LABOR RENNER

Gene test information

FACTOR V LEIDEN

- **Background**

  The factor V Leiden polymorphism (1691G->A, R506Q) in the factor V gene (F5) is present in approximately 3% of the general population, and in about 20-50% of patients with a history of unexplained recurrent venous thrombosis. The presence of a glutamine (Q) instead of an arginine (R) residue removes a site in coagulation factor V that is normally cleaved by activated protein C, and is as-sociated with resistance to activated protein C. Presence of this polymorphism substantially increases the lifetime risk of venous thrombosis.

- **Factor V Leiden (F5 R506Q) genotypes**

Genotype	Frequency	Commentary
F5 RR	92%	Wild type genotype. No Factor V Leiden variant detectable.
F5 RQ	8%	Heterozygous for factor V Leiden. The relative risk of venous thrombosis is increased approximately 3- to 8-fold.
F5 QQ	< 0.1%	Homozygous for factor V Leiden. The relative risk of venous thrombosis is increased approximately 20- to 80-fold.

- **Indications for testing**

  According to the College of American Pathologists (CAP) Consensus Conference Statement, testing for factor V Leiden is recommended in patients with

  - a history of recurrent VTE, first VTE at younger than 50 years, or first unprovoked VTE at any age,
  - a first VTE at an unusual anatomic site, such as the cerebral, mesenteric, portal, or hepatic veins,
  - a first VTE, at any age, in a subject with a first degree family member with a VTE before the age of 50 years,
  - a first VTE related to pregnancy, the puerperium, oral contraceptive use, or hormone replacement therapy,
  - unexplained pregnancy loss during the second or third trimester.

References:

Renner W, Köppel H, Hoffmann C, Schallmoser K, Stanger O, Toplak H, Wascher TC, Pilger E. Prothrombin G20210A, factor V Leiden, and factor XIII Val34Leu: common mutations of blood coagulation factors and deep vein thrombosis in Austria. Thromb Res. 2000;99:35-9.

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