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

Physician: Doctor, Example

Patient: Patient, Example

DOB	6/20/2001
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Cytogenomic SNP Microarray - Fetal

ARUP test code 2002366

Maternal Contamination Study Fetal Spec	See Note This specimen shows a mixed cell population with a ratio of 90/10. It is unknown if the fetal sample represents the majority or minority population. Please submit a maternal sample for further correlation. This result has been reviewed and approved by [redacted]
Maternal Specimen	No
Cytogenomic SNP Microarray - Fetal	Normal (Ref Interval: Normal) Test Performed: Cytogenomic SNP Microarray- Fetal (ARRAY FE) Specimen Type: Direct amniocytes (uncultured cells collected from pour-off at first feed of cultures) Indication for Testing: Hydrops --- 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. NOTE: MCC was detected in a concurrent STR analysis with a mixed cell population with a ratio of 90/10. It is unknown if the fetal sample represents the majority or minority population. Please submit a maternal sample for further correlation. 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 HD Suite (Thermo Fisher Scientific) according to validated protocols within the Genomic Microarray Laboratory at ARUP Laboratories

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

- This assay is designed to detect alterations to DNA copy number state (gains and losses), copy-neutral alterations (regions of homozygosity; ROH) that indicate an absence- or loss-of-heterozygosity (AOH or LOH), and certain alterations to ploidy state due to errors at fertilization or early embryonic cell division (i.e. triploidy, molar pregnancy)
- AOH may be present due to molar pregnancy, 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 or likely pathogenic are generally reported based on information available at the time of review
- CNVs classified as VUS are generally reported when found to have suspected clinical relevance based on information available at the time of review, or when meeting size criteria
- 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 1 Mb and gains greater than 2 Mb are generally reported, dependent on genomic content
- Regions of homozygosity (ROH) are generally reported when a single terminal ROH is greater than 3 Mb and a single interstitial ROH is greater than 10-20 Mb (dependent upon chromosomal location and likelihood of imprinting disorder) or when total autosomal homozygosity is greater than 5 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)
- Most cases of tetraploidy

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 - Fetal

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.

Chromosome Analysis, Amniotic Fluid, with Reflex to Genomic Microarray

ARUP test code 2008367

Chromosome Analysis, Amniotic Fluid	See Note	(Ref Interval: Normal)

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

Specimen received

Specimen type: Amniotic Fluid
Reason for referral: Hydrops
Test performed: Chromosome Analysis

Laboratory analysis

Number of cells counted: 15
Number of colonies counted: 15
Number of cells analyzed: 15
Number of cells karyotyped: 15
ISCN Band level: 400
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]

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

INTERPRETIVE INFORMATION: Chromosome Analysis, Amniotic Fluid

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 AF w/Rflx to Array

See Note

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

[redacted]

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	22-357-108478	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Specimen	22-357-108478	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Chromosome Analysis, Amniotic Fluid	22-357-108478	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cytogenomic SNP Microarray - Fetal	22-357-108478	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Chrom Analysis AF w/Rflx to Array	22-357-108478	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