



ampli set FVII R-353-Q ^{CE IVD} **45 tests**
 detection of R-353-Q polymorphism of the factor VII gene.

cat 1312

Factor VII coagulation factor is a vitamin-K dependent protease that plays an important role in the extrinsic system of coagulation. It is synthesized by the hepatic cells and it is secreted in the inactivated form as single peptide chain. The activation (FVIIa) consists of a proteolytic snip followed by the disulfide bond of two peptide chains. Genetic and environmental factors can influence the plasmatic levels of Factor VII. Particularly, the third part of the causes of the variations of its plasmatic level may due to genetic polymorphism of the gene encoding FVII. The more common polymorphisms are: 1) the substitution G-A in the exon 8 of the gene, responsible of the substitution R(arginine) – Q(glutamine) in the amino acid 353. Particularly, the homozygosis 353 Arg-Arg is related to the presence of higher plasmatic level of F VII. 2) The insertion/deletion of 10 nucleotides in position -323 in the promoter region of the gene(-323 P0/P10). Recently, other polymorphism are been identified in the promoter region of the gene:1)substitution T-C in position -122 (-122 T/C),2) the substitution G-T in position -401 (-401G/T) ;3) the substitution G-A in position -402 (-401 G/A). In 1999 has been demonstrated that the more rare polymorphic variants -401T and 402A are associated respectively to a decreased and to an increased transcriptional activity of the gene. It is evident that the polymorphic form 353Q and -401T may be considered as protective against thrombotic events. The **ampli set FVII R-353-Q** allows the detection of the polymorphism R(arginine)-Q (glutamine) of the amino acid 353. The detection of the mutation is carried out performing the amplification with specific primers of a fragment of 312 bp, followed by restriction section due to MspI enzyme. The allele 353 Arg (the more common) shows an additional cleavage site, as the MspI enzyme digests the product PCR in three fragments of 205, 67 and 40 bp. The product of PCR of the rare allele 353 Glu is digested in two fragments of 272 and 40 bp.

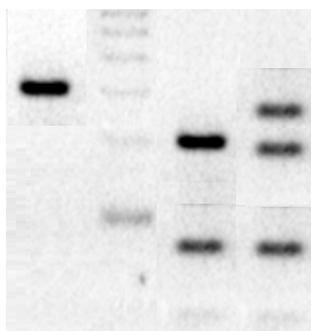
Principle of method: A) extraction of genomic DNA; B) amplification; C) enzymatic digestion; D) detection on agarose gel

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

ANALYSIS OF RESULTS

The yield of amplification is a fragment of 312 bp. The next restriction section made by the MspI enzyme can be done the following results:

PCR Marker 1 2



1	2	3
Homozigotic subject	Heterozigotic subject	Homozigotic subject
353 Arg/Arg	353 Arg/Glu	353 Glu/Glu
3 bands	4 bands	2 bands
	272 bp	272 bp
205 bp	205 bp	
67 bp	67 bp	
40 bp	40 bp	40 bp

References

Arteriosclerosis and Thrombosis 1991, 11, 3:540-6.
Hum Genet 1993, 90:574-576.
Arterioscler Thromb Vasc Biol 1996, 16: 72-76.
Thromb Haemost 1998 : 80: 281-5.
Blood, Vol 93, N. 10, 1999: 3432-3441.