



## KIT FOR THE DETECTION OF VAL34LEU POLYMORPHISM OF THE FACTOR XIII

**AMPLI-set-FXIII Val34Leu**

**Cat.n. 1.317**

The development of thrombosis 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-γ-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 α and γ 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.

**Tests:** 45

### REAGENTS AND STORAGE

| <u>AMPLIFICATION</u>     |       |
|--------------------------|-------|
| PCR mix                  | -20°C |
| H <sub>2</sub> O sterile | -20°C |
| Taq Polymerase (5U/μl)   | -20°C |
| Dde I enzyme (10 U//μl)  | -20°C |
| Digestion buffer 10X     | -20°C |
| Positive normal control  | -20°C |

**Stability:** over 12 months if correctly stored.

### 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<br>Absence of mutation<br>Normal patient | 2<br>Presence of mutation<br>Heterozygote subject | 3<br>Presence of mutation<br>Homozygote subject |
|--|---|---|
| 1 fragment                                 | 3 fragments                                       | 2 fragments                                     |
| 192 bp                                     | 192 bp<br>161 bp<br>31 bp                         | 161 bp<br>31 bp                                 |

Usually, the resolution of agarose gel weakens the visualization of the band of 31 bp. The molecular diagnosis is guaranteed from the visualization of other fragments.

### References:

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