

# FXIII G103T POLYMORPHISM

## ORDERING INFORMATION

REF: GEN-012-25 RDM Code: 1737859/R  
Tests: 25 Reactions: 31  
REF: GEN-012-50 RDM Code: 2164384/R  
Tests: 50 Reactions: 62  
CND Code: W0106010499  
Manufacturer: BioMol Laboratories s.r.l.

## CONTENTS OF THE KIT

The kit consists of reagents for Real-Time PCR amplification  
\*reagents for the extraction of genomic DNA are not supplied in the kit

For in vitro diagnostic use



## PRODUCT CHARACTERISTICS

Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS**.  
Detection of the G103T polymorphism of the gene coding for FXIII factor, by Real-Time PCR technique. Optimized kit for Real-Time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx.

## SCIENTIFIC BACKGROUND

Several genetic alterations, in particular those affecting physiological anticoagulants (antithrombin III, proteins C and S) and the procoagulant systems (factor V Leiden, prothrombin, fibrinogen), have been identified as risk factors for venous thromboembolism. Coagulation factor XIII (FXIII) is a transglutaminase that plays an important role in the final stage of blood coagulation, where it catalyzes the formation of covalent bonds between fibrin monomers to produce clot stabilization and resistance to fibrinolysis. Hereditary FXIII deficiency causes severe bleeding and a high risk of miscarriage in women with the homozygous mutation. Although several polymorphisms have been identified in the gene encoding the FXIII A subunit (Val34Leu, Pro564Leu, Val650Ile and Glu651Gln), the Val34Leu polymorphism is the most important functional polymorphism capable of influencing FXIII activation. This polymorphism is a G>T substitution at position 103 in exon 2, three amino acids away from the thrombin cleavage site that occurs in Arg37-Gly38. The release of the activating peptide is accelerated in this genetic condition. The less frequent allele (Leu34) has been described as a protective factor against myocardial infarction and venous thrombosis.

## CLINICAL SIGNIFICANCE

Venous thromboembolism (VTE), usually involving deep vein thrombosis, pulmonary embolism, or both, is a complex, multifactorial disorder in which a number of conditions interact and contribute to increased individual risk culminating in the development of venous occlusions. Thrombophilia is commonly defined as a propensity to develop venous thromboembolism based on a hypercoagulable condition attributable to inherited or acquired disorders involving blood clotting or fibrinolysis.

- § J Clin Med. 2024 Nov 15;13(22):6871. doi: 10.3390/jcm13226871. Recurrent Early Pregnancy Loss and Congenital Thrombophilia: A Prospective Study
- § Thrombophilic gene polymorphisms and recurrent pregnancy loss: a systematic review and meta-analysis. J Assist Reprod Genet. 2023 Jul;40(7):1533-1558. doi: 10.1007/s10815-023-02823-2.
- § J Clin Med. 2022 Jun 15;11(12):3454. doi: 10.3390/jcm11123454. Coagulation Factor XIII Val34Leu Polymorphism in the Prediction of Premature Cardiovascular Events-The Results of Two Meta-Analyses
- § Int J Mol Sci. 2021 Feb 12;22(3):1459. doi: 10.3390/ijms22031459. Factor XIII-A in Diseases: Role Beyond Blood Coagulation
- § Effect of factor XIII levels and polymorphisms on the risk of myocardial infarction in young patient. Balogh L, Mol Cell Biochem. 2018 Feb 26.
- § Association of the F13A1 Val34Leu polymorphism and recurrent pregnancy loss: A meta-analysis. Eur J Obstet Gynecol Reprod Biol. 2017 Aug; 215:234-240. doi: 10.1016/j.ejogrb.2017.06.032. Epub 2017 Jun 23.
- § Association of the F13A1 Val34Leu polymorphism and recurrent pregnancy loss: A meta-analysis. Eur J Obstet Gynecol Reprod Biol. 2017 Aug; 215:234-240.
- § Genetic association between FXIII and  $\beta$ -fibrinogen genes and women with recurrent spontaneous abortion: a meta-analysis. J Assist Reprod Genet. 2015 May;32(5):817-25. doi: 10.1007/s10815-015-0471-9. Epub 2015 Apr 11.
- § Blood coagulation factor XIII-A subunit Val34Leu polymorphisms and intracerebral hemorrhage risk: A meta-analysis of case-control studies. Br J Neurosurg. 2015;29(5):672-7.
- § Risk for early pregnancy loss by factor XIII Val34Leu: the impact of fibrinogen concentration. J Clin Lab Anal. 2013 Nov;27(6):444-9. doi: 10.1002/jcla.21626.



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| DESCRIPTION                          | LABEL                      | VOLUME       |              | STORAGE |
|--------------------------------------|----------------------------|--------------|--------------|---------|
|                                      |                            | GEN-012-25   | GEN-012-50   |         |
| Mix oligonucleotides and probes      | Mix Val34Leu FXIII 10X     | 1 x 77,5 µl  | 2 x 77,5 µl  | -20°C   |
| Mix buffer and Taq-polymerase enzyme | Mix Real-Time PCR 2X       | 1 x 387,5 µl | 2 x 387,5 µl | -20°C   |
| Deionized H <sub>2</sub> O           | Deionized H <sub>2</sub> O | 1 x 1 ml     | 1 x 1 ml     | -20°C   |
| Genomic DNA or recombinant DNA       | Control 1                  | 1 x 22 µl    | 2 x 22 µl    | -20°C   |
| Genomic DNA or recombinant DNA       | Control 2                  | 1 x 22 µl    | 2 x 22 µl    | -20°C   |
| Genomic DNA or recombinant DNA       | Control 3                  | 1 x 22 µl    | 2 x 22 µl    | -20°C   |

## TECHNICAL CHARACTERISTICS

COD. GEN-012-25 / COD. GEN-012-50

|                                                 |                                                                                                                                    |
|-------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------|
| STABILITY                                       | 18 months                                                                                                                          |
| REAGENTS STATUS                                 | Ready to use                                                                                                                       |
| BIOLOGICAL MATRIX                               | Genomic DNA extracted from whole blood, tissue, cells                                                                              |
| POSITIVE CONTROLS                               | Recombinant DNA for at least 3 analytical sessions (GEN-012-25)<br>Recombinant DNA for at least 6 analytical sessions (GEN-012-50) |
| VALIDATED INSTRUMENTS                           | Biorad CFX96 Dx, Biorad Opus Dx and Agilent AriaDx                                                                                 |
| TECHNOLOGY                                      | Real-time PCR; oligonucleotides and specific probes; 2 FAM/HEX fluorescence channels                                               |
| RUNNING TIME                                    | 85 min                                                                                                                             |
| THERMAL CYCLING PROFILE                         | 1 cycle at 95 °C (10 min); 45 cycles at 95 °C (15 sec) + 60 °C (60 sec)                                                            |
| ANALYTICAL SPECIFICITY                          | Absence of non-specific pairings of oligonucleotides and probes;<br>absence of cross-reactivity                                    |
| LIMIT OF DETECTION (LOD)                        | ≥ 0,016 ng of genomic DNA                                                                                                          |
| LIMIT OF BLANK (LOB)                            | 0% NCN                                                                                                                             |
| REPRODUCIBILITY                                 | 99,9%                                                                                                                              |
| DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY | 100%/98%                                                                                                                           |