



ampli set ACE^{CE IVD}

45 tests

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detection of polymorphism (I/D) OF the gene encoding for Angiotensin I - Converting Enzyme (ACE).

The development of thrombotic disease is one of the major cause of morbidity and mortality . An alteration of homeostasis is the main mechanism of thrombosis. The cause of this unbalance may be genetic.

It underlines the importance of the interaction between genes and environment in the thrombotic pathologies. Many genes have been correlated with the risk of atherosclerosis and thrombotic pathologies. Among these , polymorphisms of the gene encoding for Angiotensin I –converting Enzyme (ACE) have been correlated with to coronary disease and myocardial infarction. ACE is a key enzyme in the regulation of blood pressure. It is responsible of conversion of angiotensin I to angiotensin II, a potent vasoconstrictor and involved in the cellular proliferation , causing atherosclerosis. Many studies suggest that almost 50% of variability of plasmatic level of ACE enzyme depends on the polymorphism insertion/deletion (I/D) in intron 16 of the gene. I and D Alleles are linked to the presence or absence of a fragment of 287 bp in this intronic region. The presence of D allele is joined to a major plasmatic level of the enzyme and to a major risk of hypercoagulability and endothelial damage.

The research of polymorphism (deletion/insertion in the intron 16) is performed with the amplification with specific primers followed by detection on 2% agarose gel . The presence of the D Allele produces a fragment of 190 bp, whereas the presence of the I allele produce a fragment of ~ 490 bp.

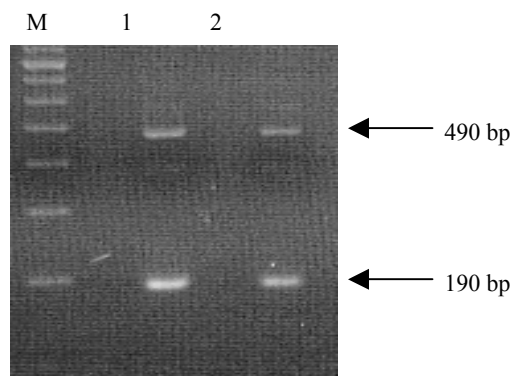
Principle of Assay: A) extraction of genomic DNA; B) amplification; C) detection on agarose gel.

Applicability: on extracted and purified genomic DNA from whole blood samples.

Stability: over 12 months if correctly stored (Agarose gels, if protected by light, can be stored 1 year at room temperature).

ANALYSIS OF RESULTS

The product of PCR of Allele D is a fragment of 190 bp. The product of PCR of Allele I is of □ 490 bp. Samples 1 and 2 are heterozygote I/D



References

- J. Clin Invest.* (1990); 86:1343-46.
- Am. J. Hum. Genet.* 51: 197-205 (1992).
- Molecular and Cellular Endocrinology* 107 (1995) 215-219.
- Blood*, vol. 95, 5, 2000 pp. 1517-1532.
- Atherosclerosis* 157 (2001) 57-64.
- Am Heart J* (2000) 140 (5): 760-5