ORDERING INFORMATION:

THROMBOPHILIA

PRODUCT	Code	N° Tests	Application
REALQUALITY **RQ-ACE (I/D)**	RQ-75-4M	50	Manual
	RQ-75-6M	100	
	RQ-75-4A	50	GQ X120 e GQ Max
	RQ-75-6A	100	
REALQUALITY **RQ-FACTOR V Y1702C**	RQ-69-4M	50	Manual
	RQ-69-6M	100	
	RQ-69-4A	50	GQ X120 e GQ Max
	RQ-69-6A	100	
REALQUALITY **RQ-PAI-1 4G/5G**	RQ-119-4M	50	Manual
	RQ-119-6M	100	
	RQ-119-4A	50	GQ X120 e GQ Max
	RQ-119-6A	100	
REALQUALITY **RS-FACTOR V H1299R**	RQ-111-4M	50	Manual
	RQ-111-6M	100	
	RQ-111-4A	50	GQ X120 e GQ Max
	RQ-111-6A	100	
REALQUALITY **RS-FACTOR II G20210A**	RQ-27-4M	50	Manual
	RQ-27-6M	100	
	RQ-27-4A	50	GQ X120 e GQ Max
	RQ-27-6A	100	
REALQUALITY **RS-FACTOR V LEIDEN**	RQ-25-4M	50	Manual
	RQ-25-6M	100	
	RQ-25-4A	50	GQ X120 e GQ Max
	RQ-25-6A	100	
REALQUALITY **RS-MTHFR A1298C**	RQ-31-4M	50	Manual
	RQ-31-6M	100	
	RQ-31-4A	50	GQ X120 e GQ Max
	RQ-31-6A	100	
REALQUALITY **RS-MTHFR C677T**	RQ-29-4M	50	Manual
	RQ-29-6M	100	
	RQ-29-4A	50	GQ X120 e GQ Max
	RQ-29-6A	100	

THROMBOPHILIA DUPLEX NEW

PRODUCT	Code	N° Tests	Application
REALQUALITY **THROMBO FII-FVL**	RQ-177-4M	50	Manual
	RQ-177-6M	100	
	RQ-177-4A	50	GQ X120 e GQ Max
	RQ-177-6A	100	

A duplex screening kit for the detection and genotyping of Factor II G20210A and Factor V Leiden mutations.

REALQUALITY **THROMBO MTHFR**	RQ-178-4M	50	Manual
	RQ-178-6M	100	
	RQ-178-4A	50	GQ X120 e GQ Max
	RQ-178-6A	100	

A duplex screening kit for the detection and genotyping of MTHFR C677T and MTHFR A1298C mutations.

HEMOCHROMATOSIS

PRODUCT	Code	N° Tests	Application
REALQUALITY **RQ-HEMO S65C**	RQ-43-4M	50	Manual
	RQ-43-6M	100	
	RQ-43-4A	50	GQ X120 e GQ Max
	RQ-43-6A	100	
REALQUALITY **RS-HEMO C282Y**	RQ-39-4M	50	Manual
	RQ-39-6M	100	
	RQ-39-4A	50	GQ X120 e GQ Max
	RQ-39-6A	100	
REALQUALITY **RS-HEMO H63D**	RQ-41-4M	50	Manual
	RQ-41-6M	100	
	RQ-41-4A	50	GQ X120 e GQ Max
	RQ-41-6A	100	

EQA Programmes (VEQ)

PRODUCT	Pkg	Code
Molecular Genetics of Thrombophilia (Factor V Leiden) *	3 challenges	UN-FVLEQA
SCHEME 5A - HFE Typing *	3 challenges	UN-SCHEME5A

