

# Factor 5 Leiden mutation <sup>^</sup>, 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).

**Stability:** 2 weeks at 2 to 8°C

**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/>