MAYO CLINIC LABORATORIES

Test Definition: JAMLM

Acute Myeloid Leukemia (AML), FISH, Bone Marrow

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 (JAMLM)	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.

FISH 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.

A separate FISH panel will be performed to detect the defining genetic abnormalities for myelodysplasia-related AML.

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.

FISH 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. Khoury 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*, *KTM2A*, 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

Jacksonville

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

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Acute Myeloid Leukemia (AML), FISH, Bone Marrow

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