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 ACP resistance are not determined by this method.

Specimen Requirements
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.

Test Request Form (TRF)
A completed CMDL 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.

<table>
<thead>
<tr>
<th>Order Codes</th>
<th>CPT Codes</th>
<th>TAT</th>
</tr>
</thead>
<tbody>
<tr>
<td>F5LD (Factor V Leiden, single mutation analysis)</td>
<td>81241, G0452</td>
<td>1 wk</td>
</tr>
</tbody>
</table>

**References**


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