

G1001C polymorphism of the FV gene (FV Cambridge)

ORDERING INFORMATION

REF: GEN-062-25
Tests: 25 Reactions: 31
REF: GEN-062-50
Tests: 50 Reactions: 62
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-RESEARCH USE ONLY**. Detection of the FV Cambridge G1001C polymorphism (rs118203906) by Real-Time PCR technique. Kit optimized for Real-time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx.

SCIENTIFIC BACKGROUND

Venous thromboembolism (VTE), which usually involves deep vein thrombosis, pulmonary embolism, or both conditions, is a complex, multifactorial disorder in which a number of conditions interact and contribute to an increased individual risk, culminating in the development of venous occlusive disorders. Thrombophilia is commonly defined as a predisposition to developing venous thromboembolism based on a state of hypercoagulability attributable to inherited or acquired disorders involving blood coagulation or fibrinolysis.

Among environmental risk factors, some may lead to increased hypercoagulability, such as cancer, surgery, trauma or fractures, immobilisation, pregnancy and the postpartum period, long-distance travel, hospitalisation, catheterisation and acute infection; others may be considered predisposing conditions, such as age, sex, race/ethnicity, body mass index and obesity, the use of oral contraceptives or hormone therapy, corticosteroids or statins, diet, physical activity, sedentary time and air pollution.

CLINICAL SIGNIFICANCE

Furthermore, venous thromboembolism has a strong genetic basis, with approximately 50–60% of the variation in incidence attributable to genetic factors. Certain genetic susceptibility variants that contribute to the risk have been identified in candidate genes, such as those encoding Factor V and prothrombin.

Cambridge Factor V is a genetic variant of coagulation factor V, characterised by the substitution of the amino acid arginine with threonine at position 306 (Arg306→Thr, AGG→ACG) of the protein.

This mutation alters the cleavage site of activated protein C (APC), reducing factor V's ability to be inhibited and thus promoting excessive pro-coagulant activity, resulting in resistance to APC.

The mutation is associated with an increased risk of venous thrombosis, although it is extremely rare in the general population.

§ Williamson D, Brown K, Luddington R, Baglin C, Baglin T. Factor V Cambridge: a new mutation (Arg306→Thr) associated with resistance to activated protein C. *Blood*. 1998 Feb 15;91(4):1140-4. PMID: 9454742.

§ Norström E, Thorelli E, Dahlbäck B. Functional characterization of recombinant FV Hong Kong and FV Cambridge. *Blood*. 2002 Jul 15;100(2):524-30. doi: 10.1182/blood-2002-02-0343. PMID: 12091344.

§ Saadatnia M, Salehi M, Movahedian A, Shariat SZ, Salari M, Tajmiriahi M, Asadimobarakkeh E, Salehi R, Amini G, Ebrahimi H, Kheradmand E. Factor V Leiden, factor V Cambridge, factor II GA20210, and methylenetetrahydrofolate reductase in cerebral venous and sinus thrombosis: A case-control study. *J Res Med Sci*. 2015 Jun;20(6):554-62. doi: 10.4103/1735-1995.165956. PMID: 26600830; PMCID: PMC4621649.

§ van der Neut Kolfschoten M, Dirven RJ, Vos HL, Tans G, Rosing J, Bertina RM. Factor Va is inactivated by activated protein C in the absence of cleavage sites at Arg-306, Arg-506, and Arg-679. *J Biol Chem*. 2004 Feb 20;279(8):6567-75. doi: 10.1074/jbc.M308574200. Epub 2003 Dec 2. PMID: 14660667.

§ Bos MH, Meijerman DW, van der Zwaan C, Mertens K. Does activated protein C-resistant factor V contribute to thrombin generation in hemophilic plasma? *J Thromb Haemost*. 2005 Mar;3(3):522-30. doi: 10.1111/j.1538-7836.2005.01181.x. PMID: 15748243.



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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-062-25	GEN-062-50	
Mix oligonucleotides and probes	Mix FV Cambridge 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 5X	1 x 155 µl	2 x 155 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-062-25 / GEN-062-50

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissues, cells
POSITIVE CONTROLS	Recombinant DNA for at least 3 analytical sessions (GEN-062-25) Recombinant DNA for at least 6 analytical sessions (GEN-062-50)
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 oligonucleotides and probes; absence of cross-reactivity
LIMIT OF DETECTION (LOD)	≥ 5 ng of genomic DNA
LIMIT OF BLANK (LOB)	>40 Cq
REPRODUCIBILITY	99,9%
SPECIFICITY / SENSITIVITY	99,9%/98%