

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

Physician: Doctor, Example

Patient: Patient, Example

DOB 2/12/1948
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

Negative

Indication for testing: Assess genetic risk for thrombosis.

NEGATIVE: The Factor II, prothrombin G20210A mutation, was not detected. Other causes of elevated prothrombin levels and hereditary forms of venous thrombosis have not been excluded.

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

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

4848



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),

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	23-026-116720	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Prothrombin (F2) G20210A Variant	23-026-116720	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

will not be detected.

END OF CHART

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

Patient: Patient, Example
ARUP Accession: 23-026-116720
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Page 2 of 2 | Printed: 2/1/2023 11:41:42 AM
4848