MAYO CLINIC LABORATORIES

Test Definition: MSTF

Myeloid Sarcoma, FISH, Tissue

Overview

Useful For

Supporting the diagnosis of myeloid sarcoma when coordinated with a surgical pathology consultation

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_I099	Interphases, 25-99	No, (Bill Only)	No
_I300	Interphases, >=100	No, (Bill Only)	No
_IL25	Interphases, <25	No, (Bill Only)	No
_PADD	Probe, +1	No, (Bill Only)	No
_PB02	Probe, +2	No, (Bill Only)	No
_PB03	Probe, +3	No, (Bill Only)	No
_PBCT	Probe, +2	No, (Bill Only)	No

Testing Algorithm

This test does not include a pathology consult. If a pathology consultation is requested, PATHC / Pathology Consultation should be ordered, and the appropriate fluorescence in situ hybridization (FISH) test will be performed at an additional charge.

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.

The test panel includes analysis for the disease-associated abnormalities using the probes listed below:
t(8;21), [M2], *RUNX1T1/RUNX1*
t(11q23;var), [M0-M7], *MLL* (*KMT2A*)
inv(16), [M4, Eos], *MYH11/CBFB*
t(15;17), [M3], *PML/RARA*
t(9;22), *BCR/ABL1*

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

Appropriate ancillary probes may be performed at consultant discretion to render comprehensive assessment. Any additional probes will have the results included within the final report and will be performed at an additional charge.

Method Name

Fluorescence In Situ Hybridization (FISH)

NY State Available

Yes

Specimen

Specimen Type
Tissue

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

Necessary Information
A reason for referral and pathology report are required in order for testing to be performed. Send information with specimen. Acceptable pathology reports include working drafts, preliminary pathology or surgical pathology reports.

Specimen Required
Specimen Type: Tissue
Preferred: Tissue block
Collection Instructions: Submit a formalin-fixed, paraffin-embedded tumor tissue block. Blocks prepared with alternative fixation methods may be acceptable; provide fixation method used.
Acceptable: Slides
Collection Instructions: For each probe set ordered, 2 consecutive, unstained, 5 micron-thick sections placed on positively charged slides, and 1 hematoxylin and eosin-stained slide.

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

Specimen Minimum Volume
For each probe set ordered, 2 consecutive, unstained, 5 micron-thick sections placed on positively charged slides.
Include 1 hematoxylin and eosin (H and E)-stained slide.

Reject Due To
All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Tissue	Ambient (preferred)		
	Refrigerated		

Clinical & Interpretive

Clinical Information
Myeloid sarcomas are tumors made up of myeloblasts or immature myeloid cells that occur in extramedullary sites or in bone. They can occur concurrently with acute or chronic myeloid leukemia (AML or CML) or may precede the leukemia

or other myeloid neoplasms. They may also be the initial manifestation of relapse of a previously treated primary AML in remission. Due to this extramedullary presentation, the bone marrow may have a low number of myeloblasts due to a lack of bone marrow involvement.

The most common abnormalities seen in myeloid sarcomas are fusion of *RUNX1T1/RUNX1* (t[8;21][q22;q22]), *PML/RARA* (t[15;17][q24;q21]), *BCR/ABL1* (t[9;22][q34;q11.2]), inversion of *MYH11/CBFB* (inv[16][q13.1q22]), and rearrangements of *MLL* (*KMT2A*; t[11q23;var]).

In general, AML patients with an inv(16), t(8;21), t(9;22), or t(15;17) have a favorable prognosis, while AML patients with a rearrangement of t(11q23) have an unfavorable prognosis. Thus, the detection of these abnormalities in an extramedullary presentation of AML can be prognostically important.

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 a given probe set.

A positive result supports the diagnosis of a myeloid sarcoma.

A negative result does not exclude the diagnosis of a myeloid sarcoma.

Cautions

Paraffin-embedded tissues that have been decalcified may not be successful for fluorescence in situ hybridization (FISH) analysis. FISH studies will be attempted if sufficient tumor is present for analysis. However, if no FISH signals are observed post-hybridization, the case will be released indicating a lack of FISH results.

Supportive Data

Fluorescence in situ hybridization (FISH) analysis was performed on 25 noncancerous formalin-fixed paraffin-embedded tissue control specimens with the results used to generate the normal cutoff value for each probe set. A retrospective data review of FISH analysis performed on myeloid sarcomas identified 3 cases with *RUNX1T1/RUNX1* fusion, 1 case with *BCR/ABL1* fusion, 3 cases with rearrangement of *MLL* (*KMT2A*), 4 cases with *PML/RARA* fusion, and 4 cases with *MYH11/CBFB* fusion.

Clinical Reference

1. Slovak ML, Kopecky KJ, Cassileth PA, et al: Karyotypic analysis predicts outcome of preremission and postremission therapy in adult acute myeloid leukemia: a Southwest Oncology Group/Eastern Cooperative Oncology Group Study. Blood. 2000;96:4075-4083

2. Swerdlow SH, Campo E, Harris NL, et al: WHO Classification of Tumors of Haematopoietic and Lymphoid Tissues. International Agency for Research on Cancer; 2008:140-141

3. Grimwade D, Hills RK, Moorman AV, et al: Refinement of cytogenetic classification in acute myeloid leukemia: determination of prognostic significance of rare recurring chromosomal abnormalities among 5876 younger adult patients treated in the United Kingdom Medical Research Council trials. Blood. 2010;116:354-365

Performance

Method Description

This test is performed using a commercially available *MLL* (*KMT2A*) dual-color break-apart strategy probe (BAP), a laboratory developed *MYH11/CBFB* dual-color, dual-fusion (D-FISH) strategy probe, and commercially available *RUNX1T1/RUNX1, BCR/ABL1*, and *PML/RARA* D-FISH strategy probes. Formalin-fixed paraffin-embedded tissues are cut at 5 microns and mounted on positively charged glass slides. The selection of tissue and the identification of target areas on the hematoxylin and eosin (H and E)-stained slide are performed by a pathologist. Using the H and E-stained slide as a reference, target areas are etched with a diamond-tipped etcher on the back of the unstained slide to be assayed. The probe is hybridized to the appropriate target area and 2 technologists each analyze 50 interphase nuclei (100 total for each probe set) with the results expressed as the percent of abnormal nuclei.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

7 to 10 days

Specimen Retention Time

Slides and H and E used for analysis are retained by the laboratory in accordance to CAP and NYS requirements. Client provided paraffin blocks and extra unstained slides (if provided) will be returned after testing is complete.

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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 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 US Food and Drug Administration.

CPT Code Information

88291
88271 x 2 (if appropriate)

88271 x 2 (if appropriate)
88271 (if appropriate)
88271 x 2 (if appropriate)
88271 x 3 (if appropriate)
88274 w/modifier 52 (if appropriate)
88274 (if appropriate)
88275 (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MSTF	Myeloid Sarcoma, FISH, Ts	In Process

Result ID	Test Result Name	Result LOINC® Value
52084	Result Summary	50397-9
52086	Interpretation	69965-2
52085	Result Table	93356-4
54576	Result	62356-1
CG735	Reason for Referral	42349-1
CG736	Specimen	31208-2
52087	Source	31208-2
52088	Tissue ID	80398-1
52089	Method	85069-3
52090	Released By	18771-6
55121	Additional Information	48767-8
53839	Disclaimer	62364-5