GENOTYPING KIT – VENOUS THROMBOSIS

Venous thrombosis is a common disease and affects 1 in 1000 individuals annually. Today, several genetic defects have been identified and shown to be associated with an increased risk of developing thrombosis. The most frequent inherited risk factor for venous thrombosis is the **R/Q 506 mutation in the factor V gene (Factor V Leiden)**. This mutation is found in Caucasians with prevalences of the mutant allele ranging from 1-2% up to about 15% in certain regions. In the Austrian and German population the average prevalence has been found to be 5%. The mutation conveys resistance to cleavage of factor V by activated protein C at position R 506 and is associated with low APC ratios.
While the risk for thrombosis in heterozygous patients is increased about 3.5 to 7-fold, it is about 80-fold increased in homozygous individuals.

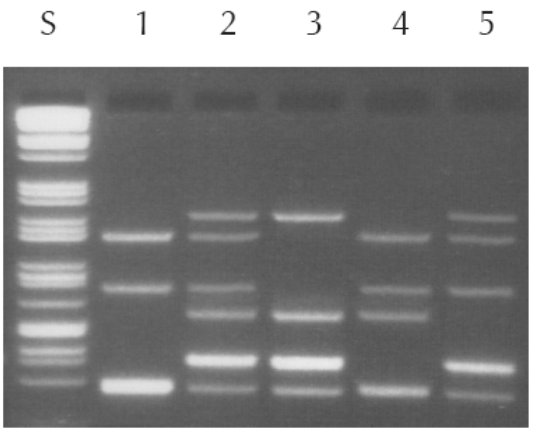
A recently found common mutation in the 3'-untranslated region of the prothrombin gene **(Factor II:202010 G/A)**, with a prevalence of 1-2% in the Austrian population, has been shown to be associated with elevated levels of factor II activity and antigen. It occurs more frequently in patients with thrombosis than in healthy control groups and represents an independent risk factor of thrombosis.

A homozygous mutation in the gene for the enzyme 5.10-methylene-tetrahydrofolate reductase **(MTHFR: A/V 223**) that results in the conversion of alanine to valine occurs with a prevalence of about 12% in the Austrian population. It has been linked to elevated levels of homocysteine and is considered to be another mild genetic risk factor for venous thrombosis.

Test Parameters

- R/Q 506 (G/A 1691) mutation in the factor V gene (Factor V Leiden)
- 20210 G/A mutation in the prothrombin gene (Factor II)
- A/V 223 (677 C/T) mutation in the 5.10-methylene-tetrahydrofolate reductase gene (MTHFR)

Examples of PCR Products



Separation of allele-specific PCR products
Lane S: size marker, Lanes 1 to 5: Samples

Interpretation

Sample No.	1	2	3	4	5
Factor V	wt	he	ho	wt	he
MTHFR	wt	he	ho	he	wt
Factor II	wt	he	he	wt	he

wt: wildtype
he: mutant, heterozygous
ho: mutant, homozygous

Test principle

Preparation of DNA

↓

DNA amplification (PCR)

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Analysis of PCR products by gel electrophoresis

Features

- quick and easy sample preparation from citrated blood
- testing for three mutations in one test tube („multiplex“ PCR)
- Mastermix ready to use
- simple analysis of PCR products by gel-electrophoresis on ready-to-use gels
- easy determination of genotype

Literature

1. Zenz W. et al. Factor V Leiden and prothrombin gene C 20210 A variant in children with ischemic stroke Thromb. Haemost (1998), 80 (5); 763-766
2. Rintelen C. et al. No evidence for an increased risk of venous thrombosis in patients with factor V Leiden by the homozygous 677 C toT mutation in the metbyleneterahydrofolate-reductase gene Blood Coagul.Fibrinol. (1999), 10; 61-69
3. Lalouschek W. et al. C677T MTHFR mutation and Factor V Leiden mutation in patients with TIA/minor stroke: A case control study Thromb. Res. (1999), 93; 61-69
4. Mustafa S. et al. Clinical features of thrombophilia in families with gene defects in protein C or protein 5 combined with factor V Leiden Blood Coagul. Fibrinol. (1998), 9; 85-89

Ordering information

REF TC11170 Genotyping Kit – Venous Thrombosis 22 T.

Technoclone GmbH
Brunner Str. 59/5
1230 Vienna, Austria
Tel: +43 1 86373-10
Fax: +431 86373-44

Technoclone Deutschland GmbH
Hans Bunte Str. 8-10
69123 Heidelberg, Germany
Tel: +49 6221 830 441
Fax: +49 6221 830 437

Technoclone Ltd.
Eclipse House 7, Curtis Road,
Dorking/Surrey RH4 1 EJ
Tel: +44 1306 888 777
Fax: +44 1306 883 883

sales@technoclone.com http://www.technoclone.com

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