

FV LEIDEN G1691A POLYMORPHISM

For in vitro diagnostic use



ORDERING INFORMATIONS

REF: GEN-001-25 RDM Code: 1718429/R
Tests: 25 Reactions: 31
REF: GEN-001-50 RDM Code: 2255477/R
Tests: 50 Reactions: 62
CND Code: W0106010103
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

PRODUCT CHARACTERISTICS

Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS**. Detection of the G1691A FV Leiden polymorphism 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 (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 occlusives. 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 the environmental risk factors, some can lead to increased hypercoagulability, for example cancer, surgery, trauma or fracture, immobilisation, 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.

§ Int J Mol Sci. 2024 May 11;25(10):5228. doi: 10.3390/ijms25105228. The Etiology of the Thrombotic Phenomena Involved in the Process of Coronary Artery Disease-What Is the Role of Thrombophilic Genes in the Development of This Pathology?

§ J Hum Reprod Sci. 2023 Oct-Dec;6(4):352-357. doi: 10.4103/jhrs.jhrs_137_23. Epub 2023 Dec 29. Combined Parental Thrombophilia Gene Mutation Defects in Couples with Repeated Pregnancy Loss

§ Ann Hematol. 2024 Aug 21. doi: 10.1007/s00277-024-05926-2. Online ahead of print. Venous thromboembolism risk in adults with hereditary thrombophilia: a systematic review and meta-analysis

§ Thrombophilia Screening: Not So Straightforward. Moore GW. Semin Thromb Hemost. 2024 May 11. doi: 10.1055/s-0044-1786807.

§ Turk J Med Sci. 2024 Jun 12;54(4):682-687. doi: 10.55730/1300-0144.5837. eCollection 2024. Can prothrombotic gene variants and Apol rs5069 polymorphism be the predictors of early myocardial infarctions?

§ Laboratory biomarkers for venous thromboembolism risk in patients with hematologic malignancies: A review. Thromb Res. 2018 Mar; 163:138-145. doi: 10.1016/j.thromres.2018.01.037. Epub 2018 Jan 31.

§ Pregnancy, thrombophilia, and the risk of a first venous thrombosis: systematic review and bayesian meta-analysis. Croles FN, Nasserinejad K, Duvekot JJ, Kruij MJ, Meijer K, Leebeek FW. BMJ 2017; 359 doi: https://doi.org/10.1136/bmj.j4452

§ J Res Med Sci. 2015 Jun; 20 (6):554-62. Factor V Leiden, factor V Cambridge, factor II G20210, and methylenetetrahydrofolate reductase in cerebral venous and sinus thrombosis: A case-control study.

CLINICAL SIGNIFICANCE

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.

The identification of the factor V Leiden (G1691A) missense mutation (Arg506Gln) causing factor V resistance to the anticoagulant action of activated protein C represents a landmark in understanding the basis of hereditary thrombotic risk. The FVL mutation is, in fact, the most common hereditary defect that predisposes to venous thrombosis



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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-001-25	GEN-001-50	
Mix oligonucleotides and probes	Mix G1691A FV Leiden 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 3	1 x 22 µl	2 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-001-25 / COD. GEN-001-50

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-001-25), Recombinant DNA for at least 6 analytical sessions (GEN-001-50)
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx and Agilent AriaDx, Hyris bCUBE, Hyris bCUBE3 with Hyris bAPP
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%