



LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 03/18/2024 14:25 Received: 03/18/2024 14:25

Test Name	Result	Flag	Ref-Ranges	Units	Site
MPN Core Diagnostics Panel					
Clinical Indication:	PV				QCRL
Specimen Source:	Whole Blood				QCRL
Block/Specimen ID:	123456				QCRL
JAK2 V617F Mutation	NOT DETECTED				QCRL
Reference Range:	NOT DETECTED				
JAK2 Exon 12 Mutation	NOT DETECTED				QCRL
Reference Range:	NOT DETECTED				
CALR Exon 9 Mutation	NOT DETECTED				QCRL
Reference Range:	NOT DETECTED				
MPL Exon 10 Mutation	NOT DETECTED				QCRL
Reference Range:	NOT DETECTED				
Gene 1	SEE NOTE				QCRL
MPL					
Amino Acid 1	SEE NOTE				QCRL
p.Trp515Leu					
Mutation Frequency 1	SEE NOTE				QCRL
84.1					
Mutation Type 1	SEE NOTE				QCRL
missense					
Exon 1	SEE NOTE				QCRL
Exon 10					

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

F918000035
WX0000003827
Printed D&T: 03/18/24 14:29

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002365

Kajal V. Sitwala, MD, PhD - Medical Director
Form: MM RL1
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Nucleotide Change 1	SEE NOTE				QCRL
NM_005373.2:c.1544G>T					
Reference 1	SEE NOTE				QCRL
COSM27289					
Gene 2	.				QCRL
Amino Acid 2	.				QCRL
Mutation Frequency 2	.				QCRL
Mutation Type 2	.				QCRL
Exon 2	.				QCRL
Nucleotide Change 2	.				QCRL
Reference 2	.				QCRL
Interpretation	SEE NOTE				QCRL

A mutation is detected in codon 515 (exon 10) of MPL. Mutations of this type are associated with essential thrombocythemia (ET) and primary myelofibrosis (PMF). MPL mutational analysis can be considered on sequential samples to assess for treatment response.

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

Assay Details	SEE NOTE				QCRL
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This Next-Generation Sequencing based assay interrogates DNA from leukocytes for the presence of mutations in exon 12 and exon 14 of JAK2 (including codon 617), exon 9 of CALR and exon 10 of MPL (including codons 505 and 515). The sensitivity of mutation detection is approximately 5% but may vary depending on the mutation type. Insertions up to 30bp and deletions up to 52bp have been successfully detected by the assay. 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); JAK2 exon 12 mutations with PV; CALR exon 9 indels and MPL exon 10 mutations with ET and PMF. Increasing allele burden of JAK2 V617F in MPNs has been shown in a number of studies to be associated with increased symptoms including pruritis, splenomegaly, and leukocytosis. Results of this assay should

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be correlated with morphology and other laboratory testing for final diagnosis and classification. To further evaluate for MPNs if this assay is negative, additional testing options including BCR-ABL1 rearrangement, Leukovantage MPN, or CSF3R mutation analysis can be considered.

DNA was aligned to GRCh37 (hg19) for analysis. The transcripts IDs used as reference sequences are ENST00000381652 (JAK2), ENST00000316448 (CALR) and ENST00000372470 (MPL).

For additional information, please refer to <http://education.QuestDiagnostics.com/faq/FAQ211> (This link is being provided for informational/educational purposes only.)

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics. 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

Reported Date: 03/18/2024 14:29 MPNCP

Performing Site:

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

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