ampli set FV Y1702C CE IVD 45 tests cat 1316

detection of Y1702C polymorphism of the Factor V gene.

The heterogeneity of clinic phenotype and the variability of thrombotic events showed by patients with familiarity for thrombotic disease have led to the hypothesis that the predisposition to these type of disorders may be due to many genetic factors. Recently, a complex haplotype of Factor V (HR2), which includes 13 different polymorphisms, has been reported. Among them, 7 cause an amino acid substitution and a functional modification of the protein, leading to an excess of plasmatic isoform FV1 concentration, more thrombogenic.
It isn't clear if haplotype HR2 alone could be a factor of thrombotic risk. It is sure that the risk of clinical thrombotic events in subjects carriers of the F V Leiden mutation is increased.
The detection of the Y1702C polymorphism is carried out starting with an amplification using specific primers of a fragment of 120 bp, following by a restriction section due to *Acc* I.The mutation is confirmed by the loss of a cleavage site for the enzyme *Acc I*.

Principle of method: 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.

ANALYSIS OF RESULTS

The yield of amplification is a fragment of 703 bp. The next restriction section made by the *Rsa I* enzyme can be done the following results:

1	2	3
Absence of mutation Homozygous Normal Patient	Presence of mutation Eterozygosis Mutant patient	Presence of mutation Homozygous mutant patient
1 fragment	3 fragments	2 fragments
703 bp	**703 bp**	
	493 bp	**493 bp**
	210 bp	**210 bp**

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