

LABORATORY REPORT

Example Client, XYZ123 1234 Warde Road Ann Arbor MI 48108 **EXAMPLE, REPORT**

WX0000003039 M 12/05/1988 34 Y

Molecular

Collected: 02/19/2022 06:16 Received: 02/19/2022 06:16

Test Name Result Flag Ref-Ranges Units Site

Prothrombin 20210A Mutation Analysis

Prothrombin 20210A Mutation Analysis

Heterozygous

AB

WMRL

Both the wild type (WT) F2 gene and the c.*97G>A pathogenic variant (G20210A) were detected indicating a heterozygous c.*97G>A genotype for this specimen. Heterozygosity for the c.*97G>A variant is associated with overproduction of F2 prothrombin and an increased risk for venous thrombosis.

Prothrombin thrombophilia is inherited in an autosomal dominant manner and adults heterozygous for the c.*97G>A variant have a 2 to 5-fold increased risk of thrombosis. The prevalence of c.*97G>A heterozygosity is 2-5% in individuals of European descent. The variant is extremely rare in individuals of African, Asian, or Native American descent. The clinical expression of prothrombin thrombophilia is variable. Many heterozygous individuals never develop thrombosis. Patients heterozygous for both c.*97G>A variant and the Factor V Leiden mutation often experience earlier onset of thrombosis that tends to be more severe than presence of the individual alleles. Genetic counseling is recommended to help determine the benefit of testing asymptomatic family members.

This test was performed using the cobas® Factor II Test (Roche) - an in vitro diagnostic device that uses real-time quantitative Polymerase Chain Reaction (qPCR) for the detection and genotyping of the human Factor II (F2) gene. The test detects the presence of the wild type (WT) F2 gene and the pathogenic c.*97G>A variant (also known as G20210A) in genomic DNA isolated from whole blood specimens as an aid in diagnosing patients with suspected thrombophilia. The cobas® Factor II Test and the cobas z 480 analyzer are used together for automated amplification and detection. The limit of detection for this test is 0.1 ng/uL of genomic DNA (2.5 ng/PCR reaction).

Reported Date: 02/19/2022

06:16

F2PM
Performing Site:

WMRL: WARDE MEDICAL LABORATORY 300 West Textile Road Ann Arbor MI 48108

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

D419000008 WX0000003039 Printed D&T: 12/01/23 10:15 Ordered By: CLIENT CLIENT WX0000000000001595

Kajal V. Sitwala, MD, PhD - Medical Director Form: MM RL1 PAGE 1 OF 1