HOMOCYSTEINE
(5,10-METYLENTETRAHYDRO-FOLATEREDUCTASE, MTHFR, C667T AND A1298C)

PATHOGENESIS

The amino acid homocysteine (HC) is considered (together with high cholesterol rate, LDL and lipoprotein-a, low values of HDL, high pressure, diabetes, nicotine addiction and positive familiar anamnesis) a factor at risk for what regards cardiovascular diseases (CVD), for heart disease (HD) and for angina pectoris and ictus.

High rate of HC are provoked both from a lack of vitamins (they're involved in its metabolism: B6, B12 or folic acid) and from enzymatic defects. Provisionally, it was noticed that there's in vitro a cytotoxic effect of the HC on the vessels' endothelin. Moreover, it was even demonstrated that high rages of HC are observed in patients with illnesses such as the angina pectoris, myocardial infarction and ictus. Recently, a Norwegian study correlated the HC rate and the mortality in patients with a coronary disease. The group with the higher results of HC resulted to have a mortality 4.5 times greater than patients with lower rates. The high rate of HC was associated in many studies to polymorphisms of the enzyme MTHFR, which is responsible of the metabolism of HC in blood, which reduces the activity. In particular, polymorphisms C677T and A1298C contribute to an increase of the risk for the CVS and for deep vein thrombosis (DVT), and they have an high prevalence in patients with CVD (0.41 for C677T and 0.27 for A1298C). Many studies linked the contemporary presence of the mutation C677T and the lox rate of folic acid in blood at an increased risk for the CVD. Homozygotes patients for the polymorphism C677T are associated to high concentrations of HC serum, in particular patients with a low rate of folic acid. Heterozygotes for both polymorphisms have anyway an increased risk for CVD and HC. The polymorphism A1298C in general population isn't associated to high levels of HC, but in patients with CVD it represents a factor at risk for an high rate of HC.

TEST

By means of PCR two fragments of the gene MTHFR are amplified. Here, two polymorphisms are digested by two different restriction enzymes. The obtained fragments are separated through electrophoresis and the results of the digested fragments allow to distinguish the wild-type (normal), the heterozygote (a normal allele and a mutated one) and homozygote (two mutated alleles).

SAMPLE TAKING

Blood/EDTA, 5 ml.

EXECUTION

Daily

COST

According to the federal charge rate of the analyses (2205.06) TP 105

OBSERVATIONS

Recent studies demonstrated a significant association between C677T and A1298C polymorphisms in women and a reduced intrauterine growth.

Laboratorio di diagnostica molecolare

Further information or bibliographic references can be asked to the laboratory.