

Factor V Leiden DNA Screen

Background

The factor V Leiden polymorphism (Arg-506-Gln, 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 instead of an arginine residue removes a site in factor V that is normally cleaved by activated protein C, and is associated with in vitro resistance to activated protein C. Presence of this polymorphism substantially increases the lifetime risk of venous thrombosis. This DNA-based test detects the underlying defect present in almost all cases of resistance to activated protein C.

Indications for Testing

- Evaluate recurrent or familial venous thrombosis
- Carrier testing in family already known to carry the factor V Leiden mutation
- Evaluate recurrent pregnancy loss

Ordering

1. Obtain blood sample - (see Sample Requirements below).
2. Fill out a Clinical Lab Request - Genetics for each patient (available at <http://depts.washington.edu/labweb/Divisions/MolDiag/MolDiagGen/index.htm>). Request: "Factor V Leiden"
3. Call Laboratory Medicine Community Services at (206)598-6066 to arrange the best method of shipment.

Sample Requirements and Specimen Handling

Whole blood - EDTA (purple top) - 5 mL.
Samples should be received within 72 hours of collection.
Samples may be refrigerated until shipped.
For prenatal diagnosis specimens, consult laboratory.
Heparin (green top) tubes are not acceptable.

Test Frequency and Reporting

Test results usually within 1 week of specimen receipt. A written interpretative report is provided.

References

- Svensson PJ, Dahlback B. Resistance to activated protein C as a basis for venous thrombosis. *N Eng J Med* 330:517-522, 1994.
- Bertina RM et al. Mutation in blood coagulation factor V associated with resistance to activated protein C. *Nature* 369:64-67, 1994.
- Ridker PM et al. Mutation in the gene coding for coagulation factor V and the risk of myocardial infarction, stroke, and venous thrombosis in apparently healthy men. *N Eng J Med* 332:912-917, 1995.
- Kujovich JL, Goodnight SH. Factor V Leiden thrombophilia. In: *GeneReviews at GeneTests: Medical Genetics Information Resource (database online)*. Copyright, University of Washington, Seattle. 1997-2003. Available at <http://www.genetests.org>
- Rey E, Kahn SR, David M, Shrier I. Thrombophilic disorders and fetal loss: a meta-analysis. *Lancet* 361:901-908, 2003.