

FV Y1702C (A5279G) POLYMORPHISM

ORDERING INFORMATIONS

REF: GEN-037-25 RDM Code: 2248815/R
Tests: 25 Reactions: 31
REF: GEN-037-50 RDM Code: 2248816/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 Y1702C (A5279G) polymorphism of the FV gene by Real-Time PCR technique. Kit optimized for Real-Time PCR instrumentation Biorad CFX96, Biorad Opus Dx and Agilent AriaDx.

SCIENTIFIC BACKGROUND

Venous thromboembolism (VTE), usually involving deep vein thrombosis, pulmonary embolism, or both, is a complex, multifactorial disorder in which several 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. Among acquired risk factors, some may lead to increased hypercoagulability, for example, cancer, surgery, injury or fracture, immobilization, pregnancy and the postpartum period, long-distance travel, hospitalization, catheterization and acute infection and others may be considered as predisposing conditions, such as age, gender, race/ethnicity, body mass index and obesity, use of oral contraceptive or hormone therapy, corticosteroids or statins, diet, physical activity, sedentary weather and air pollution.

CLINICAL SIGNIFICANCE

Furthermore, venous thromboembolism has a strong genetic basis, with approximately 50-60% of the variance in incidence attributable to genetic effects. Some genetic susceptibility variants that contribute to risk have been identified in candidate genes, such as factor V Leiden and prothrombin. A missense mutation in factor V resulting from a nucleotide residue substitution A → G at position 5279 in exon 15 has recently been identified, causing the Tyr → Cys substitution of amino acid 1702 (Y1702C) in the A3 domain of FV. This mutation appears to cause a deficiency in this factor, increasing the resistance of factor V to the anticoagulant action of activated protein C.

§ Genotyping analysis of the factor V Nara mutation, Hong Kong mutation, and 16 single-nucleotide polymorphisms, including the R2 haplotype, and the involvement of factor V activity in patients with recurrent miscarriage. Blood Coagul Fibrinolysis. 2017 Jun; 28(4):323-328.

§ Genetic modulation of the FV (Leiden)/normal FV ratio and risk of venous thrombosis in factor V Leiden heterozygotes. J Thromb Haemost. 2012 Jan; 10(1):73-80.



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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-037-25	GEN-037-50	
Mix oligonucleotides and probes	Mix FV Y1702C (A5279G) 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 ₂ O	Deionized H ₂ 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 2	1 x 22 µl	2 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-037-25 / COD. GEN-037-50

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissue, cells
CONTROLS	Recombinant DNA for at least 3 analytical sessions (GEN-037-25) Recombinant DNA for at least 6 analytical sessions (GEN-037-50)
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; 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%