ampli set F XIII Val34Leu CE IVD 45 tests cat 1317

detection of Val34Leu polymorphism of the Factor XIII

The development of thrombotis 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 an tetramer made by two subunits A and two subunits B ($A_2 B_2$) . 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 □and□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

Greenberg CS, Birckbichler PJ, Rice RH. FASEB J. 1991; 5: 3071-3077.
Muszbek L, Yee V, Hevessy Z. Thromb Res. 1999; 94: 271-305.
Board PG, Losowsky MS, Miloszewski KJ. Blood Rev. 1993; 7:229-242.
Kangsadalampai S, Board PG. Blood, 1998, 92; 2766-2770.
Endler G, Funk M, Haering D et al. British Journal of Haematology, 2003, 120; 310-314.
Balogh I, Szoke G, Karpati L et al. Blood, 2000, 96; 2479-2486.

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