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Julio C. Delgado, MD, MS, Director of Laboratories

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/17/1985
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 Pinar Bayrak-Toydemir, M.D., Ph.D.
	INTERPRETIVE INFORMATION: Maternal Cell Contamination, Fetal Specimen Please refer to fetal report for interpretation.
Maternal Specimen	Yes
Cytogenomic SNP Microarray - Fetal	Normal (Ref Interval: Normal) Specimen Received Specimen Type: Direct (uncultured) villi Reason for Referral: Abnormal maternal serum screen: Increased risk for T21; Abnormal ultrasound: increased NT Test Performed: ARRAY FE --- NORMAL MICROARRAY RESULT Genetic results ISCN: arr(1-22)x2,(XY)x1 (hg19) (normal male result) The cytogenomic microarray analysis indicated no clinically significant abnormalities and is consistent with a male 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

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

	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 1 Mb and duplications smaller than 2 Mb may not be reported. 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 15 Mb (dependent upon chromosomal location and likelihood of imprinting disorder), or when the total autosomal LCSH proportion is greater than 10 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. This test was developed and its performance characteristics determined by ARUP Laboratories. The results are not intended to be used as the sole means for clinical diagnosis or patient management decisions. This result has been reviewed and approved by Allen N. Lamb, Ph.D., FACMG
	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, Chorionic Villus with Reflex to Chromosome Analysis or Genomic Microarray

ARUP test code 2011131

Chorionic Villus, FISH	See Note	(Ref Interval: Normal)

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Specimen Received
Specimen Type: Chorionic Villi
Reason for Referral: Abnormal Maternal Serum Screen (T21 1/90)
1st Trimester Screen, NT= 95 Percent
Test Performed: FISH, CVS 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 AneuVysion probe kit (Abbott Molecular). 50 interphase cells were scored for each probe.

This analysis showed no evidence for a numerical abnormality for chromosomes 13, 18, 21, X, or Y.

Sex chromosomes: XY (male)

FISH analysis performed on CVS presumes that the fetal chromosome complement is accurately reflected in the extra-embryonic tissue. There are rare examples in which the karyotype of the CVS is not consistent with that of the fetus.

NOTE: 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 include 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 Denise I. Quigley, Ph.D., FACMG

INTERPRETIVE INFORMATION: Flourescence in Situ Hybridization, CVS
Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	18-247-115024	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Specimen	18-247-115024	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cytogenomic SNP Microarray - Fetal	18-247-115024	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Cytogenomic SNP Microarray - Fetal	18-247-115024	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Chorionic Villus, FISH	18-247-115024	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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