Overview

Useful For

Detecting a neoplastic clone associated with the common chromosome abnormalities seen in patients with myelodysplastic syndromes or other myeloid malignancies

Evaluating specimens in which standard cytogenetic analysis is unsuccessful

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

Reflex Tests

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
</tr>
</thead>
<tbody>
<tr>
<td>_I099</td>
<td>Interphases, 25-99</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_I300</td>
<td>Interphases, &gt;=100</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_IL25</td>
<td>Interphases,</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_PADD</td>
<td>Probe, +1</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_PB02</td>
<td>Probe, +2</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_PB03</td>
<td>Probe, +3</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>_PBCT</td>
<td>Probe, +2</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
</tbody>
</table>

Testing Algorithm

Chromosome analysis is recommended as first-tier testing for myelodysplastic syndrome; FISH analysis should only be ordered if chromosome analysis is not successful. If this test is ordered concurrently with a chromosomal study (CHRBM or CHRHB), testing will be held pending the results of the chromosome test. If the chromosome results are complete and informative, this test will be canceled.

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.

See Myelodysplastic Syndrome: Guideline to Diagnosis and Follow-up in Special Instructions.

Indicate if the entire panel is to be performed or if only an individual probe set (ie, -7/7q-) is desired. If the patient is being tracked for known clonal abnormalities, indicate which probes should be analyzed.

Panel includes testing for the following abnormalities using the probes listed:

-5/5q-, D5S630/EGR1

-7/7q-, D7S486/D7Z1

+8, D8Z2/MYC

11q23 rearrangement, MLL(KMT2A)
17p-, TP53/D17Z1

20q-, D20S108/20qter

inv(3) or t(3;3), RPN1/MECOM

-When an MLL (KMT2A) rearrangement is identified, reflex testing will be performed to identify the translocation partner. Probes include identification of t(4;11)(q21;q23) AFF1/MLL, t(6;11)(q27;q23) MLTT4/MLL, t(9;11)(p22;q23) MLLT3/MLL, t(11;16)(q23;p13.3) MLL/CREBBP, t(11;19)(q23;p13.1) MLL/ELL, or t(11;19)(q23;p13.3) MLL/MLLT1.

-When 3 copies of MECOM are observed with no fusion with RPN1, reflex testing using the MECOM/RUNX1 probe set will be performed to identify a potential t(3;21)(q26.2;q22) rearrangement.

-When 3 copies of RPN1 are observed with no fusion with MECOM, reflex testing using the PRDM16/RPN1 probe set will be performed to identify a potential t(1;3)(p36;q21).

Special Instructions

- Myelodysplastic Syndrome: Guideline to Diagnosis and Follow-up

Method Name
Fluorescence In Situ Hybridization (FISH)

NY State Available
Yes

Specimen

Specimen Type
Varies

Advisory Information

If this test is ordered in conjunction with AMLF / Acute Myeloid Leukemia (AML), FISH, it will be canceled as a duplicate test request because these same probes are performed as part of the AMLF panel.

Chromosome analysis is recommended as first-tier testing; see CHRBM / Chromosome Analysis, Hematologic Disorders, Bone Marrow, or CHRHB / Chromosomes, Hematologic Disorders, Blood. This second-tier test should only be ordered if chromosome analysis is not successful, as it does not increase the sensitivity for detection of myelodysplastic syndrome (MDS) for classic abnormalities (ie, -5/5q-, -7/7q-). If this test is ordered concurrently with a chromosomal study (CHRBM or CHRHB), testing will be held pending the results of the chromosome test. If the chromosome results are complete and informative, this test will be canceled. This assay can be helpful if a complete chromosome study is not achieved (<20 metaphases), 1 abnormal metaphase (nonclonal) with a classic abnormality is observed, or an unresolved structural abnormality is observed.

Shipping Instructions

Advise Express Mail or equivalent if not on courier service.

Necessary Information

1. Provide a reason for referral with each specimen and bone marrow pathology report (if available). The laboratory will not reject testing if this information is not provided, but appropriate testing and interpretation may be compromised or delayed.
2. A pathology and/or flow cytometry report may be requested by the Genomics Laboratory to optimize testing and aid in interpretation of results.

