Riverbend Regional Medical Center

CYTOGENETICS LABORATORY — Department of Pathology & Laboratory Medicine
1200 Halsted Avenue, Lakeshore, IL 60000 | CLIA 14D9999999

CHROMOSOME ANALYSIS & FISH — HEMATOLOGIC MALIGNANCY PANEL

Patient: DOE, JONATHAN R.	**MRN:** MRN-4471902	**Accession:** CG-26-00871
DOB: 1967-09-14 (58 y / Male)	**Collected:** 2026-04-22	**Reported:** 2026-04-29
Specimen: Bone marrow aspirate	**Ordering:** A. Patel, MD	**Status:** FINAL

CLINICAL INDICATION

58-year-old man with three weeks of progressive fatigue, dyspnea on exertion, gingival bleeding, and scattered lower-extremity ecchymoses. CBC on admission: WBC 38.4 x10^9/L, hemoglobin 7.9 g/dL, platelets 31 x10^9/L. Peripheral smear with 41% circulating blasts, some with folded nuclei and abundant cytoplasm. No prior hematologic history, no prior chemotherapy or radiation. Bone marrow performed to evaluate for acute leukemia.

CONVENTIONAL CHROMOSOME ANALYSIS (G-BANDED)

Tissue cultured	Bone marrow, unstimulated
Cultures	24-hour and 48-hour, synchronized
Cells counted	20
Cells karyotyped	5
Band resolution	400-425 bphs

ISCN RESULT:

`46,XY[20]`

Twenty metaphase cells were analyzed and five were fully karyotyped. All cells showed an apparently normal male chromosome complement. No clonal numerical or structural abnormality was identified at the stated band resolution. A normal karyotype does not exclude a submicroscopic abnormality; correlation with FISH and molecular studies is advised.

FLUORESCENCE IN SITU HYBRIDIZATION (FISH)

Interphase FISH performed on cultured bone marrow using the acute myeloid leukemia panel. 200 interphase nuclei scored per probe.

Probe / target	Abnormality screened	Result	% nuclei
RUNX1-RUNX1T1	t(8;21)(q22;q22)	NEGATIVE	0.5%
CBFB	inv(16)/t(16;16)(p13;q22)	NEGATIVE	1.0%
PML-RARA	t(15;17)(q24;q21)	NEGATIVE	0.5%
KMT2A (MLL)	11q23.3 rearrangement	NEGATIVE	1.5%
MECOM (EVI1)	3q26.2 rearrangement	NEGATIVE	1.0%
-5/5q- (EGR1)	deletion 5q31	NEGATIVE	1.5%
-7/7q- (D7S486)	monosomy 7 / del(7q)	NEGATIVE	1.0%
TP53 (17p13.1)	deletion 17p	NEGATIVE	2.0%

All probe signal counts were within the laboratory's established normal cutoff range. No evidence of the targeted rearrangements or copy-number abnormalities.

INTERPRETATION

Normal male karyotype (46,XY) with a negative AML FISH panel. No recurrent cytogenetic abnormality of acute myeloid leukemia was detected by either method. This places the case in the cytogenetically normal category. Cytogenetically normal AML is frequently driven by gene-level mutations that are not visible by karyotype or FISH; molecular testing (NPM1, FLT3, CEBPA, and a broader myeloid panel) is required for classification and risk stratification and has been ordered separately (see accession MMD-26-NGS-22150).

Reviewed and electronically signed by Helena V. Brandt, PhD, FACMG, Director of Cytogenetics, 2026-04-29. This is one component of a combined hematopathology workup and should be interpreted together with the morphology, flow cytometry, and molecular reports.