# Factor V (F5) Leiden (R506Q) Mutation by Real-Time PCR

# **Clinical Indication and Relevance**

Please refer to "American College of Medical Genetics Consensus Statement on Factor V Leiden Mutation Testing".

http://www.acmg.net/StaticContent/StaticPages/Factor\_V.pdf

# Methodology

DNA is isolated from the patient sample and the Factor V gene containing the Leiden mutation site is PCR-amplified and analyzed using an allelic discrimination assay employing primers and TaqMan probes. Results are reported as normal, heterozygous or homozygous for the factor V Leiden mutation.

#### **Sensitivity**

N/A

#### **Turn-around Time**

Five to ten working days

# **Sample Requirements**

#### **Collect**

Peripheral blood (PB): 3-5 mL (1 mL minimum), in purple top (sodium EDTA) tube; yellow top (ACD) tube acceptable.

### **Transport**

Ambient or 2-8°C (wet ice or cold packs). Do not freeze.

### **Stability**

Ambient - 8 hours; refrigerated - 48 hours.

# **Unacceptable Samples**

Serum or plasma; frozen peripheral blood; clotted blood; severely hemolyzed blood samples.

# **CPT Code(s)**

81241: F5 (coagulation Factor V) gene analysis, Leiden variant

G0452-26: Molecular pathology procedure; physician interpretation and report