** distribution exclusively for the Italian market*

CE IVD

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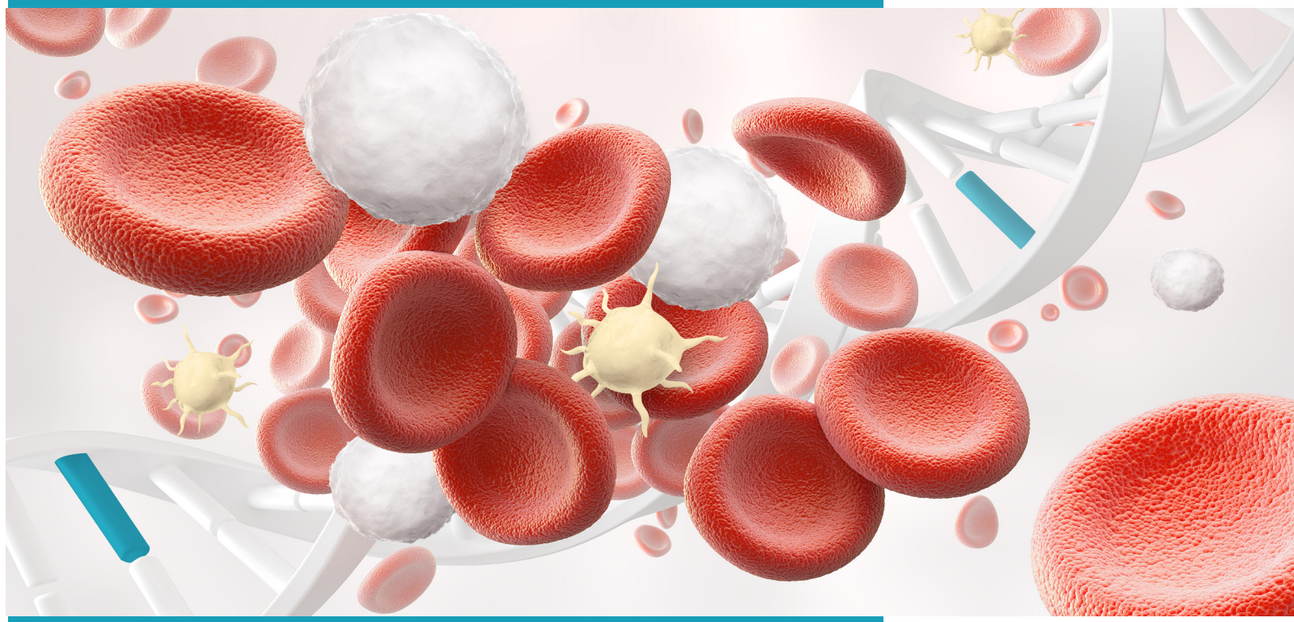
THROMBOPHILIA_HEMOCHROMATOSIS_Brochure_e20240322

REALQUALITY THROMBOPHILIA & HEMOCHROMATOSIS

Kits for the discrimination of the most important allelic mutations related to thrombophilia and hemochromatosis by Real-Time PCR



Thrombophilia

ACE I-D

FACTOR V Y1702C

PAI-1 4G-5G

FACTOR V H1299R

FACTOR-II G20210A

FACTOR V Leiden

MTHFR A1298C

MTHFR C677T

Hemochromatosis

HEMO H63D

HEMO S65C

HEMO C282Y

Thrombophilia Duplex

FII-FVL

MTHFR A1298C-C677T

AB ANALITICA ADVANCED BIOMEDICINE

THROMBOPHILIA

Thrombophilia is commonly defined as any acquired or hereditary disorder associated with an increased risk to develop thromboembolic phenomena. These phenomena occur when blood circulation is blocked by clots, originating in veins or derived from a thrombus in another area of the body. Most commonly, thrombi develop in superficial or deep veins of the legs, but can also be found in veins of the brain, the retina, the liver or in mesenteric veins.
Thrombosis, that is not the result of a genetic defect, often occurs in elderly persons as a consequence of strong environmental risks factors, including surgery, bone fracture or cancer. In contrast, hereditary thrombosis is associated with an onset at earlier age, due to the presence of one or more genetic defects caused by gene-to-gene and/or gene-to-environment interactions.
Genetic alterations of different blood components may directly or indirectly influence blood homeostasis, thus triggering a prothrombotic state. Such alterations may lead to the loss of function of natural anticoagulants (e.g. Protein C, Protein S, Antithrombin), to an increased activity of procoagulant factors (e.g. Prothrombin, Factor V, Factor VII, Factor IX, Factor XIII, MTHFR, MTRR) or to diminished fibrinolytic activity (e.g. PAI-1, TAFI).
REALQUALITY THROMBOPHILIA KITS allow the detection of different mutation responsible to the development of venous thrombosis.

