



ampli set β Fibrinogen-455G/A^{CE IVD} 45 tests cat 1352
detection of 455G/A polymorphism of the β -Fibrinogen gene

The development of thrombotic pathologies is one of the major cause of morbidity and mortality. The alteration of haemostatic system is the central mechanism of the thrombotic events. The cause of the alterations may be also genetic. It underlines the importance of the interaction between genes and environment in the thrombotic pathologies. Fibrinogen is a glycoprotein dimer where every chain is made of three polypeptide chains α , β and γ with a molecular weight of 340000 D. The genes encoding for the three chains are located in a cluster of almost 50 kb on the long arm of chromosome 4 (q23-q32). Many mutations are reported in the promoter of the gene encoding the β chain of Fibrinogen (polymorphism Bcl I and -455 G/A). The synthesis of this chain may be the restrictive step in the construction of the total protein complex of Fibrinogen. Therefore, genetic polymorphism regulating the synthesis of β chain are very interesting. Particularly, the presence of allele 455A (20-25% of European population) of polymorphism -455 G/A causes an increase of plasmatic levels of Fibrinogen.

The detection of the G455A polymorphism is performed with an amplification using specific primers of a fragment of 1300 bp, following by a restriction section due to *Hae III*.

Principle of Assay: A) extraction of genomic DNA; B) amplification; C) enzymatic digestion; D) 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 yield of amplification is a fragment of 1300 bp. The next restriction section made by the *Hae III* enzyme can be done the following results:

1 Absence of mutation Normal subject	2 Presence of mutation Heterozygote subject	3 Presence of mutation Homozygote mutated subject
3 fragments	4 fragments	3 fragments
575 bp	957 bp	957 bp
383 bp	575 bp	
343 bp	383 bp	
	343 bp	343 bp

References

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