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

Physician: Doctor, Example

Patient: Patient, Example

DOB	Unknown
Gender:	Unknown
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Chromosome Analysis, Constitutional Blood, with Reflex to Genomic Microarray

ARUP test code 2005763

Chromosome Analysis Constitutional Blood

See Note (Ref Interval: Normal)

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---------------------------------------------------------
Test Performed: Chromosome Analysis
Specimen Type: Peripheral Blood
Indication for Testing: Short stature

Number of cells counted: 20
Number of cells analyzed: 5
Number of cells karyotyped: 5
ISCN band level: 550
Banding method: G-Banding
---------------------------------------------------------
RESULT
Normal Karyotype (Female)

46,XX

This specimen is being reflexed to genomic microarray
---------------------------------------------------------
INTERPRETATION
This analysis showed a normal result.

The standard cytogenetic methodology used in this analysis may
not detect small rearrangements or low-level mosaicism and
cannot detect submicroscopic deletions or duplications that are
detectable by genomic microarray analysis.

Health care providers with questions may contact an ARUP genetic
counselor at (800) 242-2787 ext. 2141.

This result has been reviewed and approved by [redacted]

INTERPRETIVE INFORMATION: Chromosome Analysis
                          Constitutional 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.

Counseling and informed consent are recommended for genetic
testing. Consent forms are available online.
```

EER Chrom Analysis PB 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

ARUP test code 2003414

Cytogenomic SNP Microarray

Normal (Ref Interval: Normal)

Test Performed: Cytogenomic SNP Microarray (CMA SNP)
Specimen Type: Peripheral blood
Indication for Testing: Short stature

RESULT SUMMARY
Normal Microarray Result (Female)

RESULT DESCRIPTION
No clinically significant copy number changes or regions of homozygosity were detected.

INTERPRETATION
This analysis showed a normal result.

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

Cytogenomic Nomenclature (ISCN):
arr(X,1-22)x2

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 an absence- or loss-of-heterozygosity (AOH or LOH)
- AOH may be present due to parental relatedness (consanguinity) or uniparental disomy (UPD)
- LOH may be present due to acquired UPD (segmental or whole chromosome)
- The detection sensitivity (resolution) for any particular genomic region may vary dependent upon 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
- In general, the genome-wide resolution is approximately 25-50 kb for copy number changes and approximately 3 Mb for ROH (See reporting criteria)
- The limit of detection for 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
- Copy number variant (CNV) analysis is performed in accordance with recommendations by the American College of Medical Genetics and Genomics (ACMG), using standard 5-tier CNV classification

terminology: pathogenic, likely pathogenic, variant of uncertain significance (VUS), likely benign, and benign
- CNVs classified as pathogenic, likely pathogenic, or variant of uncertain significance are generally reported, based on information available at the time of review
- Known or expected pathogenic CNVs affecting genes with known clinical significance but which are unrelated to the indication for testing will generally be reported
- Variants that do not fall within the standard 5-tier CNV classification categories may be reported with descriptive language specific to that variant
- In general, recessive disease risk and recurrent CNVs with established reduced penetrance will be reported
- For a list of databases used in CNV classification, please refer to ARUP Constitutional CNV Assertion Criteria, which can be found on ARUP's Genetics website at www.aruplab.com/genetics
- CNVs classified as likely benign or benign that are devoid of relevant gene content or reported as common findings in the general population, are generally not reported
- CNV reporting (size) criteria: losses greater than 50 kb and gains greater than 400 kb are generally reported, dependent on genomic content
- ROH are generally reported when a single terminal ROH is greater than 3 Mb and a single interstitial ROH is greater than 10-15 Mb (dependent upon chromosomal location and likelihood of imprinting disorder) or when total autosomal homozygosity is greater than 3 percent (only autosomal ROH greater than 3 Mb are considered for this estimate)

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)

Data Sharing
In cooperation with the National Institutes of Health's effort to improve understanding of specific genetic variants, ARUP submits HIPAA-compliant, de-identified (cannot be traced back to the patient) genetic test results and health information to public databases. The confidentiality of each sample is maintained. If you prefer that your test result not be shared, call ARUP Laboratories at (800) 242-2787 ext. 3301. Your de-identified information will not be disclosed to public databases after your request is received, but a separate request is required for each genetic test. Additionally, patients have the opportunity to participate in patient registries and research. To learn more, visit ARUP's Genetics website at www.aruplab.com/genetics.

This result has been reviewed and approved by [redacted]

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

INTERPRETIVE INFORMATION: CYTOGENOMIC SNP MICROARRAY

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.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis Constitutional Blood	23-159-122938	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cytogenomic SNP Microarray	23-159-122938	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chrom Analysis PB w/Rflx to Array	23-159-122938	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