



# 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:

**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

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

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, Mills 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

**Fees & Codes**

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**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