ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/17/1972
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Factor V Leiden (F5) R506Q Mutation

ARUP test code 0097720

FACV Specimen	Whole Blood
Factor V Leiden (F5) R506Q Mutation	Heterozygous *

Indication for testing: Assess genetic risk for thrombosis.

HETEROZYGOUS: One copy of the factor V Leiden variant, c.1601G>A; p.Arg534Gln, was detected. This is associated with activated protein C resistance and a four to eight fold increased risk for venous thrombosis in comparison to individuals without this variant. Genetic consultation is recommended.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Factor V Leiden (F5) R506Q Mutation

CHARACTERISTICS: Venous thromboembolism (VTE) is multifactorial caused by a combination of genetic and environmental factors. The Factor V Leiden (FVL) variant is the most common cause of inherited VTEs, accounting for over 90 percent of activated protein C (APC) resistance. Because the FVL variant eliminates the APC cleavage site, factor V is inactivated slower, thus persisting longer in blood circulation, leading to more thrombin production. Other genetic risk factors for VTE include, male sex and variants in antithrombin, protein C, protein S, or factor XIII. Non-genetic risk factors include, age, smoking, prolonged immobilization, malignant neoplasms, surgery, pregnancy, oral contraceptives, estrogen replacement therapy, tamoxifen and raloxifene therapy.
INCIDENCE OF FACTOR V LEIDEN VARIANT: Approximately 5 percent of Caucasians, 2 percent of Hispanics, 1 percent of African Americans and 0.5 percent of Asians are heterozygous; homozygosity occurs in 1 in 1500 Caucasians.
INHERITANCE: Semi-dominant; both heterozygotes and homozygotes are at increased risk for VTE.
PENETRANCE: Lifetime risk of VTE is 10 percent for heterozygotes and 80 percent of homozygotes.
CAUSE: The pathogenic gain of function in the F5 gene variant c.1601G>A (p.Arg534Gln). Legacy nomenclature: R506Q (1691G>A)
CLINICAL SENSITIVITY: 20-50 percent of individuals with an isolated VTE have the FVL variant.
METHODOLOGY: Polymerase chain reaction and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. F5 gene mutations, other than p.Arg534Gln, will not be detected.

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
FACV Specimen	23-027-131227	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Factor V Leiden (F5) R506Q Mutation	23-027-131227	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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