ARUP LABORATORIES

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Tracy I. George, MD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/22/1934
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

H=High, L=Low, *=Abnormal, C=Critical

Cytogenomic SNP Microarray - Oncology

ARUP test code 2006325

Cytogenomic Microarray SNP - Oncology

Abnormal * (Ref Interval: Normal)

Specimen Type: Bone Marrow
Reason for Referral: AML
Test Performed: CMA ONC

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TEST RESULT

ABNORMAL MICROARRAY RESULTS

Summary of Clinically Significant Alterations (See Note: only large CNVs evaluated by this analysis due to low-level clonal burden):

-Deletions of:
  - 5q (terminal, including EGR1)
  - 17p (interstitial, including TP53)
  - 17q (interstitial, including NF1 and RARA)
  - 18q12.1q23 (interstitial)

-Loss of Chromosome 16
-Gain of Chromosome 21 (4 copies total)

Sex chromosome complement: XY (male)

RESULT DESCRIPTION:

The above abnormalities were observed at a low-level, approximately 10-15 percent in the sample, consistent with a somatic/acquired origin.

NOTE: Due to the presence of low-level clonal burden, this analysis could only evaluate genomic alterations greater than approximately 10-20 Mb in size.

No other significant copy number changes or regions of homozygosity were detected.

INTERPRETATION:
This is a complex genomic profile that includes recurrent alterations in de novo or secondary MDS and AML, including deletion 5q (terminal), monosomy 16, deletion 17p, deletion 18q, and tetrasomy 21. The deletion breakpoint on proximal 17p appears to include the TP53 gene, which is generally associated with poor prognosis. Please correlate this result with clinical and other laboratory findings.

NOTE: FISH using the AML Panel probes was performed concurrently on this sample and was consistent with this genomic profile.

Cytogenetic nomenclature (ISCN):
arr[GRCh37] 5q11.1q35.3(49430268_180719789)x1-2
arr[GRCh37] 16p13.3q24.3(85880_90155062)x1-2
arr[GRCh37] 17p13.3p13.1(604905_8342877)x1-2
arr[GRCh37] 17q11.1q23.2(25270397_58916261)x1-2
arr[GRCh37] 18q12.1q23(27449191_78014123)x1-2
arr[GRCh37] 21q11.2q22.3(15006457_48097372)x2-3

Test Information:
Chromosomal microarray analysis (CMA) was performed using Affymetrix CytoScan HD microarray. This microarray consists of 2,696,550 oligonucleotide probes across the genome, including 1,953,246 unique non-polymorphic probes, and 743,304 SNP (single nucleotide polymorphism) probes. Patient hybridization parameters are normalized to a reference set derived from 100 individuals with normal microarray results. Genomic linear positions are given relative to NCBI build 37 (hg19). Detected aberrations are reported when found to have clear or suspected clinical relevance; aberrations devoid of relevant gene content or reported as common findings in the general population may not be reported.

This microarray and associated software (Chromosome Analysis Suite) are manufactured by Affymetrix and used by ARUP Laboratories for the purpose of identifying DNA copy number gains and losses associated with large chromosomal imbalances. This analysis will not detect all forms of polyploidy, balanced

rearrangements (e.g. inversions and balanced chromosomal translocations), small deletions, point mutations, and some mosaic conditions. While this assay has been extensively validated by ARUP Laboratories and other clinical laboratories per ACMG guidelines, it is not feasible to validate every potential genomic imbalance in the human genome. Furthermore, this technique only identifies the regions of imbalance; it does not provide information regarding the arrangement or mechanisms responsible. For these reasons, we may recommend that some chromosomal microarray results be characterized by fluorescence in situ hybridization (FISH) or standard chromosome analysis.

The functional resolution of this assay varies across different samples dependent upon the size of the abnormality, probe density, tumor content and quality of the DNA obtained. On average, the limit of detection will vary from less than 100 kilobases for samples with high tumor content (generally greater than 70 percent) to several megabases for samples with lower tumor content (25-35 percent). The limit of detection for loss of heterozygosity (LOH) is approximately 3 megabases.

This result has been reviewed and approved by [redacted],
[redacted]

INTERPRETIVE INFORMATION: Cytogenomic Microarray
SNP - Oncology
Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement B: aruplab.com/CS

EER CMA ONC

EERUnavailable

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Cytogenomic Microarray SNP - Oncology	20-057-403616	2/25/2020 9:00:00 AM	2/27/2020 5:30:13 AM	3/6/2020 7 28:00 PM
EER CMA ONC	20-057-403616	2/25/2020 9:00:00 AM	2/27/2020 5:30:13 AM	3/6/2020 7 28:00 PM

END OF CHART

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