

# We Make Complexity Simpler

Coagulation Disorders in a wide range of pathologies: multifactorial genetic analysis by Duplica<sup>Real Time</sup> Genotyping kit

The role of coagulation factor polymorphisms and homocysteine level in cardiovascular diseases is well known. Factor V (FV) R506Q (Leiden mutation) and prothrombin (PT) 20210G/A mutation, relatively frequent functional polymorphisms in population, have been found to play a major role as risk factors in cardiovascular diseases.

Both factors, enhancing thrombin generation by prothrombinase complex, throughout different mechanisms. The heterogeneity of clinical phenotypes and different onsets of thrombosis observed in thrombophilic people, have led to the hypothesis that predisposition to venous thrombosis results from combination of several genetic defects.

Several studies also demonstrated that elevation of homocysteine (hyper-homocysteinemia) is a risk factor for cardiovascular diseases, strokes and venous thromboembolisms.

Mild hyper-homocysteinemia is quite common and can be caused by nutritional deficiency, certain treatments and by common gene polymorphisms like MTHFR mutations.

Multiparametric analysis of the most important polymorphisms is EuroClone Diagnostica concept for diagnosis of thrombotic events due to genetic disorders.

#### References:

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2. Lentz SR. Mechanisms of homocysteine-induced atherothrombosis. *J Thromb Haemost* 2005; 3: 1646-54.
3. Homocysteine Studies Collaboration. Homocysteine and risk of ischemic heart disease and stroke: a meta-analysis. *JAMA* 2002;288:2015-22.
4. Wald DS, Law M, Morris JK. Homocysteine and cardiovascular disease: evidence on causality from a meta-analysis. *BMJ* 2002; 325:1202.
5. den Heijer M, Lewington S, Clarke R. Homocysteine, MTHFR and risk of venous thrombosis: a meta-analysis of published epidemiological studies. *J Thromb Haemost* 2005; 3: 292-9.

# Coagulation Disorders in a wide range of pathologies: multifactorial genetic analysis by Duplica<sup>®</sup> Real Time Genotyping kit

Ref.	Description	Platform	Format
<b>Single Thermal Profile</b>			
EER001032	Duplica <sup>®</sup> Real Time Factor II G20210A Genotyping Kit	SC	32 tests
EER002032	Duplica <sup>®</sup> Real Time Factor V G1691A Genotyping Kit	SC	32 tests
EER010032	Duplica <sup>®</sup> Real Time Factor V H1299R Genotyping Kit *available on request	SC, RQ	32 tests
EER041032	Duplica <sup>®</sup> Real Time Next Factor V H1299R Kit	ABI, Bio-Rad	32 tests
EER013032	Duplica <sup>®</sup> Real Time Factor V Y1702C Genotyping Kit *available on request	SC, RQ	32 tests
EER024032	Duplica <sup>®</sup> Real Time Factor V R306T (Cambridge) Genotyping Kit *available on request	SC, RQ	32 tests
EER022032	Duplica <sup>®</sup> Real Time Factor XIII V34L Genotyping Kit *available on request	SC, RQ	32 tests
EER017032	Duplica <sup>®</sup> Real Time PAI-I Genotyping Kit	SC, RQ	32 tests
EER003032	Duplica <sup>®</sup> Real Time MTHFR C677T Genotyping Kit	SC	32 tests
EER007032	Duplica <sup>®</sup> Real Time MTHFR A1298C Genotyping Kit	SC	32 tests
EER008032	Duplica <sup>®</sup> Real Time MTRR A66G Genotyping Kit *available on request	SC	32 tests
EER009032	Duplica <sup>®</sup> Real Time MTR A2756G Genotyping Kit *available on request	SC	32 tests
<b>Common Thermal Profile 1</b>			
EER037032	Duplica <sup>®</sup> Real Time Mix & Match Factor II G20210A Kit	RQ, ABI, Bio-Rad, LC	32 tests
EER038032	Duplica <sup>®</sup> Real Time Mix & Match Factor V G1691A Kit	RQ, ABI, Bio-Rad, LC	32 tests
EER039032	Duplica <sup>®</sup> Real Time Mix & Match MTHFR C677T Kit	RQ, ABI, Bio-Rad, LC	32 tests
EER040032	Duplica <sup>®</sup> Real Time Mix & Match MTHFR A1298C Kit	RQ, ABI, Bio-Rad, LC	32 tests
<b>Common Thermal Profile 2</b>			
EER042032	Duplica <sup>®</sup> Real Time Mix & Match MTRR A66G Kit	RQ, ABI, Bio-Rad, LC soon	32 tests
EER043032	Duplica <sup>®</sup> Real Time Mix & Match MTR A2756G Kit	RQ, ABI, Bio-Rad, LC soon	32 tests
<b>Suitable for QIASymphony AS</b>			
EER027032	Duplica <sup>®</sup> Real Time Factor V G1691A Genotyping Kit for Rotor-Gene Q	RQ	32 tests
EER031032	Duplica <sup>®</sup> Real Time Factor V H1299R Genotyping Kit for Rotor-Gene Q	RQ	32 tests
EER028032	Duplica <sup>®</sup> Real Time Factor II G20210A Genotyping Kit for Rotor-Gene Q	RQ	32 tests
EER029032	Duplica <sup>®</sup> Real Time MTHFR C677T for Rotor-Gene Q	RQ	32 tests
EER030032	Duplica <sup>®</sup> Real Time MTHFR A1298C for Rotor-Gene Q	RQ	32 tests

\* product licensed by Hoffman-La Roche / # available on request

## Intended use:

Duplica<sup>®</sup> Real Time detection Kit are assays based on allelic discrimination (Wild Type and Mutated) of different polymorphisms used as indicators of genetic predisposition in cardiovascular diseases.

**Platform legend:** **SC** SmartCycler (Cepheid), **RQ** Rotor-Gene Q (Qiagen), **ABI** Applied Biosystems platforms, **Bio-Rad** Bio-Rad platforms, **MX** Stratagene, **LC** LightCycler 480 (Roche).

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