Factor 5 Leiden mutation ^, Proaccelerin

General:

Every year, 0.1% of the population are affected by a thrombotic event. One of the causes is decreased activities of coagulation inhibitors such as AT3, protein C and protein S. However the most common cause is a mutation in the factor V gene. The mutation causes pathological APC resistance by replacement of arginine with glutamine in the factor V protein at position 506. The proteolytic splicing site for activated protein C is thereby transformed - and thus leads to an increased pro-coagulatory effect of factor V.

Indication: Suspicion of thrombosis predisposition (e.g. planned oral contraception), in familial cumulative thromboembolism, thromboembolism at young age, increased or borderline APC resistance (can be influenced by contraceptives).

Material: 2 ml EDTA blood

TAT: up to 7 days, FML

Method: PCR

Units: qualitative

Note: If a patient obtains anticoagulative treatment, the investigation of APC resistance could be impaired. We recommend testing for mutations in these cases.

Results and interpretation:

Normal genotype (Wildtype)	none of the factor V alleles carries the Leiden mutation.
Heterozygous genotype	one of the factor V alleles carries the Leiden mutation leading to a 7-fold increased risk of thrombosis, additional exogenous risk factors should be avoided.
Homozygous mutant genotype	both factor V alleles carry the Leiden mutation leading to an 80-100 fold risk of thrombosis! Other members of the family should be included in examinations.

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit http://www.fml-dubai.com/parameter-listings/

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