Test Definition: MPLJM

MPL Exon 10 Variant Detection, Reflex, Bone Marrow (Non-Orderable)

Overview

Useful For

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

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

Evaluating for variants in *MPL* in an algorithmic process

Method Name

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Sanger Sequencing

NY State Available

No

Specimen

Specimen Type

Bone Marrow

Specimen Required

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Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: None
Specimen Volume: 3 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.
Note: Extracted DNA from bone marrow is not acceptable.

Specimen Minimum Volume

1 mL

Reject Due To

Gross hemolysis	Reject
Moderately to severely clotted	Reject
Extracted DNA from outside laboratory	Reject

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

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An interpretive report will be provided.

Interpretation

The interpretive report includes an overview of the findings.

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. Rumi E, Pietra D, Ferretti V, et al. JAK2 or CALR mutation status defines subtypes of essential thrombocythemia with substantially different clinical course and outcomes. Blood. 2014;123(10):1544-1551
3. Greenfield G, McMullin MF, Mils K. Molecular pathogenesis of the myeloproliferative neoplasms. J Hematol Oncol. 2021;14(1):103
4. Khoury JD, Solary E, Abla O, et al. The 5th edition of the World Health Organization classification of haematolymphoid tumors: myeloid and histiocytic/dendritic neoplasms. Leukemia 2022; 36:1703-1719

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

2 to 10 days

Specimen Retention Time

Bone marrow: 2 weeks; Extracted DNA: 1 year

Performing Laboratory Location

Mayo Clinic Jacksonville Clinical Lab

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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.

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