

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 8/15/1993
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Prothrombin (F2) c.*97G>A (G20210A) Pathogenic Variant

ARUP test code 0056060

PT PCR Specimen whole Blood

Prothrombin (F2) G20210A Variant

Homozygous *

Indication for testing: Assess genetic risk for thrombosis.

HOMOZYGOUS: Two copies of the Factor II, prothrombin G20210A mutation were detected. This genotype is associated with elevated prothrombin levels and an increased risk for venous thrombosis.

Recommendations: If clinically indicated, testing for other inherited or acquired thrombophilic disorders is recommended including DNA testing for the factor V Leiden mutation, measurement of total plasma homocysteine concentration, serological assays for anticardiolipin antibodies, multiple phospholipid-dependent coagulation assays for lupus inhibitor, protein C activity, protein S activity or free protein S antigen, and antithrombin activity.

This result has been reviewed and approved by [REDACTED]

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

BACKGROUND INFORMATION: Prothrombin (F2) c.*97G>A (G20210A) Pathogenic Variant

CHARACTERISTICS: The Factor II, c.*97G>A (G20210A) pathogenic variant is a common genetic risk factor for venous thrombosis associated with elevated prothrombin levels leading to increased rates of thrombin generation and excessive growth of fibrin clots. The expression of Factor II thrombophilia is impacted by coexisting genetic thrombophilic disorders, acquired thrombophilic disorders (eg, malignancy, hyperhomocysteinemia, high factor VIII levels), and circumstances including: pregnancy, oral contraceptive use, hormone replacement therapy, selective estrogen receptor modulators, travel, central venous catheters, surgery, and organ transplantation.

INCIDENCE: Approximately 2 percent of Caucasians and 0.3 percent of African Americans are heterozygous; homozygosity occurs in 1 in 10,000 individuals.

INHERITANCE: Incomplete autosomal dominant.

PENETRANCE: The risk of thrombosis is increased 2-4 fold for heterozygotes and further increased for homozygotes.

CAUSE: Homozygosity or heterozygosity for F2 c.*97G>A (G20210A).

PATHOGENIC VARIANT TESTED: F2 c.*97G>A (G20210A).

CLINICAL SENSITIVITY FOR VENOUS THROMBOSIS: Approximately 10 percent.

METHODOLOGY: Polymerase chain reaction and fluorescence monitoring.

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.

LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. F2 gene variants, other than c.*97G>A (G20210A), 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
PT PCR Specimen	22-325-144078	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Prothrombin (F2) G20210A Variant	22-325-144078	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