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Prothrombin, Factor 2 Mutation ^ (20210)

General:

S. R. Poort and colleagues (1996) described a so far unknown inherited mutation in the coagulating factor II: a base exchange of guanine to adenine at position 20210 of the factor II gene. Since 87% of positive carriers show an increased prothrombin level, it is assumed that the mutated gene is important for gene expressional regulation. Vice versa, increased prothrombin levels alone do not confirm the Factor II mutation. A confirmation is only possible on the molecular genetic level. Homozygous carriers are seriously at risk of developing a thrombotic event. The heterozygous form of the factor-II mutation (concerning only one allele) is the second most frequent cause of venous thrombosis after the factor-V Leiden mutation. The prevalence of the mutation within the normal population is approx. 2%. Eighteen % of the patients with familial thrombosis and 6.2% of the patients without familial history are carriers of the mutation. The thrombosis risk in (heterozygous) mutation carriers is approximately three fold increased compared to non-carriers.

OMIM:	176930
Gene:	Factor II, Locus 11q11
Indication:	Family history of thromboembolic events, thromboembolic event at young age.
Material:	2 ml EDTA blood
TAT:	upto 7 days, FML
Method:	PCR
Note:	Signed consent necessary!

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DAkkS Deutsche Akkreditierungsstelle D-ML-13455-01-00

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*performed in a collaborating laboratory (Germany)

Results and interpretation	
Normal genotype (wildtype)	none of the two alleles carries the 20210 mutation.
Heterozygous genotype	one of the alleles carries the 20210 mutation, the risk of thrombosis is approx. 3 times increased.
Homozygous mutant genotype	both alleles carry the 20210 mutation, the risk of thrombosis is increased considerably.

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

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