Factor V Leiden (R506Q) Mutation, Blood

**Test Definition: F5DNA** 

# **Reporting Title:** Factor V Leiden (R506Q) Mutation, B **Performing Location:** Rochester

MAYO CLINIC

ABORATORIES

### Ordering Guidance:

This assay will only detect the F5 c.1601G>A; p.Arg534Gln (rs6025) variant associated with factor V Leiden thrombophilia. To detect other pathogenic alterations in the F5 gene of a patient with a laboratory diagnosis of coagulation factor V deficiency, order F5NGS / F5 Gene Next Generation Sequencing, Varies.

This assay will not will not detect alterations in individuals with activated protein C (APC)-resistance caused by mechanisms other than the F5:c.1601G>A, p.Arg534Gln variant. Coagulation-based activated protein C (APC)-resistance ratio (mixing with factor V-deficient plasma) is recommended as the initial screening assay for APC-resistance. Depending on the assay system, the APC-resistance ratio may be indeterminate for patients with a lupus anticoagulant or extremely high heparin levels. For more information, see APCRV / Activated Protein C Resistance V (APCRV), Plasma or APCRR / Activated Protein C Resistance V (APCRV), with Reflex to Factor V Leiden, Blood and Plasma.

### Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.
Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: Yellow top (ACD solution B), light-blue top (sodium citrate)
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube. Do not aliquot.

### Specimen Minimum Volume:

1 mL

### Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Coagulation Patient Information (T675) in Special Instructions

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)	14 days	



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Frozen	14 days	
Refrigerated	14 days	

## **Result Codes:**

Result ID	Reporting Name	Туре	Unit	LOINC®
21838	Factor V Leiden (R506Q) Mutation, B	Alphanumeric		21668-9
21839	F5DNA Interpretation	Alphanumeric		69049-5
21841	F5DNA Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

# **Supplemental Report:**

No

## **CPT Code Information:**

81241

# **Reference Values:**

Negative