

Thrombotic Mutations

Factor II, Factor V, Factor XIII, MTHFR A1298C,
MTHFR C677T, PAI-1 Genotyping

Mutations Genotyping

/ Coagulation factors

Faktor II – prothrombin

- hypothyrombinemia

Faktor V – proaccelerin, labile factor

- Leiden mutation
- up to 10 times higher thrombosis development risk

Faktor XIII – Laki-Lorand factor, fibrin stabilizing factor

- affects the quality of thrombus

NEW PRODUCT

/ Methylene tetrahydrofolate reductase (MTHFR)

MTHFR A1298C and **MTHFR C677T**

- accumulation of homocysteine in plasma
- increased risk of repeated miscarriages in I. trimester
- risk of venous thromboembolism

/ Acute-phase proteins

PAI-1 – plasminogen activator inhibitor-1

- decreased fibrinolysis
- atherothrombosis marker
- genotype 4G/4G – up to 38% risk of miscarriage

Key Facts

- / Disruption of balance between coagulation and fibrinolysis
 - hypothyrombinemia
 - venous thromboembolism
 - pulmonary embolism
 - cardiovascular diseases
 - risk of vascular diseases of placenta, fetal growth disorders, repeated miscarriages, premature births

Diagnostics

- / Examination of hemocoagulation and its inhibitors
 - / Monitoring of acute-phase proteins levels
- / Targeted genotyping of thrombophilic mutations
 - minimizes the risk of life-threatening conditions
 - minimizes risks for mother and fetus during pregnancy

/ The PCR kit is designed to detect polymorphism of the *F13A1* gene encoding the subunit A of FXIII factor by the real-time Polymerase Chain Reaction (PCR) method.

The method is based on amplification and detection of the target sequence using allele specific fluorophore labelled probes. Target sequence is a single nucleotide replacement of guanine by thymine in site 103 (G103T). Presence of wild-type allele (G103G) is detected in the FAM fluorescent channel and mutant allele (T103T) in the HEX fluorescent channel. In case of heterozygous genotype (G103T) a signal is detected in both channels. Detection kit contains Ready to Use MasterMix and utilizes "hot start" technology, minimizing non-specific reactions and assuring maximum sensitivity. Kit is designed for *in vitro* diagnostics.

Technology	real-time PCR
Target Sequence	<i>F13A1</i> gene
Specificity	V34L polymorphism of the <i>F13A1</i> gene (G103T)
Detection limit	2 ng/μl
Qualitative Detection	mutation present × not present
Evaluation	G/G – standard homozygote (wild type) T/T – mutant homozygote G/T – heterozygote
Validated Sample Material	whole blood
Kit Storage	-20 °C ± 5 °C
Validated Isolation Methods	croBEE NA16 Nucleic Acid Extraction System GeneProof PathogenFree DNA Isolation Kit
Validated Real-Time PCR Devices	croBEE Real-Time PCR System LightCycler® 2.0 LineGene 9600 Plus Rotor-Gene 3000/6000 SLAN® Real-Time PCR System
Required Detection Channels	FAM, HEX
Quality Control	regularly tested by INSTAND e.V. External Quality Assessment panels – results at www.geneproof.com
Certification	CE IVD for <i>in vitro</i> Diagnostics Use

Product name	Technology	Cat. No.	
		50 reactions	100 reactions
Thrombotic Mutations			
GeneProof Factor II Prothrombin PCR Kit	real-time PCR	FII/050	FII/100
GeneProof Factor V Leiden PCR Kit	real-time PCR	FV/050	FV/100
GeneProof Factor XIII V34L PCR Kit	real-time PCR	FXIII/050	FXIII /100
GeneProof MTHFR A1298C PCR Kit	real-time PCR	M1298/050	M1298/100
GeneProof MTHFR C677T PCR Kit	real-time PCR	M677/050	M677/100
GeneProof PAI-1 Genotyping PCR Kit	real-time PCR	PAI/050	PAI/100