

## LABORATORY REPORT

Example Client, XYZ123 1234 Warde Road Ann Arbor MI 48108

#### **EXAMPLE, REPORT W**

WX000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 09/25/2023 09:32 Received: 09/25/2023 09:32

**Test Name** Result Flag Ref-Ranges Units Site

JAK2 V617F Cascading Reflex

QCRL Clinical Indication: NA OCRI **WB EDTA** Specimen Source: QCRL Block/Specimen ID: NA QCRL JAK2 V617F Mutation NOT DETECTED

Reference Range: NOT DETECTED Test Performed at: Quest Diagnostics Nichols Institute 33608 Ortega Highway

San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

QCRL **CALR Exon 9 Mutation** NOT DETECTED

Reference Range: NOT DETECTED Test Performed at:

Quest Diagnostics Nichols Institute 33608 Ortega Highway

San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

QCRL NOT DETECTED JAK2 Exon 12 Mutation

Reference Range: NOT DETECTED Test Performed at:

Quest Diagnostics Nichols Institute

33608 Ortega Highway

San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

QCRL MPL Exon 10 Mutation NOT DETECTED

Reference Range: NOT DETECTED Test Performed at: Quest Diagnostics Nichols Institute 33608 Ortega Highway

San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

QCRL CSF3R Exon 14/17 Mutation **NOT DETECTED** 

Reference Range:

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

F325000022 WX000003827 Printed D&T: 09/25/23 09:37 Ordered By: KAJAL SITWALA, MD, PhD

WX0000000002365

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

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NOT DETECTED

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QCRL Gene QCRL Amino Acid OCRI **Mutation Frequency** QCRL Mutation Type QCRL Exon QCRL **Nucleotide Change** OCRI Reference QCRL Interpretation SFF NOTE

No mutations were detected in codon 617 or exon 12 of JAK2, or exon 9 of CALR, or exon 10 (codons 505 and 515) of MPL, or exons 14 and 17 of CSF3R. Insertions up to 30bp and deletions up to 52bp have been successfully detected by the assay. The absence of mutation does not exclude the presence of a myeloproliferative neoplasm. See below for additional recommendations on testing.

This data was reviewed and interpreted by Charles Ma, PhD. HCLD(ABB)

Assay Details

SEE NOTE

QCRL

This PCR-based advanced sequencing assay interrogates DNA from leukocytes for the presence of mutations in codon 617 and exon 12 of JAK2, exon 9 of CALR, exon 10 (codons 505 and 515) of MPL and exons 14 and 17 of CSF3R. The sensitivity of mutation detection is approximately 5%, but may vary depending on the particular mutation type. Alterations outside of the tested areas of these genes will not be detected. Synonymous or known non-synonymous polymorphic changes (SNPs) are not reported. JAK2 V617F mutation is associated with myeloproliferative neoplasms (MPNs), including polycythemia vera (PV), essential thrombocythemia (ET) and primary myelofibrosis (PMF);  ${\tt JAK2}$ exon 12 mutations with PV; CALR exon 9 indels with ET and PMF; MPL exon 10 mutations with ET and PMF and CSF3R mutations with atypical chronic myeloid leukemia (aCML), chronic neutrophilic leukemia (CNL) and other myeloid neoplasms. Increasing allele burden of JAK2 V617F in MPNs has been shown in a number of studies to be associated with increased symptoms including classification. To further evaluate for MPNs if this assay is negative, additional testing for BCR-ABL1 rearrangement (test code 91065 or 12070) can be performed.

Tests not indicated have been cancelled and credited.

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<u>Test Name</u> <u>Result</u> <u>Flag Ref-Ranges</u> <u>Units</u> <u>Site</u>

DNA was aligned to GRCh37(hg19) for analysis.

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

San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

Billing Only - CALR Exon 9 Mutation

BILL

Billing Only - JAK2 exon 12 Mutation

BILL

GCRL

GURL

GUR

Performing Site:

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

**Reported Date:** 2023.09.25 9:37 JAKCR

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

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