FACTOR V (LEIDEN, R506Q)

PATOGENESI

In the cascade process leading to blood clotting, activated protein C (APC) has a limiting effect on thrombin formation, as it digests factors V and VIII (coagulation accelerators) by proteolysis. The "activated protein C resistance" phenotype is caused in more than 90% of cases by mutations in the gene coding for Factor V, mainly the Leiden mutation. In patients with the Leiden mutation, the APC is no longer able to inactivate Factor V and therefore to "stop" coagulation. Other predisposing genetic factors for thrombophilia such as antithrombin deficiency, protein C, protein S or hyper-homocystenemia may be present with a factor V mutation. and forced fixed assets. Oral contraceptives are considered in themselves already a risk factor for thrombosis, in healthy women it is spoken of 2-8 times and this risk increases to 35 and 150-500 times if we are in the presence of the Leiden mutation in heterozygous form or respectively homozygous.
60% of thromboses in pregnancy are caused by resistance to activated protein C and in 90% of these we have the presence of the Leiden mutation. EDTA blood, 5 ml. Daily.

EPIDEMIOLOGY

The Leiden mutation has a prevalence of 2-10% in the white population but is not present in Africans and Asians. The R506Q defect in heterozygous form is associated with a 5-10 times greater thrombotic risk, while for the homozygous defect the risk is increased by 50-100 times.

TEST

A fragment of the gene in which the mutation is encoded is amplified by PCR. After amplification, the product is analyzed by restriction fragment analysis. The result distinguishes between a gene with a mutation and a gene without.

SAMPLE TAKING

Blood/EDTA, 5 ml.

EXECUTION

Daily.

Laboratorio di diagnostica molecolare

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