PROTHROMBIN
(FACTOR II, G2021A)

PATHOGENESIS

The Prothrombin (factor II) is the precursor of serine-protease thrombin, which catalyzes the last reaction in coagulation's cascade: the transformation from fibrinogen to fibrin. The thrombin is a very important enzyme for what regards the equilibrium in coagulation and anticoagulation, because it potentiates the coagulation through a positive synthesis and it promotes the anticoagulation through a mechanisms linked to the protein C. Recently, it was identified a new variant (G20210A) in region 3' of the gene (chromosome 11) that codifies for the prothrombin. This polymorphism is associated to an high risk of thromboembolic events (thrombosis, myocardium heart break, pulmonary embolism, etc.).

EPIDEMIOLOGY

The polymorphism G20210A was found in the 6.2% of patients with thrombosis and in 1-2% of normal patients (relative risk of 2.3 for mutation's carriers). In patients with congenital thrombophilia the mutation was identified in 18% of patients. The same polymorphism was associated to an high risk of the myocardium heart break.

TEST

A fragment of the gene is amplified through PCR. In this fragment it is codified the mutation and after the amplification the gene is digested by means of enzymatic digestion. The obtained fragments are divided through electrophoresis. The restriction enzyme distinguishes between gene with or without mutation.

SAMPLE TAKING

Blood/EDTA, 5 ml.

EXECUTION

Daily.

COST

According to the federal charge rate of the analyses (2205.02) TP 105.

Laboratorio di diagnostica molecolare

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