

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 / Myeloproliferative Neoplasm, JAK2 V617F with Reflex to CALR and MPL, Varies

Method Name

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

Sanger Sequencing

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*.

Reject Due To

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



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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 variant is present in 95% to 98% of patients with polycythemia vera, 50% to 60% of patients with primary myelofibrosis (PMF), and 50% to 60% of patients with 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 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 patients with PMF. 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, *JAK2* V617F with Reflex to *CALR* and *MPL*, Varies.

An interpretive report will be provided.

Interpretation

An interpretation will be provided under the MPNR / Myeloproliferative Neoplasm, *JAK2* V617F with Reflex to *CALR* and *MPL*, Varies

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.



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In rare cases, a variant 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 MPL exon 10 amplified using standard polymerase chain reaction. The entire exon 10 sequence is obtained using Sanger sequencing (BigDye terminator V1.1 cycle sequencing kit from Applied Bioscience) 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

Whole blood, bone marrow: 2 weeks; Extracted DNA: 3 months

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

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

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

LOINC® Information



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Varies

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