

Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: arup, arup

Patient: Test, PAC 13 MUT

DOB

Sex: Male

Patient Identifiers: 46901

Visit Number (FIN): 47239

Collection Date: 3/3/2023 10:35

Factor XIII (F13A1) V34L Variant

ARUP test code 2003220

Factor XIII Variant Specimen whole Blood

Factor XIII (F13A1) V34L Variant Negative

Indication for testing: Assess genetic risk for thrombosis.

NEGATIVE: The Factor XIII (F13A1), V34L sequence variant, was not detected.

Recommendations: This result should be interpreted in the context of clinical and 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 [REDACTED]

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.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 non-genetic factors not assessed by this assay.

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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Test, PAC 13 MUT

ARUP Accession: 23-062-103952

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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	23-062-103952	3/3/2023 10:35:00 AM	3/3/2023 10:35:24 AM	4/6/2023 2:29:00 PM
Factor XIII (F13A1) V34L Variant	23-062-103952	3/3/2023 10:35:00 AM	3/3/2023 10:35:24 AM	4/6/2023 2:29:00 PM

END OF CHART

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

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