PRODUCT CHARACTERISTICS:

- Includes dUTP/UNG system for contamination prevention and a fluorescence normalizer.
- Validated on the most common Real-Time PCR thermocyclers:
  - Applied Biosystems™ 7500 Fast (Applied Biosystems).
  - Applied Biosystems™ 7300 Real-Time PCR System (Applied Biosystems).
  - Applied Biosystems™ StepOne (Applied Biosystems).
  - CFX96™ Dx Real-Time System (Bio-Rad).
  - CFX96™ Dx Real-Time PCR Detection Systems for In Vitro Diagnostics (IVD) (Bio-Rad)*.
  - CFX96™ Real-Time PCR Detection System (Bio-Rad).
  - CFX96™ Real-Time PCR Detection System-IVD (Bio-Rad)*.
  - LightCycler® 480 Real-Time PCR System version II (Roche).
  - Rotor-Gene® Q MDx (QIAGEN)*.
  - Mic qPCR Cycler (Mic – Bio Molecular Systems)*.
  - AriaDx Real-Time PCR System (Agilent Technologies)*.

  *thermocyclers validated also for THROMBOPHILIA DUPLEX KIT
- Available also in automatic format for **GENEQUALITY® X120** and **GENEQUALITY® Max** platform.
- Easy interpretation of results with **AB-SNP-REPORT SOFTWARE**.

SPECIMENS:

Validated on DNA extracted from whole peripheral blood.

SHELF LIFE:

18 months.

FLUOROPHORES:

Fluorophores	Target
FAM	WT allele
HEX	MUT allele

HEMOCHROMATOSIS

Hemochromatosis is an autosomal recessive disorder of the iron metabolism that affects approximately 0.2 - 0.5 % of the Caucasian population. It is characterized by an excessive accumulation of iron in the body, which is caused by an increased absorption of dietary iron by the intestinal mucosa. The excess iron is first deposited in the liver tissue, causing the organ to swell and over time leading to irreversible damage, like liver cirrhosis. Other organs where the excess iron is stored are the heart, the pancreas (significantly increased risk of diabetes), the endocrine organs (particularly the pituitary gland and testicles), and the joints. If genetically determined hemochromatosis is diagnosed early and is treated appropriately, predisposed individuals may never develop any symptoms. For this reason, early diagnosis of a genetic predisposition is extremely important.
It has been shown that mutations in the HFE gene cause the disorder. Certain mutations in this gene lead to the synthesis of an altered protein, which is unable to interact with transferrin receptors, which in turn forces the transport of iron through the intestinal mucosa. The two most frequent mutations found in the HFE gene are the C282Y mutation (substitution of a cysteine by a tyrosine) and the H63D mutation (substitution of the histidine by aspartic acid). A third type of mutation, S65C (substitution of a serine with a cysteine), is found in significantly fewer patients.
REALQUALITY HEMOCHROMATOSIS KITS allow the detection of different mutation related to hemochromatosis.

PRODUCT CHARACTERISTICS:

- Includes dUTP/UNG system for contamination prevention and a fluorescence normalizer.
- Validated on the most common Real-Time PCR thermocyclers:
  - Applied Biosystems™ 7500 Fast (Applied Biosystems).
  - Applied Biosystems™ 7300 Real-Time PCR System (Applied Biosystems).
  - Applied Biosystems™ StepOne (Applied Biosystems).
  - CFX96™ Dx Real-Time System (Bio-Rad).
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Fluorophores	Target
FAM	WT allele
HEX	MUT allele

Automation and Data analysis