MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: November 9, 2021
Effective Date: December 13, 2021

Acute Myeloid Leukemia (AML), FISH, Pediatric, Varies

Test ID: AMLPF

Useful for:
Detecting a neoplastic clone associated with the common chromosome abnormalities and classic rearrangements seen in pediatric/young adult patients with acute myeloid leukemia (AML)

An adjunct to conventional chromosome studies in patients with AML

Evaluating specimens in which standard cytogenetic analysis is unsuccessful

Testing Algorithm:
This test includes a charge for the probe application, analysis and professional interpretation of results for 13 probe sets (26 individual fluorescence in situ hybridization [FISH] probes). Additional charges will be incurred for all reflex or additional probe sets performed.

The diagnostic pediatric/young adult FISH panel includes testing for the following abnormalities using the FISH probes listed:

inv(16), [M4, Eos], MYH11/CBFB
t(8;21), [M2], RUNX1T1/RUNX1
t(15;17), [M3], PML/RARA
11q23 rearrangement, [M0-M7], MLL (KMT2A)
t(6;9), [M2,M4], DEK/NUP214
inv(3) or t(3;3), [M1,2,4,6,7], RPN1/MECOM
t(8;16), [M4,M5], KAT6A/CREBBP
t(1;22), [M7], RBM15/MKL1
-5/5q-, D5S630/EGR1
-7/7q-, D7Z1/ D7S486
12p13 rearrangement, ETV6
inv(16), GLIS2/CBFA2T3
11p15.4 rearrangement, NUP98

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) MLLT4(AFDN)/MLL, t(9;11)(p22;q23) MLLT3/MLL, t(10;11)(p12;q23) MLLT10/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.

In the absence of RPN1/MECOM fusion, when an extra MECOM signal is identified, reflex testing using the MECOM/RUNX1 probe set will be performed to identify a potential t(3;21)(q26.2;q22) rearrangement.

In the absence of RPN1/MECOM fusion, when an extra RPN1 signal is identified, reflex testing using the PRDM16/RPN1 probe set will be considered to identify a potential t(1;3)(p36;q21).

In the absence of MYH11/CBFB fusion, when an extra CBFB signal is identified, reflex testing will be performed using the CBFB break-apart probe set to evaluate for the presence or absence of an *CBFB* rearrangement.

In the absence of PML/RARA fusion, when an extra or atypical RARA signal is identified, testing using a break-apart RARA probe set will be performed to identify a potential variant translocation involving *RARA*; example: t(17;var)(q21;?).

When an ETV6 rearrangement is identified, reflex testing using the MNX1/ETV6 probe set will be performed to identify a potential t(7;12)(q36;p13) rearrangement.

When a NUP98 rearrangement is identified, reflex testing using the HOXA9/NUP98 probe set will be performed to identify a potential t(7;11)(p15;p15.4) rearrangement.

Reflex Tests:

Test ID	Reporting Name	Available Separately	Always Performed
AMLPB	Probe, Each Additional (AMLPF)	No (Bill Only)	No

Methods:

Fluorescence In Situ Hybridization (FISH)

Reference Values:

An interpretive report will be provided.

Specimen Requirements:

Preferred Specimen Type: Bone marrow

Preferred Container/Tube: Yellow top (ACD)

Acceptable Container/Tube: Green top (heparin) or lavender top (EDTA)

Specimen Volume: 2-3 mL

Minimum Volume: 1 mL

Collection Instructions:

1. It is preferable to send the first aspirate from the bone marrow collection.
2. Invert several times to mix bone marrow.

Acceptable Specimen Type: Blood

Preferred Container/Tube: Yellow top (ACD)

Acceptable Container/Tube: Green top (heparin) or lavender top (EDTA)

Specimen Volume: 6 mL

Minimum Volume: 2 mL

Collection Instructions:

1. Invert several times to mix blood.

Note:

1. A reason for testing and a flow cytometry and/or a bone marrow pathology report, if available, should be submitted 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. If this information is not provided, an appropriate indication for testing may be entered by Mayo Clinic Laboratories.

2. If the patient has received an opposite sex bone marrow transplant prior to specimen collection for this protocol, note this information on the request.

Specimen Stability Information:

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Refrigerated	

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.

Fluorescence in situ hybridization (FISH) is not a substitute for conventional chromosome studies because the latter detects many chromosome abnormalities associated with other hematological disorders that would be missed by this FISH panel test.

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 a hematopathologist).

CPT Code:
88271x26, 88275x13, 88291 x1-FISH Probe, Analysis, Interpretation; 13 probe sets

88271x2, 88275x1-FISH Probe, Analysis; each additional probe set (if appropriate)

Day(s) Performed: Monday through Friday **Report Available:** 7 to 10 days

Questions

Contact Joshua Couchene Laboratory Technologist Resource Coordinator at 800-533-1710.