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 (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

<table>
<thead>
<tr>
<th>Gross hemolysis</th>
<th>Reject</th>
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<tbody>
<tr>
<td></td>
<td>Paraffin embedded bone marrow aspirate clot or biopsy blocks Slides Paraffin shavings Moderately to severely clotted</td>
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</table>

Specimen Stability Information

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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<tbody>
<tr>
<td>Varies</td>
<td>Varies</td>
<td>7 days</td>
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Clinical and 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) and Time(s) Test Performed

Monday through Friday

Analytic Time

7 days

Maximum Laboratory Time

10 days

Specimen Retention Time

DNA stored for 3 months
Performing Laboratory Location
Rochester

Fees and 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 Regional Manager. 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 U.S. 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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<th>Order LOINC Value</th>
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