

# FACTOR V LEIDEN

## DESCRIPTION

Mendelian Inheritance in Man number: [227400](#)

Click here for [Gene Reviews](#) Clinical Summary.

Factor V Leiden refers to a specific substitution of G to A at nucleotide 1691 in the Factor V gene. This mutation causes factor V to be inactivated at a significantly slower rate than normal factor V. This delayed inactivation increases the risk for venous thrombotic event (VTE) in the individual. The clinical features of factor V Leiden are extremely variable and depend on the number of factor V alleles and environmental factors. Some individuals with the factor V Leiden mutation may never develop venous thrombosis. Individuals heterozygous for the factor V Leiden mutation are at an increased risk for venous thrombosis by approximately four- to eightfold. Individuals homozygous for the factor V Leiden mutation are at an increased risk by up to 80-fold.

## INDICATIONS FOR DIRECT TESTING

- A first VTE before 50 years of age
- A first unprovoked VTE at any age
- Recurrent VTE
- Venous thrombosis at unusual sites (such as cerebral, mesenteric, portal, and hepatic veins)
- VTE during pregnancy, the puerperium, or in association with oral contraceptive use or hormone replacement therapy
- A first VTE and a strong family history of VTE
- At risk individuals in families with known factor V Leiden mutations

## TESTING METHODOLOGY

The Factor V Leiden molecular genetic test is an allele-specific based approach described by [Hezard et al. \(1997\)](#), for the detection of the 1691G>A mutation. DNA is extracted and amplified using allele-specific primers for the normal and factor V Leiden alleles. The amplicons produced have different sizes, and are separated by agarose gelelectrophoresis. This method allows for complete genotyping of individuals for the factor V Leiden allele.

## SPECIMEN REQUIREMENTS

We require a minimum of 1 milliliter of whole blood. Blood samples must be collected in EDTA (purple topped) tubes.

## TRANSPORT

If the specimen is from clinics within UAB or Kirklin Clinic, please call 934-5562 for pickup. If specimens are being sent from some other location, please ship via **UPS Overnight Priority**. Specimens must be packaged to prevent breakage and absorbent

material must be included in the package to absorb liquids in the event that breakage occurs. Also, the package must be shipped in double watertight containers (e.g. a specimen pouch + the shipping companies Diagnostic Envelope). Please contact us (Email – [mgl@genetics.uab.edu](mailto:mgl@genetics.uab.edu), Phone – 205-934-5562) prior to sample shipment and provide us with the date of shipment and the tracking number of the package.

**TURN AROUND TIME:** 7-10 working days

**CPT CODES AND PRICES:** \$170 USD

83891, 83894 (x2), 83900 (x2), 83912

**Please note that prices listed correspond to institutional rates; please contact the lab for insurance rates.**

Payment may be made by Money order, Cashier's check, VISA or MasterCard. We will also bill requesting institutions. Please call for more information.

## **REQUIRED FORMS**

### [Test Requisition Form](#)

**Note:** Requests for Molecular Genetic testing for Factor V Leiden will **not** be accepted for the following reasons:

- No label (patients full name and date of collection) on the specimens
- No referring physician's or genetic counselor's names and addresses
- No billing information if being paid for by an institution
- No informed consent

**For more information, test requisition forms, or sample collection, please call: 205-934-5562.**

## **REFERENCES**

[Hézar, N., Corneillet, P., Droulle, C., Gillot, L., Potron, Gerard, and Nguyen, P. \(1997\) Factor V Leiden: detection in whole blood by ASA PCR using an additional mismatch in an antepenultimate position. \*Thrombosis Research\*. 88\(1\): 59-66.](#)

[Hézar, N., Cornillet-Ldfebvre, P., Gillot, L., Potron, G., Nguyen, P., \(1998\) Multiplex ASA PCR for a simultaneous determination of Factor V Leiden gene, G>A 20210 Prothrombin gene and C>T 677 MTHFR gene mutations. \*Thromb. Haemost.\* 79: 1054-1055.](#)

[Reich LM, Bower M, Key NS. \(2003\) Role of the geneticist in testing and counseling for inherited thrombophilia. \*Genet Med.\* 2003 May-Jun;5\(3\):133-43.](#)