

Patient: [REDACTED]
DOB: [REDACTED] Age: [REDACTED] Gender: [REDACTED]
Patient Identifiers: [REDACTED]
Visit Number (FIN): [REDACTED]

Client: [REDACTED]
Physician: [REDACTED]

ARUP Test Code: 2011132
Collection Date: 10/27/2016
Received in lab: 10/28/2016
Completion Date: 11/03/2016

Interpretation

Specimen Received
Specimen Type: Peripheral Blood
Reason for Referral: Qualitative Platelet Defects
Test Performed: FISH, AML Panel

ABNORMAL FISH RESULTS

5q31 (EGR1): deletion present
11q23 (KMT2A; also known as MLL): amplification present

NORMAL FISH RESULTS

3q21.3q26.2 (RPN1/MECOM): translocation or inversion not detected
7cen (D7Z1), 7q31 (D7S486): deletion / monosomy not detected
t(8;21)(q22;q22) (RUNX1T1;RUNX1): translocation not detected
11q23 (KMT2A; also known as MLL): rearrangement not detected
16q22 (CBFB): rearrangement not detected

DIAGNOSTIC IMPRESSION:

Fluorescence in situ hybridization (FISH) analysis was performed with the RPN1/MECOM, EGR1, D7Z1, D7S486, RUNX1/RUNX1T1 (also known as AML1/ETO), KMT2A (MLL), and CBFB probes (Abbott Molecular). 200 interphase cells were scored for each probe combination.

This analysis showed evidence of:

- deletion 5q31 involving the EGR1 locus in 129/200 (64.5 percent) cells scored,
- 3-11 copies of the MLL locus at 11q23 in 154/200 (77.0 percent) cells scored. Examination of metaphase spreads present on the interphase FISH slide used in this analysis showed a signal pattern consistent with amplification of this locus.

FISH analysis with the remaining probes showed normal results.

MLL amplification is a recurrent abnormality in de novo and therapy-related myeloid neoplasms. In general, MLL amplification is associated with a poor prognosis. Please correlate these results with clinical and other laboratory findings in this patient

Reference:

Mohamed A. MLL amplification in leukemia. Atlas Genet Cytogenet Oncol Haematol. May 2010. URL: <http://AtlasGeneticsOncology.org/Anomalies/MLLampliID1547.html>

ISCN:

nuc ish(RPN1,MECOM)x2[200],
(EGR1x1)[129/200], (D7Z1,D7S486)x2[200],
(RUNX1T1,RUNX1)x2[200],
amp(KMT2A)[154/200],



Patient: [REDACTED]
ARUP Accession: 16-301-103940

Acute Myeloid Leukemia Panel by FISH

Patient: [REDACTED] | Date of Birth: [REDACTED] | Gender: [REDACTED] | Physician: [REDACTED]
Patient Identifiers: [REDACTED] | Visit Number (FIN): [REDACTED]

(CBFBx2) [200]

This result has been reviewed and approved by [REDACTED],
Ph.D., FACMG
Electronic Signature

Test developed and characteristics determined by ARUP Laboratories. See Compliance
Statement A: aruplab.com/CS



Patient: [REDACTED]
ARUP Accession: 16-301-103940