MAYO CLINIC LABORATORIES

Test Definition: MPLR

MPL Exon 10 Mutation Detection, Reflex, Varies

Overview

Useful For

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

Evaluates for mutations in *MPL* in an algorithmic process for the MPNR / Myeloproliverative Neoplasm (MPN), *JAK2* V617F with reflex to *CALR* and *MPL*.

Method Name

Only orderable as a reflex. For more information see MPNR / Myeloproliferative Neoplasm (MPN), *JAK2* V617F with Reflex to *CALR* and *MPL.*

NY State Available

Yes

Specimen

Specimen Type

Varies

Specimen Required

Only orderable as a reflex. For more information see MPNR / Myeloproliferative Neoplasm (MPN), *JAK2* V617F with reflex to *CALR* and *MPL*.

Specimen Minimum Volume

Blood and Bone marrow: 0.5 mL

Reject Due To

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

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies	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 mutation in *JAK2* characterized as c.G1849T; p. Val617Phe (V617F). The *JAK2* V617F is present in 95% to 98% of polycythemia vera (PV), and 50% to 60% of primary myelofibrosis (PMF) and essential thrombocythemia (ET). It has also been described infrequently in other myeloid neoplasms, including chronic myelomonocytic leukemia and myelodysplastic syndrome. Detection of the *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 mutation (20%-30% of PMF and ET) and MPL exon 10 mutation (5%-10% of PMF and 3%-5% of ET). Mutations in *JAK2*, *CALR*, and *MPL* are essentially mutually exclusive. A *CALR* mutation 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 MPNR / Myeloproliferative Neoplasm (MPN), *JAK2* V617F with reflex to *CALR* and *MPL*.

An interpretive report will be provided.

Interpretation

An interpretation will be provided under the MPNR / Myeloproliferative Neoplasm (MPN), *JAK2* V617F with reflex to *CALR* and *MPL*.

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.

In rare cases, a mutation other than the V617F may be present in an area that interferes with primer or probe binding and cause a false-negative result.

Performance

Method Description

Genomic DNA is extracted from bone marrow and the *MPL* exon 10 amplified using standard PCR. The entire exon 10

sequence is obtained using Sanger sequencing (BigDye terminator V1.1 cycle sequencing kid from Applied Bioscience) with analysis on an automated genetic analyzer.(Applied Biosystems 3130; Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
7 to 10 days

Specimen Retention Time
DNA stored for 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. This test 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

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MPLR	MPL Exon 10 Mutation Detection, R	62948-5

Result ID	Test Result Name	Result LOINC® Value
36672	Final Diagnosis	22637-3