ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/6/1994
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Genomic SNP Microarray, Products of Conception

ARUP test code 2005633

SNP Microarray, Products of Conception

Abnormal * **(Ref Interval: Normal)**

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Test Performed: Genomic SNP Microarray, Products of Conception
(ARRAY POC)
Specimen Type: Products of Conception (Tissue: Villi)
Indication for Testing: POC
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RESULT SUMMARY
Abnormal Microarray Result (Male)

Trisomy 16

Classification: Pathogenic
Copy number change: 16p13.3q24.3 gain
Size: 90.1 Mb
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RESULT DESCRIPTION
This analysis showed a gain of all probes on chromosome 16,
indicating an additional copy (trisomy) of this chromosome.

INTERPRETATION
This result is consistent with a diagnosis of trisomy 16.
Trisomy 16 has a highly variable phenotype which may include
intrauterine growth restriction, intrauterine fetal demise,
heart defects, and other anomalies. Autosomal trisomy is the
most frequent type of chromosomal abnormality in pregnancy loss
and is usually sporadic.

Recommendation:
Genetic counseling

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

References:
1) Gardner and Amor. Gardner and Sutherlands Chromosome
Abnormalities and Genetic Counseling, 5th ed. New York, NY:
Oxford University Press; 2018.
2) Benn. Trisomy 16 and trisomy 16 Mosaicism: a review. Am J Med
Genet. 1998 Sep 1;79(2):121-33. PMID: 9741470.
3) Milunsky. Genetic Disorders and the Fetus: Diagnosis,
Prevention and Treatment. 7th edition. West Sussex, UK: John
Wiley and Sons; 2016.

Cytogenomic Nomenclature (ISCN):
arr(16)x3

Technical Information
- This assay was performed using the CytoScan(TM) HD Suite
(Thermo Fisher Scientific) according to validated protocols
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H=High, L=Low, *=Abnormal, C=Critical

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), 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.

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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

A portion of this analysis was performed at the following location(s):
ARUP Laboratories Site CG-WA#2

INTERPRETIVE DATA: Genomic SNP Microarray,
Products of Conception
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
SNP Microarray, Products of Conception	23-221-115520	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