



LABORATORY REPORT

QC ACCOUNT (WARDE)
300 W. TEXTILE
ANN ARBOR MI 48108

EXAMPLE, REPORT
WX0000003481 F 12/08/1988 34 Y

Referral Testing

Collected: 03/18/2021 10:48 Received: 03/18/2021 10:48

Table with 6 columns: Test Name, Result, Flag, Ref-Ranges, Units, Site. Row 1: Factor XIII V34L Mutation Analysis, SEE NOTE, QCRL

RESULT: HETEROZYGOUS POSITIVE FOR THE p.V34L VARIANT IN THE FACTOR XIII GENE

Interpretation: This patient has one copy of the p.V34L variant in the Factor XIII gene. The p.V34L variant has been associated with a protective effect against myocardial infarction and an increased risk for intracerebral hemorrhage.

Factor XIII (FXIII) plays a crucial role in fibrin crosslinking during the coagulation process. One variant in the FXIII gene, p.V34L, is associated with higher rates of FXIII activity and appears to be protective against myocardial infarction (MI). The presence of the p.V34L variant (with a frequency of 0.50 in the white population) causes an approximate 126% increase in FXIII activity in heterozygotes and 176% in homozygotes (Thromb Haemost 1998; 80:704). The protective effect of elevated FXIII activity is not well understood but it may interfere with fibrin crosslinking directly or indirectly.

The p.V34L variant is detected by polymerase chain reaction amplification of a portion of exon 2 of the FXIII gene, single nucleotide primer extension, and detection of fluorescent extension products on an automated DNA sequencer.

This analysis evaluates only the specified mutation in the FXIII gene and cannot detect other FXIII mutations, or mutations in other genes, that may similarly affect FXIII activity. Furthermore, genetic variation and other factors can affect the accuracy of direct mutation testing. Therefore, the results of this testing should always be interpreted in light of the appropriate clinical and familial data. For assistance with the interpretation of these results, please contact your local Quest Diagnostics genetic counselor or call 1-866-GENEINFO (436-3463).

This test is performed pursuant to a license agreement with Orchid Biosciences, Inc.

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols Institute San Juan Capistrano. It has not been cleared or approved by FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

Test Performed at:
Quest Diagnostics Nichols Institute
33608 Ortega Highway

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



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Test Name Result Flag Ref-Ranges Units Site
San Juan Capistrano, CA 92690-6130 I Maramica MD, PhD, MBA
Reported Date: 03/18/2021 10:54 F13MA

Performing Site:

QCRL: QUEST DIAGNOSTICS REFERENCE LAB CAPISTRANO 33608 Ortega Highway San Juan Capistrano CA 92675

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

C318000004 Ordered By: CLIENT CLIENT
WX0000003481 WX00000000002063
Printed D&T: 11/30/23 14:16

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