

Client: Example Client ABC123 123 Test Drive Salt Lake City, UT 84108 UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB 1/20/1990 **Gender:** Female

Patient Identifiers: 01234567890ABCD, 012345

Visit Number (FIN): 01234567890ABCD **Collection Date:** 00/00/0000 00:00

Factor XIII (F13A1) V34L Variant

ARUP test code 2003220

Factor XIII Variant Specimen

Whole Blood

Factor XIII (F13A1) V34L Variant

Heterozygous

Indication for testing: Assess genetic risk for thrombosis.

HETEROZYGOUS: One copy of the Factor XIII (F13A1), V34L sequence variant was detected. In Caucasians, this genotype is associated with a decreased risk for pulmonary embolism and deep vein thrombosis, a modest risk reduction for myocardial infarction and a slight decrease in coronary artery disease.

Recommendations: This result should be interpreted in the context of clinical and other laboratory findings as thrombotic risk is influenced by other genetic and non-genetic factors not assessed by this assay. If clinically indicated, testing for other inherited or acquired thrombophilic disorders is recommended; consider DNA testing for the factor V Leiden mutation, measurement of total plasma homocysteine concentration, serological assays for anticardiolipin antibodies and multiple phospholipid-dependent coagulation assays for lupus inhibitor.

This result has been reviewed and approved by

H=High, L=Low, *=Abnormal, C=Critical

4848



Background Information: Factor XIII (F13A1) V34L Variant

Characteristics: The Factor XIII (F13A1), 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.Va134Leu. Clinical Sensitivity: Varies by ethnicity. Methodology: Polymerase chain reaction and fluorescence Methodology: Polymerase chain reaction and fluorescence methodology: Polymerase Chain reaction and Tluorescence 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 non-genetic factors not assessed by this assess.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Factor XIII Variant Specimen	23-125-134509	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Factor XIII (F13A1) V34L Variant	23-125-134509	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

this assay.

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

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