MAYO CLINIC LABORATORIES

Test Definition: MPLJM

MPL Exon 10 Variant Detection, Reflex, Bone Marrow

Overview

Useful For

Aiding in the distinction between a reactive cytosis and a chronic myeloproliferative disorder

Evaluating for variants in *MPL* in an algorithmic process

Method Name

Only orderable as a reflex. For more information see MPNJM / Myeloproliferative Neoplasm, *JAK2* V617F with Reflex to *CALR* and *MPL*, Bone Marrow.

Sanger Sequencing

NY State Available

No

Specimen

Specimen Type

Bone Marrow

Specimen Required

Only orderable as a reflex. For more information see MPNJM / Myeloproliferative Neoplasm, *JAK2* V617F with Reflex to *CALR* and *MPL*, Bone Marrow.

Container/Tube: Lavender top (EDTA)
Specimen Volume: 2 mL
Collection Instructions:
1. Invert several times to mix bone marrow.
2. Send specimen in original tube. **Do not** aliquot.
3. Label specimen as bone marrow.

Specimen Minimum Volume

0.5 mL

Reject Due To

Gross hemolysis	Reject
Moderately to	Reject

severely clotted	

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Bone Marrow	Ambient (preferred)	7 days	
	Refrigerated	7 days	

Clinical & Interpretive

Clinical Information

The Janus kinase 2 gene (*JAK2*) codes for a tyrosine kinase (*JAK2*) that is associated with the cytoplasmic portion of a variety of transmembrane cytokine and growth factor receptors important for signal transduction in hematopoietic cells. Signaling via *JAK2* activation causes phosphorylation of downstream signal transducers and activators of transcription (STAT) proteins (eg, STAT5) ultimately leading to cell growth and differentiation. *BCR-ABL1*-negative myeloproliferative neoplasms (MPN) frequently harbor an acquired single nucleotide variant in *JAK2* characterized as c.G1849T; p. Val617Phe (V617F). *JAK2* V617F is present in 95% to 98% of polycythemia vera and 50% to 60% of primary myelofibrosis (PMF) and essential thrombocythemia (ET) cases. It has also been described infrequently in other myeloid neoplasms, including chronic myelomonocytic leukemia and myelodysplastic syndrome. Detection of *JAK2* V617F is useful to help establish the diagnosis of MPN. However, a negative *JAK2* V617F result does not indicate the absence of MPN. Other important molecular markers in *BCR-ABL1*-negative MPN include *CALR* exon 9 variant (20%-30% of PMF and ET) and *MPL* exon 10 variant (5%-10% of PMF and 3%-5% of ET). Variants in *JAK2*, *CALR*, and *MPL* are essentially mutually exclusive. A *CALR* variant is associated with decreased risk of thrombosis in both ET and PMF and confers a favorable clinical outcome in PMF patients. A triple negative (*JAK2* V617F, *CALR*, and *MPL*-negative) genotype is considered a high-risk molecular signature in PMF.

Reference Values

Only orderable as a reflex. For more information see MPNJM / Myeloproliferative Neoplasm, *JAK2* V617F with Reflex to *CALR* and *MPL*, Bone Marrow.

An interpretive report will be provided.

Interpretation

An interpretation will be provided.

Cautions

A positive result is not specific for a particular subtype of myeloproliferative neoplasm and clinicopathologic correlation is necessary in all cases.

A negative result does not exclude the presence of a myeloproliferative neoplasm or other neoplastic process.

Analytical sensitivity is approximately 20%, meaning there must be about 20% of the variant DNA in the specimen for

reliable detection.

Clinical Reference

1. Tefferi A, Lasho TL, Finke CM, et al: CALR vs JAK2 vs MPL-mutated or triple-negative myelofibrosis: clinical, cytogenetic and molecular comparisons. Leukemia. 2014;28(7):1472-1477. doi:10.1038/leu.2014.3
2. Pikman Y, Lee BH, Mercher T, et al: MPLW515L is a novel somatic activating mutation in myelofibrosis with myeloid metaplasia. PLoS Med. 2006;3:e270. doi:10.1371/journal.pmed.0030270
3. Pardanani A, Levine R, Lasho T, et al: *MPL*515 mutations in myeloproliferative and other myeloid disorders: a study of 1182 patients. Blood. 2006;108:3472-3476. doi:10.1182/blood-2006-04-018879
4. Kilpivaara O, Levine RL: *JAK2* and *MPL* mutations in myeloproliferative neoplasms: discovery and science. Leukemia. 2008;22:1813-1817. doi:10.1038/leu.2008.229

Performance

Method Description

Polymerase chain reaction amplification of *MPL* exon 10 is performed on DNA isolated from the patient sample. The entire exon 10 sequence is obtained using Sanger sequencing with analysis on an automated genetic analyzer.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

7 to 10 days

Specimen Retention Time

DNA: 3 months

Performing Laboratory Location

Jacksonville

Fees & Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

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Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

81339

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MPLJM	MPL Exon 10 Mutation Detection, BM	75033-1

Result ID	Test Result Name	Result LOINC® Value
614541	Final Diagnosis	22637-3