



ampli set PAI-1 ^{CE IVD}

45 tests

cat 1351

detection of 4G/5G polymorphism in the promoter of the gene PAI-1

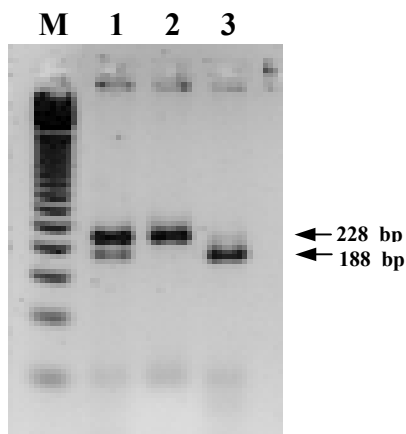
The development of thrombotic pathologies is one of the major cause of morbidity and mortality .The alteration of haemostatic system is the central mechanism of the thrombotic events. The causes of this alterations may be also genetic. It underlines the importance of the interaction between genes and environment in the thrombotic pathologies. The inhibitor of the activator of Plasminogen (PAI-1) is the main inhibitor of the activators of plasminogen (t-PA and u-PA). An increase or a decrease of PAI-1 can be responsible of thrombotic or haemorrhagic events respectively. Recently, the detection of a polymorphism in the promoter region of the PAI-1 gene has lead to evaluate PAI-1 as a factor risk of venous thrombosis. Particularly, the insertion/deletion of a residue of Guanosine in the region 675 of the promoter of the gene, named **4G/5G**, has been related with an increase of the activity of PAI-1. This polymorphism involves the bond of nuclear proteins responsible of the transcription of the gene PAI-1. The site **4G** may links only an "enhancer" region, whereas the allele **5G** links both the "enhancer" region and the "suppressor" one, resulting in a lower level of transcription. "In vivo" studies show that levels of PAI-1 are 25% higher in homozygotic subject for the allele **4G** than in homozygotic subject for allele **5G**. Homozygosis for the allele **5G** is present in 25% of normal population, and in 10% of pathological population for thrombotic events. The relationship between polymorphism **4G** and the risk of venous thrombosis needs of further investigation. Probably, the presence of the homozygosis **4G** becomes a risk factor when is joined to other genetic or environmental risk factors. The detection of **4G/5G** PAI-1 polymorphism is performed with the amplification with specific primers of a fragment of **228 bp**, followed by restriction section due to *Eci I* enzyme. The **5G** polymorphism introduces a restriction site for the enzyme. The product of PCR of the allele **4G** isn't cut, whereas the allele **5G** produces two fragments of **188bp** and **40 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 228 bp. The next restriction section made by the *Eci I* enzyme can give the following results:



M = Marker 50bp ladder
 1) = Heterozygosis **4G/5G**
 2) = Homozygosis **4G**
 3) = Homozygosis **5G** (control DNA)

Heterozygote 4G/5G	Homozygote 4G	Homozygote 5G
228 bp 188 bp 40 bp	228 bp	188 bp 40 bp

References

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