**Specimen Required**
Submit only 1 of the following specimens:

**Preferred 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.

**Acceptable Specimen Type:** 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.

**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**

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
</tr>
</thead>
<tbody>
<tr>
<td>Varies</td>
<td>Ambient (preferred)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Refrigerated</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Clinical and Interpretive**
**Clinical Information**

Myelodysplastic syndromes (MDS) primarily occur in the older adult population and have a yearly incidence of 30 in 100,000 in persons older than 70 years of age. These disorders are typically associated with a hypercellular bone marrow and low peripheral blood counts, and with significant morbidity and mortality. The eventual clinical outcome for patients with MDS relates to either bone marrow failure or transformation to acute myeloid leukemia. MDS can be either primary (de novo) or secondary (due to previous treatment with alkylating or etoposide chemotherapy, with or without radiation).

Cytogenetic studies can provide confirmatory evidence of clonality in MDS and can be used to provide clinical prognostic or diagnostic information. Clonal cytogenetic abnormalities are more frequently observed in cases of secondary MDS (80% of patients) than in primary MDS (40%-60% of patients). The common chromosomal abnormalities associated with MDS include: inv(3), -5/5q-, -7/7q-, +8, and 20q-. These abnormalities can be observed singly or in concert. In addition, MLL (KMT2A) rearrangements, t(1;3), and t(3;21) are more frequently associated with secondary MDS.

Conventional chromosome analysis is the gold standard for identification of the common, recurrent chromosome abnormalities in MDS; however, some of the subtle rearrangements associated with secondary MDS can be missed (eg, MLL abnormalities).

**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 reference range for any given probe.

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

**Cautions**

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

Bone marrow is the preferred specimen type for this FISH test. If bone marrow is not available, a blood specimen may be used if there are malignant cells in the blood specimen (as verified by hematopathology).

**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 25 normal specimens. For each probe set a series of chromosomally abnormal specimens were evaluated to confirm each probe set detected the abnormality it was designed to detect.

**Clinical Reference**


3. He R, Wiktor AE, Durnick DK, et al: Bone Marrow Conventional Karyotyping and Fluorescence In Situ

**Performance**

**Method Description**

This test is performed using commercially available and laboratory-developed probes. Deletion or monosomy of chromosomes 5, 7, trisomy of chromosome 8, and deletion or rearrangement of chromosomes 17 and 20 are detected using enumeration strategy probes. Rearrangements involving MLL (KMT2A) are detected using a dual-color break-apart (BAP) strategy probe. Dual-color, dual-fusion (D-FISH) strategy probe sets are used to detect inv(3), t(3;21), and in reflex testing when rearrangements of the MLL gene are detected. For the enumeration and BAP strategy probe sets, 200 interphase nuclei are scored; 500 interphase nuclei are scored when D-FISH probes are used. Two technologists analyze each probe set and all results are 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. to 5 p.m.

**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.
- 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-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**

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>MDSF</td>
<td>MDS, FISH</td>
<td>In Process</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Result ID</th>
<th>Test Result Name</th>
<th>Result LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>51912</td>
<td>Result Summary</td>
<td>50397-9</td>
</tr>
<tr>
<td>51914</td>
<td>Interpretation</td>
<td>69965-2</td>
</tr>
<tr>
<td>51913</td>
<td>Result Table</td>
<td>93356-4</td>
</tr>
<tr>
<td>54548</td>
<td>Result</td>
<td>62356-1</td>
</tr>
<tr>
<td>CG688</td>
<td>Reason for Referral</td>
<td>42349-1</td>
</tr>
<tr>
<td>CG689</td>
<td>Specimen</td>
<td>31208-2</td>
</tr>
<tr>
<td>51915</td>
<td>Source</td>
<td>31208-2</td>
</tr>
<tr>
<td>51916</td>
<td>Method</td>
<td>49549-9</td>
</tr>
<tr>
<td>55115</td>
<td>Additional Information</td>
<td>48767-8</td>
</tr>
<tr>
<td>53865</td>
<td>Disclaimer</td>
<td>62364-5</td>
</tr>
<tr>
<td>51917</td>
<td>Released By</td>
<td>18771-6</td>
</tr>
</tbody>
</table>