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FII-V AND MTHFR CRITICAL TESTS FOR EVALUATING THE RISK OF THROMBOSIS

CLINICAL RELEVANCE

- The Factor V Leiden mutation is the most common variant associated with inherited thrombosis. This mutation has a high prevalence in the general population (4 - 6% of US Population), and accounts for 85-95% of activated protein C resistant cases.[1] Enhanced risk of venous thrombosis, with the presence the Factor V Leiden variant, with odds ratios (ORs) of 3 to 8 in heterzygotes and 30 to 140 OR in homozygotes.[2]
- The Factor II (Prothrombin) variant gene is the second most common genetic defect for inherited thrombosis.
- Hyperhomocysteinemia is a widely recognized risk factor for coronary artery disease, venous thrombosis, and stroke. It is also involved in the pathogenesis of neural tube defects, stillbirths, and recurrent pregnancy loss. Methlyenetetrahydrofolate reductase (MTHFR) is a key enzyme in the metabolism of homocysteine.

CLINICAL UTILITY

- The risk of thrombosis is substantially increased for patients with multiple genetic risk factors (i.e. The “double hit hypothesis”) including factor V Leiden mutation, hyperhomocysteinemia, protein C deficiency, protein S deficiency and antiphospholipid antibody syndrome(s).[3]
- The increased risk of venous thrombosis in patients who are heterozygous for the prothrombin (G20210A) gene polymorphism is 3-fold. Homozygotes for this polymorphism have been described but are very uncommon. Patients with a previous, or current, thrombotic event that have the prothrombin (G20210A) gene polymorphism are potentially at increased risk for recurrence.[4]

GENETIC VARIANTS

- Factor V Leiden: G1691A
- Factor II (Prothrombin): G20210A
- MTHFR : A1298C and C677T

1. Grody W, Griffin J, Taylor A, Korf B, Heit, J. (2001) American College of Medical Genetics Consensus Statement on Factor V Leiden Leiden Mutation Testing, Genetics in Medicine, 3:2, 139-147.
2. Salomon O. et al; Single and Combined Prothrombotic Factors in Patients With Idiopathic Venous Thromboembolism;
3. Arteriosclerosis Thrombosis and Vascular Biology, 1999, 19: 511-518 © 1999 American Heart Association http://pathology.mc.duke.edu/coag/PTGlflyer2.html
4. http://pathology.mc.duke.edu/coag/PTGlflyer2.html

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