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

Physician: Doctor, Example

Patient: Patient, Example

DOB	1/5/1938
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome Analysis, Leukemic Blood with Reflex to Genomic Microarray

ARUP test code 2007131

Chromosome Analysis, Leukemic Blood — See Note (Ref Interval: Normal)

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 Test Performed: Chromosome Analysis
 Specimen Type: Peripheral Blood
 Indication for Testing: D72.820 - Lymphocytosis (symptomatic)

 Number of cells counted: 20
 Number of cells analyzed: 20
 Number of cells karyotyped: 20
 ISCN band level: 400
 Banding method: G-Banding
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 RESULT

 46,XX[20]


 This specimen is being reflexed to genomic microarray
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 Diagnostic Impression:
 Evaluation of metaphase cells from this patient revealed a
normal female chromosome complement. There were no abnormal
clones detected within the limits of the technology utilized in
this study.

 NOTE: FISH CLL P was performed on this sample and reported
under ARUP accession [redacted]. FISH results were NORMAL.

This result has been reviewed and approved by [redacted]

A portion of this analysis was performed at the following
location(s):
[redacted]

INTERPRETIVE INFORMATION: Chromosome Analysis,
                          Leukemic Blood
This test was developed and its performance characteristics
determined by ARUP Laboratories. It has not been cleared or
approved by the US Food and Drug Administration. This test was
performed in a CLIA certified laboratory and is intended for
clinical purposes.
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EER Chrom Analysis LKB w/Rflx to Array — See Note

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

Authorized individuals can access the ARUP Enhanced Report using the following link:

[redacted]

Cytogenomic SNP Microarray - Oncology

ARUP test code 2006325

Cytogenomic Microarray SNP - Oncology

Abnormal * (Ref Interval: Normal)

Test Performed: Cytogenomic SNP Microarray - Oncology (CMA ONC)
Specimen Type: Peripheral blood
Indication for Testing: Lymphocytosis

RESULT SUMMARY
Abnormal Microarray Result (Female)

Clinically Significant CNVs and/or ROH (Tier 1 and Tier 2 Variants):
- Deletion (Loss) 3p21.31 (breakpoint within SETD2)

RESULT DESCRIPTION
This deletion includes most of the SETD2 gene and was observed at approximately 30-40 percent in the sample, consistent with a somatic (acquired) origin.

INTERPRETATION
Deletions involving SETD2 are recurrent findings in acute lymphoblastic leukemia (ALL) and chronic lymphocytic leukemia (CLL). Please correlate this result with clinical and other laboratory findings.

Recommendation:
Monitor by genomic microarray analysis in future studies.

References:
1) Skucha et al. Roles of SETD2 in Leukemia-Transcription, DNA-Damage, and Beyond. Int J Mol Sci. 2019 Feb 27;20(5). PMID: 30818762.
2) Parker et al. Genomic disruption of the histone methyltransferase SETD2 in chronic lymphocytic leukaemia. Leukemia. 2016 Nov;30(11):2179-86. PMID: 27282254.
3) Licht. SETD2: a complex role in blood malignancy. Blood. 2017 Dec 14;130(24):2576-8. PMID: 29242204.

Cytogenomic Nomenclature (ISCN):
arr[GRCh37] 3p21.31(47075716_47464757)x1-2

Technical Information
- This assay was performed using the CytoScan(TM) HD Suite (Thermo Fisher Scientific) according to validated protocols within the Genomic Microarray Laboratory at ARUP Laboratories
- This assay is designed to detect alterations to DNA copy number state (gains and losses) as well as copy-neutral alterations (regions of homozygosity; ROH) that indicate a loss- or absence-of-heterozygosity (LOH or AOH)
- Copy-neutral LOH (CN-LOH) may be present due to acquired UPD (segmental or whole chromosome)
- AOH may be present due to parental relatedness (consanguinity) or uniparental disomy (UPD)
- The detection sensitivity (resolution) for any particular genomic region may vary dependent upon tumor burden, the number of probes (markers), probe spacing, and thresholds for copy number and ROH determination

- The CytoScan HD array contains 2.67 million markers across the genome with average probe spacing of 1.15 kb, including 750,000 SNP probes and 1.9 million non-polymorphic probes
- Genome-wide resolution varies from approximately 25-50 kb for copy number changes and approximately 3 Mb for ROH for samples with high tumor content (generally greater than 70 percent), to several Mb for samples with lower tumor content (20-30 percent)
- The limit of detection for clonality (mosaicism) varies dependent upon the size and type of genomic imbalance. In general, genotype mixture due to mosaicism (distinct cell lines from the same individual) or chimerism (cell lines from different individuals) will be detected when present at greater than 20-30 percent in the sample
- Genomic coordinates correspond to the Genome Reference Consortium human genome build 37/human genome issue 19 (GRCh37/hg19)

Variant Classification and Reporting Criteria
- Variant analysis is performed in accordance with recommendations by the American College of Medical Genetics and Genomics (ACMG), using tiered classification terminology
- Acquired/somatic or constitutional/germline cancer-associated copy number variants (CNVs) and ROH are classified and reported using the following clinical significance categories: Clinically Significant CNVs and/or ROH (Tier 1 and Tier 2 Variants) and Other Clonal Variants (Tier 3)
- Constitutional/germline CNVs not associated with cancer are classified according to the ACMG recommended 5-tier classification system: pathogenic, likely pathogenic, variant of uncertain significance (VUS), likely benign, and benign
-In general, only constitutional CNVs classified as pathogenic or likely pathogenic will be reported using the following clinical significance category: Other Variants (Likely Constitutional)
- Constitutional CNVs conferring non-cancer recessive disease risk will generally not be reported
- CNVs classified as Tier 4, likely benign or benign that are devoid of relevant gene content or reported as common findings in the general population, are generally not reported
- ROH are generally reported when known or suspected to be mosaic and representative of CN-LOH
- Total autosomal homozygosity (only autosomal ROH greater than 3 Mb are considered for this estimate) consistent with AOH at a level of greater than 10 percent will generally be reported; AOH less than 10 percent may be reported, dependent upon on the concern for masked CN-LOH and/or a recessive disorder

Limitations
This analysis cannot provide structural (positional) information associated with genomic imbalance. Therefore, additional cytogenetic testing by chromosome analysis or fluorescence in situ hybridization (FISH) may be recommended.

Certain genomic alterations may not or cannot be detected by this technology. These alterations may include, but are not limited to:
- CNVs below the limit of resolution of this platform
- Sequence-level variants (mutations) including point mutations and indels
- Low-level mosaicism (generally, less than 20-30 percent)
- Balanced chromosomal rearrangements (translocations, inversions and insertions)
- Genomic imbalance in repetitive DNA regions (centromeres, telomeres, segmental duplications, and acrocentric chromosome short arms)

This result has been reviewed and approved by [redacted]

A portion of this analysis was performed at the following location(s):
[redacted]

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

[redacted]

INTERPRETIVE INFORMATION: Cytogenomic Microarray
SNP - Oncology

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Leukemic Blood	23-025-402226	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chrom Analysis LKB w/Rflx to Array	23-025-402226	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cytogenomic Microarray SNP - Oncology	23-025-402226	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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