

Patient ID SA00175526	Patient Name TESTING, AMLFP_ABNORMAL	Birth Date 2001-01-01	Sex F	Age 24
Order Number SA00175526	Client Order Number SA00175526	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 06 May 2025 14:45		

Pediatric-AML panel, FISH

Result Summary

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Abnormal

Interpretation

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The result is abnormal and indicates RUNX1::RUNX1T1 fusion in 87.5% of nuclei. This result is consistent with the t(8;21) observed in the chromosome studies (reported separately).

This 8;21 translocation has been associated with both de novo and therapy-related AML and has a favorable prognosis. Molecular studies to determine KIT mutational status should be considered in core binding factor leukemias [t(8;21), inv(16)/t(16;16)] as KIT mutations confer an intermediate-risk status (Swerdlow et al., WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues. IARC Press:Lyon, 2017; page 132; NCCN Guidelines Version 3.2021, AML).

Clinical and pathologic correlation is recommended.

For monitoring response to therapy, RT-PCR for t(8;21) RUNX1::RUNX1T1 fusion is recommended, per NCCN guidelines; Test: T821Q. Although RT-PCR testing is not available on the current specimen, this testing can be performed on blood or bone marrow collected in EDTA or ACD. Please call 800-533-1710 regarding specimen requirements for RT-PCR based tests.

Result Table

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Abnormality Name	Result	Abn% Cutoff%
t(15;17) PML::RARA fusion	Normal	<4.0
inv(16)/t(16;16) CBFB::MYH11 fusion	Normal	<4.0
11q23(KMT2A sep)	Normal	<10.0
t(8;21) RUNX1::RUNX1T1 fusion	Abnormal	87.5 <4.0
-5q31(D5S630x2,EGR1x1)	Normal	<12.0
-5(D5S630,EGR1)x1	Normal	<10.0
-7q31(D7Z1x2,D7S486x1)	Normal	<12.0
-7(D7Z1,D7S486)x1	Normal	<10.0
inv(3) GATA2::MECOM fusion	Normal	<4.0
t(6;9) DEK::NUP214 fusion	Normal	<4.0
t(7;12) MNX1::ETV6 fusion	Normal	<4.0
t(8;16)KAT6A::CREBBP fusion	Normal	<4.0
inv(16)/t(16;16)CBFA2T3::GLIS2 fusion	Normal	<4.0
11p15.4(NUP98 sep)	Normal	<25.0

Result

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nuc ish (RUNX1T1,RUNX1)x3(RUNX1T1 con RUNX1)x2[175/200]

Reason for Referral

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r/o AML

Specimen

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Bone Marrow

Source

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Left iliac crest

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292

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Method

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Locus and probes	[Strategy;#Nuclei;Vendor]
3q21(GATA2), 3q26.2(MECOM)	[DFISH;200;LDT]
5p15.2(D5S630), 5q31(EGR1)	[COPY#;100;AM]
6p22.3(DEK), 9q34(NUP214)	[DFISH;200;LDT]
7CEN(D7Z1), 7q31(D7S486)	[COPY#;100;AM]
7q36(MNX1), ETV6(12p13)	[DFISH;200;LDT]
8p11.2(KAT6A), 16p13.3(CREBBP)	[DFISH;200;LDT]
8q21.3(RUNX1T1), 21q22(RUNX1)	[DFISH;200;AM]
11p15.4(3'NUP98,5'NUP98)	[BAP;100;LDT]
11q23(5'KMT2A,3'KMT2A)	[BAP;100;AM]
15q24.1(PML), 17q21(RARA)	[DFISH;200;AM]
16p13.3(GLIS2), 16q24.3(CBFA2T3)	[DFISH;200;LDT]
16p13(MYH11), 16q22(CBFB)	[DFISH;200;LDT]

Probe strategies include:
DFISH=dual color, double fusion;
BAP=break-apart probe;
COPY#=region gain and loss.
Scoring Method: Manual

Probe vendors include:

LDT = Mayo Clinic Developed

AM = Abbott Molecular, Inc (Des Plaines, IL)

Updated Gene Symbols

GATA2: previously reported as RPN1

KMT2A: previously reported as MLL

Disclaimer

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Applicable to Analyte Specific Reagent (ASR) and Laboratory Developed Tests (LDT). 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 U.S. Food and Drug Administration. This FISH test does not rule out other chromosome abnormalities.

Released By

MCR

Xinjie Xu, Ph.D.

Received: 13 May 2025 10:48

Reported: 21 May 2025 14:08

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Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292