ampli set FII CE IVD 45 tests cat 1310

detection of G20210A polymorphism of the prothrombin gene

In 1996 a single mutation in the 3'-untranslated region of the prothrombin gene was reported, resulting in a G-to-A substitution. This polymorphism is joined to an increased level of plasmatic prothrombin . This polymorphism changes the efficiency of the "processing" and the stability of his own RNA. The 20210A allele has been found in 18% of a population of patients selected for individual or familiar venous thrombotic events, in the 6,2% f patients with a first event of deep venous thrombosis and in 2,3% of health control subjects. The risk of thombotic event associated to this polymorphism is 2,8 greater than associated to to the 20210G allele. The detection of mutation G20210A is carried out using the amplification with specific primers of a fragment of 223 bp, followed by restriction section by Hind III enzyme. The gain of a restriction cleavage shows the presence of the mutation. The normal allele produces a fragment of 223 bp, whereas the mutated allele produces fragments of 197bp and 26 bp..

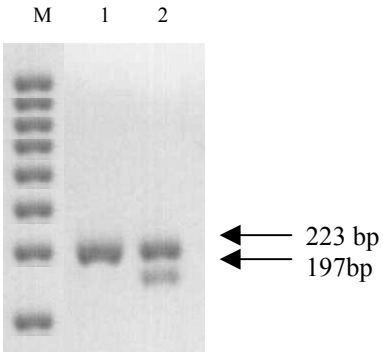
Principle of method: A) extraction of genomic DNA; B) amplification; C) enzymatic digestion; D) detection on agarose gel

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

ANALYSIS OF RESULTS

The yield of amplification is a fragment of 223 bp. **The wild type allele produces a fragment of 223 bp, whereas the mutated allele produces fragments of 197bp and 26 bp..**

1	2
No mutation HOMOZYGOTE NORMAL SAMPLE 1 fragment 223 bp	HETEROZYGOTE MUTATED SAMPLE 3 fragments 223 bp 197 bp 26 bp



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