



**ampli set F XIII Val34Leu<sup>CE IVD</sup> 45 tests**  
 detection of Val34Leu polymorphism of the Factor XIII

**cat 1317**

The development of thrombotic disease is one of the major cause of morbidity and mortality . An alteration of homeostasis is the main mechanism of thrombosis. The cause of this unbalance may be genetic.

Factor XIII of coagulation is a tetramer made by two subunits A and two subunits B (A<sub>2</sub> B<sub>2</sub> ). Subunits A have a trans-glutamine enzymatic activity, that is activated by thrombin. Factor XIII plays an important role in coagulation and in fibrinolysis; it is responsible of the stabilization of the clot of fibrin with the production of covalent bonds between the  $\alpha$  and  $\beta$  chains. Recently, many polymorphism involved in different degrees of activity of FXIII have been reported. Polymorphism C/T leading to the substitution Val34Leu in position 34 of the A chain of Factor XIII seems to protect against thrombotic diseases.

The detection of Val34Leu polymorphism is performed with an amplification with specific primers of a fragment of 192 bp, followed by a restriction section due to *Dde I* enzyme.

**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.

**ANALYSIS OF RESULTS**

The yield of amplification is a fragment of 192 bp The next restriction section made by the *Dde I* enzyme can be done the following results:

1 Absence of mutation	2 Presence of mutation Heterozygote subject	3 Presence of mutation Homozygote subject
1 fragment	3 fragments	2 fragments
192 bp	192 bp 161 bp 31 bp	161 bp 31 bp

**References**

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