



FV H1299R POLYMORPHISM

ORDERING INFORMATION

REF: GEN-015-25 Code RDM: 1744019/R
Tests: 25 Reactions: 31 x 2
REF: GEN-015-50 Code RDM: 2256370/R
Tests: 50 Reactions: 62 x 2
Code CND: W0106010499
Manufacturer: BioMol Laboratories s.r.l.

For in vitro diagnostic use



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

PRODUCT CHARACTERISTICS

Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS**. Detection of A4070G polymorphism of the FV (H1299R) gene by Real-Time PCR technique. Optimized kit for Real-Time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Hyris bCUBE and Hyris bCUBE3 with Hyris bAPP.

SCIENTIFIC BACKGROUND

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.

Factor V 1691 G→A (FV Leiden, FVL) is the most common genetic risk factor for hereditary thrombophilia resulting from a G→A nucleotide residue substitution at position 1691, causing the Arg→Glu substitution of amino acid 506 (R506Q). In addition to the FVL mutation, a substitution of a nucleotide residue A to G at residue 4070 of exon 13 of the factor V gene results in the H-R 1299 substitution of the protein and has been described as an R2 polymorphism. The R2 variation has been shown to affect plasma FV concentration and its association causes mild resistance to activated protein C. The polymorphism has been associated with an increased risk of thrombosis alone or in association in heterozygosity with the FV G1691A mutation.

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 an individual's 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 coagulation or fibrinolysis.

Among the acquired risk factors, some can cause an increase in hypercoagulability, for example cancer, surgery, trauma or fractures, 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, sex, race/ethnicity, body mass index and obesity, use of oral contraceptive or hormone therapy, corticosteroids or statins, diet, physical activity, sedentary time and air pollution.

§ A Systematic Review and Meta-Analysis of the Association between the FV H1299R Variant and the Risk of Recurrent Pregnancy Loss. *Biology (Basel)* 2022 Nov 3;11(11):1608. doi: 10.3390/biology11111608

§ 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.

§ Impaired APC cofactor activity of factor V plays a major role in the APC resistance associated with the factor V Leiden (R506Q) and R2 (H1299R) mutations. *Blood*. 2004 Jun 1;103(11):4173-9.

§ Multicenter Study. *Blood*. 1999 Nov 1;94(9):3062-6. Coinheritance of the HR2 haplotype in the factor V gene confers an increased risk of venous thromboembolism to carriers of factor V R506Q (factor V Leiden)



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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-015-25	GEN-015-50	
Oligonucleotides Mix	Mix A FV H1299R 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Oligonucleotides Mix	Mix B FV H1299R 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 775 µl	2 x 775 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 35 µl	2 x 35 µl	-20°C
Genomic DNA or recombinant DNA	Control 2	1 x 35 µl	2 x 35 µl	-20°C
Genomic DNA or recombinant DNA	Control 3	1 x 35 µl	2 x 35 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-015-25 / COD. GEN-015-50

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-015-25) Recombinant DNA for at least 6 analytical sessions (GEN-015-50)
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Hyris bCUBE and Hyris bCUBE3 with Hyris bAPP.
TECHNOLOGY	Real-time PCR; specific oligonucleotides; 1 SYBR-GREEN/FAM fluorescence channel
RUNNING TIME	150 min
THERMAL CYCLING PROFILE	1 cycle at 50 °C (2 min); 1 cycle at 94 °C (5 min); 30 cycles at 95 °C (50 sec) + 60 °C (40 sec) + 72 °C (50 sec) + 1 dissociation cycle from 70°C to 90°C with 0,2 °C increments.
ANALYTICAL SPECIFICITY	Absence of non-specific pairings of oligonucleotides; 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%