE)

ACE and Winning the Race

Biology Background

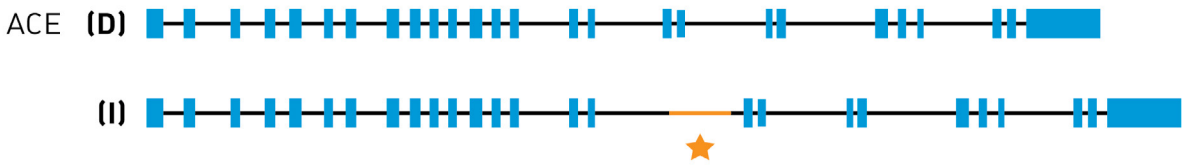
- The ACE gene produces the protein angiotensin-converting enzyme (ACE), which functions as a protease that cuts other proteins.
- ACE plays a central role in the system that controls blood pressure by regulating the volume of fluids in the body.
- ACE is located within the cell membrane.
- ACE is made in nearly all tissues of the human body, but appears to be most strongly expressed in capillaries.



Blood Vessels

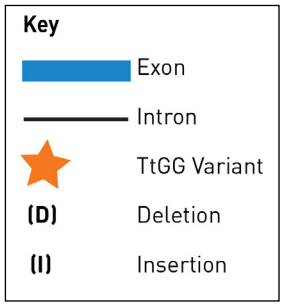
Genomic Locus

The ACE gene is located on chromosome 17 of the human genome. The ACE gene is 21,313 base pairs in length and consists of 26 exons and 25 introns.

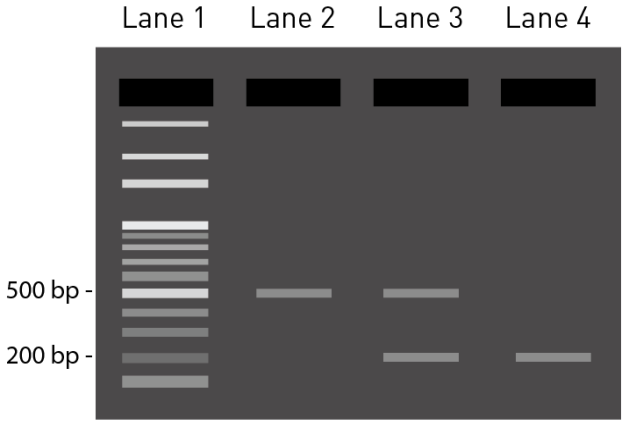


The TtGG Variant

- In this assay, you are studying one polymorphism, or variant, in ACE called an indel (short for **in**sertion/**del**etion) that is situated within intron 16 of the ACE gene (see star).
- The insertion allele (I) includes an insertion of a 287 base pair transposable element in intron 16. The deletion allele (D) does not include the 287 base pair transposable element.
- Since this insertion is in the intron of the gene, it does not directly affect the amino acid sequence of the protein.



ACE Gel



Lane 1: DNA ladder
Lane 2: Homozygous I genotype, 500 bp
Lane 3: Heterozygous I/D genotype, 200 bp, 500 bp
Lane 4: Homozygous D genotype, 200 bp

Population Genetics

- The indel variant located within intron 16 of the ACE gene has been studied for its contribution to physical endurance.
- This variant seems to affect enzymatic activity, possibly due to differences in protein levels circulating in blood plasma.

Influence on Human Health

- The presence of the insertion allele has been associated with improved endurance performance in studies of mountaineers and soldiers.
- These effects are attributed to increased mechanical efficiency in muscles, possibly due to an increase in type 1 muscle fibers.
- Variants in ACE have also been associated with heart disease, but have not been proven to cause heart disease. Heart disease is a complex disorder that has many different genetic and environmental influences.
- Remember that association does not mean causation. While ACE variants are associated with certain phenotypes and diseases, the ACE alleles do not necessarily cause those phenotypes and diseases.

Sources

» Online Mendelian Inheritance in Man (OMIM) http://www.omim.org/entry/106180
» National Center for Biotechnology Information (NCBI) Gene http://www.ncbi.nlm.nih.gov/gene/1636
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» Review on ACE and link to athletic performance: Puthucheary et al. The ACE Gene and Human Performance. Sports Medicine (2011)
» Human Protein Atlas
» UCSC Genome Browser