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

Physician: Doctor, Example

Patient: Patient, Example

DOB	4/11/1986
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	Fetal Cells Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination. This result has been reviewed and approved by [redacted]
Maternal Specimen	Yes
Cytogenomic SNP Microarray - Fetal	Normal (Ref Interval: Normal) Test Performed: Cytogenomic SNP Microarray- Fetal (ARRAY FE) Specimen Type: Direct (uncultured) amniocytes Indication for Testing: Abnormal NIPT for T18 --- RESULT SUMMARY Normal Microarray Result (Male) --- 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. Cytogenetic Nomenclature (ISCN) arr(1-22)x2,(X,Y)x1 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 - 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)

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

- 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 ARUPs 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] INTERPRETIVE INFORMATION: Cytogenomic SNP Microarray - Fetal Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS
EER Cytogenomic SNP Microarray - Fetal	EERUnavailable

Chromosome FISH, Amniotic Fluid with Reflex to Chromosome Analysis or Genomic Microarray

ARUP test code 2011130

Chromosome FISH, Prenatal	See Note	(Ref Interval: Normal)

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Specimen Received
Specimen Type: Amniotic Fluid
Reason for Referral: Abnormal Maternal Serum Screen- T18
Test Performed: AF F RFLX

NORMAL FISH RESULT

DIAGNOSTIC IMPRESSION:
Prenatal interphase fluorescence in situ hybridization (FISH) analysis was performed with chromosome enumeration probes for 13, 18, 21, X and Y using the FDA-approved AneuVysion probe kit (Abbott Molecular). 50 interphase cells were scored for each probe.

This analysis showed normal results with no evidence of aneuploidy.

Sex chromosomes: XY (male)

This specimen is being reflexed to genomic microarray

ISCN:
nuc ish(DXZ1x1,DYZ3x1,D18Z1x2),
(RB1x2,D21S259/D21S341/D21S342x2)

PLEASE NOTE: Interphase FISH will not detect approximately one third of prenatal chromosome abnormalities, which includes mosaicism for the above chromosomes, structural abnormalities, and other numerical chromosome abnormalities. Therefore, routine cytogenetic analysis or genomic microarray is recommended for the final interpretation. If either test is being performed, it will be reported separately.

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,
Prenatal FISH

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.

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	21-014-113961	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Specimen	21-014-113961	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cytogenomic SNP Microarray - Fetal	21-014-113961	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Cytogenomic SNP Microarray - Fetal	21-014-113961	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Chromosome FISH, Prenatal	21-014-113961	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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