

Overview

Useful For

Diagnosing and classifying acute myeloid leukemia using bone marrow specimens

Providing guidance for clinical management of patients

Confirming a gene fusion detected by next-generation sequencing

Tracking response to therapy

Reflex Tests

| Test Id | Reporting Name | Available Separately | Always Performed |
|---------|-------------------------------|----------------------|------------------|
| JAMMP | Probe, Each Additional (JAML) | No, (Bill Only) | No |

Testing Algorithm

A charge and CPT code is applied for each probe set hybridized, analyzed and reported.

The following fluorescence in situ hybridization (FISH) probes are orderable individually or as a set:

Dual color dual fusion probes for *PML::RARA* fusion

Dual color dual fusion probes for *RUNX1T1::RUNX1* fusion

Dual color break-apart probes for *CBFB::MYH11* fusion

Dual color break-apart probes for *KMT2A* rearrangement

Dual color dual fusion probes for *DEK::NUP214* fusion

Tri- color dual fusion probes for *BCR::ABL1* fusion

Dual color break-apart probes for *MECOM* rearrangement

Method Name

Fluorescence In Situ Hybridization (FISH)

NY State Available

No

Specimen

Specimen Type

Bone Marrow

Shipping Instructions

Specimen must arrive within 7 days of collection.

Necessary Information

The following information is required:

1. Pertinent clinical history
2. Clinical or morphologic suspicion
3. Date of collection
4. Specimen source

Specimen Required

Container/Tube: Green top (sodium heparin) or yellow top (ACD solution B)

Specimen Volume: 2 mL

Collection Instructions:

1. Invert several times to mix bone marrow.
2. Send bone marrow specimen in original tube. **Do not** aliquot.
3. Label specimen as bone marrow.

Specimen Minimum Volume

1 mL

Reject Due To

| | |
|--------------------------------|--------|
| Gross hemolysis | Reject |
| Moderately to severely clotted | Reject |

Specimen Stability Information

| Specimen Type | Temperature | Time | Special Container |
|---------------|---------------------|--------|-------------------|
| Bone Marrow | Ambient (preferred) | 7 days | |
| | Refrigerated | 7 days | |

Clinical & Interpretive**Clinical Information**

Acute myeloid leukemia (AML) has been defined by genetic abnormalities and differentiation in the 5th edition of World Health Organization classification of hematolymphoid tumors.(1)

The subtypes of AML defined by genetic abnormalities include:

Acute promyelocytic leukemia with *PML::RARA* fusion

AML with *RUNX1::RUNXT1* fusion
AML with *CBFB::MYH11* fusion
AML with *DEK::NUP214* fusion
AML with *RBM15::MRTFA* fusion
AML with *BCR::ABL1* fusion
AML with *KMT2A* rearrangement
AML with *MECOM* rearrangement
AML with *NUP98* rearrangement
AML with *NPM1* mutation
AML with *CEBPA* mutation
AML myelodysplasia-related
AML with other defined genetic alterations

Fluorescence in situ hybridization (FISH) testing detects specific gene fusions associated with AML.

Fluorescence in situ hybridization testing will not detect gene variants associated with AML.

RBM15::MRTFA fusion and *NUP98* rearrangement will not be detected in this FISH test. These two abnormalities are rare and can be detected by next-generation sequencing. FISH testing for these two abnormalities may be added to this test at later date.

Reference Values

An interpretive report will be provided.

Interpretation

Detection of a specific fusion or a rearrangement confirms a clinical diagnosis of acute myeloid leukemia (AML) and defines an AML classification. Absence of a specific gene fusion or rearrangement will not rule out presence of AML.

Cautions

Fluorescence in situ hybridization (FISH) results should be correlated with clinical and pathologic information for diagnosis and treatment.

Fluorescence in situ hybridization testing is not a substitute for conventional chromosome studies because the latter detects many other chromosome abnormalities associated with acute myeloid leukemia and other hematological disorders.

Bone marrow is the preferred specimen type. If bone marrow is not available, a blood specimen may be used if there are malignant cells in the blood specimen (as verified by a hematopathologist); see JAMLB / Acute Myeloid Leukemia (AML), FISH, Blood.

Supportive Data

Each probe was independently tested and verified on unstimulated peripheral blood specimens. Normal cutoffs were calculated based on the results of 22 normal specimens. Each probe set was evaluated to confirm the probe set detected the abnormality it was designed to detect.

Clinical Reference

1. Khouri JD, Solary E, Abla O et al. The 5th edition of the World Health Organization Classification of Haematolymphoid Tumours: Myeloid and histiocytic/dendritic neoplasms. *Leukemia*. 2022;36(7):1703-1719.
doi:10.1038/s41375-022-01613-1
2. Dohner H, Estey E, Grimwade D, et al. Diagnosis and management of AML in adults: 2017 ELN recommendations from an international expert panel. *Blood*. 2017;129(4):424-447

Performance**Method Description**

Fluorescence in situ hybridization is performed using commercially available probes, including the dual/tri-color dual fusion DNA probes for *PML::RARA*, *RUNX1T1::RUNX1*, *DEK::NUP214*, and *BCR::ABL1* fusion and break-apart probes for *CBFB*, *KMT2A*, and *MECOM* rearrangement. Two-hundred interphase nuclei are analyzed for each probe set by 2 laboratory technologists, 100 per technologist. All results are interpreted as positive or negative based on the cut-off established by the validation in this lab and reported using an International System for Human Cytogenetic Nomenclature.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

2 to 8 days

Specimen Retention Time

4 weeks

Performing Laboratory Location

Mayo Clinic Jacksonville Clinical Lab

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 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

88377 (if 1 probe set)
88377 x 2 (if 2 probe sets)
88377 x 3 (if 3 probe sets)
88377 x 4 (if 4 probe sets)
88377 x 5 (if 5 probe sets)
88377 x 6 (if 6 probe sets)
88377 x 7 (if 7 probe sets)

LOINC® Information

| Test ID | Test Order Name | Order LOINC® Value |
|---------|-----------------------|--------------------|
| JAMLM | AML FISH, Bone Marrow | 102103-9 |

| Result ID | Test Result Name | Result LOINC® Value |
|-----------|------------------------|---------------------|
| 620195 | Result Summary | 50397-9 |
| 620196 | Interpretation | 59465-5 |
| 620197 | Result | 62356-1 |
| 620198 | Reason For Referral | 42349-1 |
| 620341 | Probes Requested | 62370-2 |
| 620199 | Specimen | 31208-2 |
| 620200 | Source | 31208-2 |
| 620201 | Method | 85069-3 |
| 620202 | Additional Information | 48767-8 |
| 620203 | Disclaimer | 62364-5 |
| 620204 | Released By | 18771-6 |