Warde Medical Laboratory

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: 09/14/2023 11:25 Received: 09/14/2023 11:25

Test Name	Result	Flag	Ref-Ranges	Units	Site
MPL Mutation Analysis					
Clinical Indication:	NA				QCRL
Specimen Source:	NA				QCRL
Block/Specimen ID:	NA				QCRL
MPL Exon 10 Mutation	**DETECTED**	**AB**			QCRL
Reference Range: NOT DETECTED					
Gene	SEE NOTE				QCRL
MPL					
Amino Acid	SEE NOTE				QCRL
p.Trp515Leu					
Mutation Frequency	SEE NOTE				QCRL
29.3					
Mutation Type	SEE NOTE				QCRL
missense					
Exon	SEE NOTE				QCRL
Exon 10					
Nucleotide Change	SEE NOTE				QCRL
c.1544G>T					
Reference	SEE NOTE				QCRL
COSM18918					
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 performed

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on sequential samples to assess for treatment response.

Results reviewed by M.R. Sheikholeslami, M.D.

Assay Details SEE NOTE QCRL

This PCR-based advanced sequencing assay interrogates DNA from leukocytes for the presence of mutations in exon 10 of the thrombopoietin receptor (MPL), including codons 505 and 515. The sensitivity of mutation detection is 5% but may vary depending on the particular 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 this gene will not be detected. Synonymous or known non-synonymous polymorphic changes (SNPs) are not reported. Mutations at these sites in MPL are associated with myeloproliferative neoplasms (MPNs), particularly essential thrombocythemia (ET) and primary myelofibrosis (PMF). Results of this assay should be correlated with morphology and other laboratory testing for final diagnosis and classification. If this test is negative, additional testing that may be useful for workup of MPNs, depending on presenting hematologic features, includes BCR-ABL1 rearrangement (test code 91065 or 12070X) or mutational analysis of JAK2 V617F (polycythemia vera (PV)/ET/PMF, 92473), CALR (ET/PMF, 92475), JAK2 exon 12 (PV, 92474) or CSF3R (chronic neutrophilic leukemia, 92477). Residual material from this sample may be used except for BCR-ABL1 testing; call lab to add.

DNA was aligned to GRCh37(hg19) for analysis and transcript ID ENST00000372470 was used as reference for MPL sequence.

For additional information, please refer to
(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 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

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

Reported Date: 2023.09.14 11:26 MPLQL