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

Physician: Doctor, Example

Patient: Patient, Example

DOB	3/12/2002
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Cytogenomic SNP Microarray

ARUP test code 2003414

Cytogenomic SNP Microarray

Normal (Ref Interval: Normal)

Specimen Received
Specimen Type: Peripheral Blood
Reason for Referral: Secondary amenorrhea
Test Performed: CMA SNP

NORMAL MICROARRAY RESULT

Genetic results
ISCN: arr(1-22,X)x2 (hg19) (normal female result)

The cytogenomic microarray analysis indicated no clinically significant abnormalities and is consistent with a female chromosome complement.

Recommendations:
Genetic Counseling

Certain copy number variants (CNV) have been observed in many individuals with no phenotypic associations and are believed to be clinically insignificant. Any such CNVs which have been detected in this patient are thus not specifically listed in this report.

If you would like additional information, please contact an ARUP genetic counselor at (800) 242-2787 extension 2141. ARUP genetic counselors are available to help health care providers with test selection, result interpretation and identifying local clinical genetic services.

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. These SNP probes allow for the identification of long contiguous stretches of homozygosity (LCSH) that may suggest uniparental disomy (UPD), or regions of the genome identical by descent. Patient hybridization parameters are compared to data derived from 100 individuals with normal microarray results. Deletions smaller than 50 kb and duplications smaller than 400 kb may not be reviewed. Detected copy number variations (CNVs) are reported when found to have clear or suspected clinical relevance; CNVs devoid of relevant gene content or reported as common findings in the general population may not be reported. Regions of homozygosity are reported when a single LCSH is greater than 8-15 Mb (dependent upon chromosomal location and likelihood of imprinting disorder), or when the total autosomal LCSH proportion is

greater than 3 percent (only autosomal LCSH greater than 3 Mb are considered for this estimate). Genomic linear positions are given relative to NCBI build 37 (hg19).

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 (eg. 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.

In cooperation with the National Institutes of Health's effort to improve the 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. Confidentiality of each sample is maintained. You may choose not to share your test result by calling ARUP Laboratories at (800) 522-2787 x3301. Your de-identified information will not be shared with public databases after the request is made, but a separate request is required for each genetic test. For more information see www.aruplab.com/genetics.

This result has been reviewed and approved by [redacted]

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

INTERPRETIVE INFORMATION: CYTOGENOMIC SNP MICROARRAY

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

EER Cytogenomic SNP Microarray	EERUnavailable

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Cytogenomic SNP Microarray	19-128-132505	5/8/2019 10:50:00 AM	5/10/2019 6:40:29 AM	5/15/2019 9:42:00 AM
EER Cytogenomic SNP Microarray	19-128-132505	5/8/2019 10:50:00 AM	5/10/2019 6:40:29 AM	5/15/2019 9:42:00 AM

END OF CHART