

Overview

Useful For

Aiding in the distinction between a reactive cytosis and a myeloproliferative neoplasm when *JAK2V617F* testing result is negative

Evaluates for variants in *MPL* in an algorithmic process for MPNCM / Myeloproliferative Neoplasm, *CALR* with Reflex to *MPL*, Varies.

Method Name

Only orderable as a reflex. For more information see MPNCM / Myeloproliferative Neoplasm, *CALR* with Reflex to *MPL*, Varies.

Sanger Sequencing

NY State Available

Yes

Specimen

Specimen Type

Varies

Specimen Required

Only orderable as a reflex. For more information see MPNCM / Myeloproliferative Neoplasm, *CALR* with Reflex to *MPL*, Varies.

Submit only 1 of the following specimens:

**Specimen Type:** Blood

**Container/Tube:** Lavender top (EDTA) or yellow top (ACD solution B)

**Specimen Volume:** 4 mL

**Collection Instructions:**

1. Invert several times to mix blood.
2. Send specimen in original tube.
3. Label specimen as blood.

**Specimen Stability Information:** Ambient (preferred) 7 days/Refrigerate 7 days

**Specimen Type:** Bone marrow

**Container/Tube:** Lavender top (EDTA) or yellow top (ACD solution B)

**Specimen Volume:** 2 mL

Collection Instructions:

1. Invert several times to mix bone marrow.
2. Send specimen in original tube.
3. Label specimen as bone marrow.

Specimen Stability Information: Ambient (preferred) 7 days/Refrigerate 7 days

Specimen Type: Extracted DNA from blood or bone marrow

Container/Tube: 1.5- to 2-mL tube

Specimen Volume: Entire specimen

Collection Instructions: Label specimen as extracted DNA from blood or bone marrow and provide indication of volume and concentration of the DNA.

Specimen Stability Information: Frozen (preferred)/Refrigerate/Ambient

Specimen Minimum Volume

Blood/bone marrow: 0.5 mL

Reject Due To

Gross hemolysis	Reject
Paraffin-embedded bone marrow aspirate clot or biopsy blocks Slides Paraffin shavings Moderately to severely clotted	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical & Interpretive

Clinical Information

*JAK2* V617F variant is present in 95% to 98% of polycythemia vera (PV), and 50% to 60% of primary myelofibrosis (PMF) and essential thrombocythemia (ET). Detection of the *JAK2* V617F is useful to help establish the diagnosis of a myeloproliferative neoplasm (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 alterations (20%-30% of PMF and ET) and *MPL* exon 10 alterations (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 MPNCM / Myeloproliferative Neoplasm, *CALR* with Reflex to *MPL*, Varies.

An interpretive report will be provided.

### Interpretation

The results will be reported as 1 of the 3 following states:

- Positive for *CALR* variant
- Positive for *MPL* variant
- Negative for *CALR* and *MPL* variants

Positive variant status is highly suggestive of a myeloid neoplasm and clinicopathologic correlation is necessary in all cases.

Negative variant status does not exclude the presence of a myeloproliferative neoplasm or other neoplasms.

### 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.

### Clinical Reference

1. Klampfl T, Gisslinger H, Harutyunyan AS, et al: Somatic mutation of calreticulin in myeloproliferative neoplasms. *N Engl J Med* 2013;369:2379-2390
2. Nangalia J, Massie CE, Baxter EJ, et al: Somatic *CALR* mutation in myeloproliferative neoplasms with nonmutated *JAK2*. *N Engl J Med* 2013;369:2391-2405
3. Rotunno G, Mannarelli C, Guglielmelli P, et al: Impact of calreticulin mutations on clinical and hematological phenotype and outcome in essential thrombocythemia. *Blood* 2014;123:1552-1555
4. Tefferi A, Lasho TL, Finke CM, et al: *CALR* vs *JAK2* vs *MPL*-mutated or triple-negative myelofibrosis: clinical, cytogenetic and molecular comparisons. *Leukemia advance online publication* 21 January 2014
5. Pikman Y, Lee BH, Mercher T, et al: *MPLW515L* is a novel somatic activating mutation in myelofibrosis with myeloid metaplasia. *FLoS Med* 2006;3:e270
6. Pardanani A, Levine R, Lasho T, et al: *MPL515* mutations in myeloproliferative and other myeloid disorders: a study of 1182 patients. *Blood* 2006;15:3472

### Performance

### Method Description

Genomic DNA was extracted and Sanger sequencing used to evaluate for alterations in *MPL*, exon 10. The sensitivity of this assay is approximately 20%, such that samples containing lower percentages of mutated DNA will appear negative.(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**

Rochester

**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-MPL (myeloproliferative leukemia virus oncogene, thrombopoietin receptor, TPOR) (eg, myeloproliferative disorder), exon 10 sequence