

Assay Summary

F5 Gene Mutation Analysis

Factor V Leiden

Synopsis

The factor V Leiden mutation, also designated as 1691 G>A or R506Q, is the major heritable risk factor for venous thromboembolism^{1,2}. This mutation in the coagulation factor V gene results in resistance of factor V to inactivation by activated protein C (APC)^{3,4,5}. Approximately 5% of Caucasians are either heterozygous carriers or homozygous for the factor V Leiden mutation; the prevalences are lower for other ethnic groups^{6,7}.

Indications for testing

Individuals with a family history of deep vein thromboembolism, members of families with a previously identified factor V Leiden mutation, patients with a first episode of deep vein thromboembolism, individuals undergoing major surgery, and women initiating oral contraceptive use may consider testing for the factor V Leiden mutation.

Methodology

An allele-specific amplification method called bi-directional amplification of specific alleles (Bi-PASA)⁸ is used to analyze the factor V Leiden mutation status in patient DNA samples. The Bi-PASA method distinguishes among heterozygous carriers (one copy), homozygous carriers (two copies), and noncarriers (negative) of the factor V Leiden mutation.

Performance

The sensitivity and specificity of the assay for the factor V Leiden mutation is greater than 98%. About 90% of hereditary APC resistance is due to factor V Leiden. Approximately 50% of patients with venous thromboembolism are carriers of at least one copy of the factor V Leiden mutation.

Limitations

This assay will only detect the R506Q mutation of the factor V gene. Other mechanisms of APC resistance are not determined by this method.

Specimen Requirements

- (a) Blood samples: 2 tubes with a total of 6 ccs in ACD (yellow top) or EDTA (lavender top) tubes.
Keep at ambient temperature and ship by overnight courier. Samples must be received in our laboratory within 72 hours of draw.

Note:

- i) for infants, a minimum of 3 ccs is sufficient.
- ii) we accept DNA; at least 10 micrograms is required.

- (b) Prenatal samples: 2 T25 flasks of confluent cells sent padded to arrive on M/Tu/W.

A blood sample from the mother maybe required (2 tubes with a total of 6 ccs in ACD (yellow top) or EDTA (lavender top) tubes) for use as positive control. Maternal cell contamination studies are not done here but are required for autosomal disorders and dosage analysis on X-linked disorders. We would be happy to assist in coordinating sending out a specimen for this purpose.

Test Request Form (TRF)

A completed MDL [TRF](#) is required for each specimen. Please submit the completed TRF with the specimen. Complete testing and billing information must be provided before the specimen is processed.

<i>Order Codes</i>	<i>CPT Codes</i>	<i>TAT</i>
F5LD (Factor V Leiden, single mutation analysis)	83890, 83901, 83894, 83912	3 wks

References

1. Voorberg, J. et al. (1994). Lancet 343:1535-1536.
2. Zoller, B. and Dahlback, B. (1994). Lancet 343:1536-1538.
3. Bertina, R. M. et al. (1994). Nature 369:64-67.
4. Greengard, J. S. et al. (1994). Lancet 343:1361-1362.
5. Sun, X. et al. (1994). Blood 83:3120-3125.
6. Gregg, J. P. et al. (1997). American Journal of Medical Genetics 73:334-336.
7. Ridker, P. M. et al. (1997). JAMA 277:1305-1307.
8. Liu, Q. et al. (1997). Genome Research 7:389-398.

NOTE: This test is performed pursuant to a license agreement with Roche Molecular Systems, Inc.