

# Homocysteinemia genetic typing

## General:

Homocysteine, an intermediate product of the methionine metabolism, increases the risk for thrombosis. The pathophysiological mechanism is not exactly known. A significantly increased plasma level of homocysteine is an established risk factor for arterial and venous thrombotic events. An increased homocysteine level is acquired, e.g. in vitamin B6-, B12- or folic acid deficiency. Carriers of a frequent polymorphism in the gene coding for methylenetetrahydrofolate reductase (MTHFR-gene, chromosome 1p36.3; mutation at nucleotide position 677) show a reduced enzyme activity by which less homocysteine is remethylated to methionine. It can result in mild hyperhomocysteinemia. Insignificantly increased homocysteine serum concentrations are also found in heterocytous carriers. Testing for mutation A1298C is also available.

Indication: clarification of homocysteinuria, familial thromboses, recurrent abortions, thrombembolism.

Material: 5 ml EDTA blood

TAT: 7-10 days\*

Method: PCR

Ref.- range: see report

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