Overview

Useful For
 Detecting a neoplastic clone associated with the common chromosome abnormalities seen in patients with acute leukemia or other myeloid malignancies

Tracking known chromosome abnormalities and response to therapy in patients with myeloid malignancies

Reflex Tests

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<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
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Testing Algorithm

This test includes a charge for application of the first probe set (2 FISH probes) and professional interpretation of results.

Additional charges will be incurred for all reflex probes performed. Analysis charges will be incurred based on the number of cells analyzed per probe set. If no cells are available for analysis, no analysis charges will be incurred.

This assay includes testing for the following abnormalities using the probes listed:

- **ABL2** (1q25) rearrangement
- **FIP1L1/CHIC2/PDGFRα** (4q12) rearrangement (**CHIC2** deletion)
- **PDGFRβ** (5q33) rearrangement
- **ABL1** (9q34) rearrangement

If an **ABL1** rearrangement is identified, reflex testing will be performed using the **BCR/ABL1** dual-color, double fusion FISH probe set to evaluate for the presence or absence of **BCR/ABL1** fusion.

If the patient is being tracked for known abnormalities, indicate which probes should be used.

Method Name

Fluorescence In Situ Hybridization (FISH)
NY State Available
Yes

Specimen

Specimen Type
Varies

Shipping Instructions
Advise Express Mail or equivalent if not on courier service.

Necessary Information
Provide a reason for referral with each specimen. The laboratory will not reject testing if this information is not provided, but appropriate testing and interpretation may be compromised or delayed.

Specimen Required
Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube: Green top (sodium heparin)
Specimen Volume: 7-10 mL

Collection Instructions:
1. Invert several times to mix blood.
2. Other anticoagulants are not recommended and are harmful to the viability of the cells.

Specimen Type: Bone marrow
Container/Tube: Green top (sodium heparin)
Specimen Volume: 1-2 mL

Collection Instructions:
1. Invert several times to mix bone marrow.
2. Other anticoagulants are not recommended and are harmful to the viability of the cells.

Forms
If not ordering electronically, complete, print, and send a Hematopathology/Cytogenetics Test Request (T726) with the specimen.

Specimen Minimum Volume
Blood: 2 mL
Bone Marrow: 1 mL
Reject Due To
All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

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<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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Clinical and Interpretive

Clinical Information
Myeloid neoplasms are primary disorders of the bone marrow cells. These malignancies encompass several entities with extremely varied clinical courses, including acute myeloid leukemias (AML), chronic myeloproliferative disorders (CMPD), and myelodysplastic syndromes. The underlying genetic mechanisms associated with these malignancies are varied and only a portion of the genetic abnormalities have targeted therapies clinically available.

One group of genes, including ABL1 (Abelson murine leukemia viral oncogene homolog 1), ABL2 (Abelson murine leukemia viral oncogene homolog 2), PDGFRα (platelet-derived growth factor receptor, alpha), and PDGFRβ (platelet-derived growth factor receptor, beta) can be inappropriately activated via various genetic mechanisms and result in overexpression of their tyrosine kinase activity. Tyrosine kinase activity plays an important role in cellular signaling, division, and differentiation; overexpression may cause some cancers. The myeloid malignancies associated with these aberrantly expressed genes include AML, chronic myelogenous leukemia (CML), hypereosinophilic syndrome/systemic mast cell disease (HES/SMCD), and atypical CMPD. These translocations can also be seen in lymphoid neoplasms, including acute lymphoblastic leukemia (ALL) and lymphomas, and they can also possess a varied genetic etiology. Several clinical studies have demonstrated that the malignancies displaying overexpression of these genes are responsive to imatinib mesylate, a drug that specifically targets these genes.

Reference Values
An interpretive report will be provided.

Interpretation
A neoplastic clone is detected when the percent of cells with an abnormality exceeds the normal cutoff for any given probe.

The presence of a positive clone supports a diagnosis of malignancy.

The absence of an abnormal clone does not rule out the presence of neoplastic disorder.

Cautions
This test is not approved by the U.S. Food and Drug Administration and it is best used as an adjunct to existing clinical and pathologic information.

Supportive Data
Each probe was independently tested and verified on unstimulated peripheral blood and bone marrow specimens.
Normal cutoffs were calculated based on the results of at least 20 normal specimens. For each probe set, a series of chromosomally abnormal specimens were evaluated to confirm that each probe set detected the abnormality it was designed to detect.

**Clinical Reference**


**Performance**

**Method Description**

This test is performed using commercially available and laboratory-developed probes. Rearrangements involving ABL2, ABL1, or PDGFRB are detected using dual-color break-apart (BAP) strategy probes. FIP1L1/PRGFR fusion (with loss of the CHIC2 locus), is detected using a tricolor rearrangement probe strategy. For each probe set, 2 technologists each analyze 100 interphase nuclei (200 total) with the results expressed as the percent abnormal nuclei. (Unpublished Mayo method)

**PDF Report**

No

**Day(s) and Time(s) Test Performed**

Samples processed Monday through Sunday. Results reported Monday through Friday, 8 a.m.-5 p.m. CST.

**Analytic Time**

7 days

**Maximum Laboratory Time**

10 days

**Specimen Retention Time**

4 weeks

**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.
Test Definition: IMRGF
Imatinib Mesylate Resp Genes, FISH

- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification
This test was developed using an analyte specific reagent. Its performance characteristics were 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
88271 x 2, 88291-DNA probe, each (first probe set), Interpretation and report

88271 x 2-DNA probe, each; each additional probe set (if appropriate)
88271 x 1-DNA probe, each; coverage for sets containing 3 probes (if appropriate)
88271 x 2-DNA probe, each; coverage for sets containing 4 probes (if appropriate)
88271 x 3-DNA probe, each; coverage for sets containing 5 probes (if appropriate)
88274 w/modifier 52-Interphase in situ hybridization, <25 cells, each probe set (if appropriate)
88274-Interphase in situ hybridization, 25 to 99 cells, each probe set (if appropriate)
88275-Interphase in situ hybridization, 100 to 300 cells, each probe set (if appropriate)

LOINC® Information

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