

TEST CHANGE

Factor XIII (F13A1) V34L Variant

2003220, FAC 13 MUT

Specimen Requirements:

Patient Preparation:

Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Transport Temperature: Refrigerated.

Unacceptable Conditions: Frozen specimens in glass collection tubes.

Remarks:

Stability: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

Methodology: Polymerase Chain Reaction (PCR) / Fluorescence Monitoring

Performed: ~~Varies~~ **Mon, Thu**

Reported: 2-7 days

Note:

CPT Codes: 81400

New York DOH Approval Status: This test is New York DOH approved.

Interpretive Data:

Background Information for Factor XIII (F13A1) V34L Variant:

Characteristics: The Factor XIII (F13A1) ~~V34L~~ V34L sequence variant is a protective factor against pulmonary embolism, deep vein thrombosis, and myocardial infarction in Caucasians. ~~?~~ It may also have a slight protective effect against coronary artery disease. Limited data suggests the V34L sequence variant may also be associated with idiopathic spontaneous subconjunctival hemorrhage (SSH), but this finding has not been confirmed.

Allele Frequency: Caucasian 0.27, African American 0.17, American Indian 0.29, Asian 0.01.

Inheritance: Autosomal dominant.

Cause: Homozygosity or heterozygosity for F13A1; V34L

Variant Tested: F13A1 c.103G>T; p.Val34Leu.

Clinical Sensitivity: Varies by ethnicity.

Methodology: Polymerase chain reaction and fluorescence monitoring.

Analytical Sensitivity and Specificity: 99 percent.

Limitations: Mutations in the F13A1 or F13B genes, other than the V34L sequence variant, are not evaluated. Diagnostic errors can occur due to rare sequence variations.

The protective effect of the V34L sequence variant has not been established for ethnicities other

than Caucasian and may be altered by other genetic and nongenetic factors not assessed by this assay.

Reference Interval:

By report
