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

Physician: Doctor, Example

Patient: Patient, Example

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

Cytogenomic SNP Microarray, Family-Specific Variant

ARUP test code 3005694

Cytogenomic SNP Microarray, Fam Spec Var

Abnormal

```
RRAY FSV POS
Test Performed: Cytogenomic SNP Microarray, Family Specific
Variant (ARRAY FSV)
Specimen Type: Peripheral blood
Indication for Testing: Familial testing
------------------------------------------------------------
RESULT

16p11.2 Deletion:  DETECTED

Copy number change: 16p11.2 loss
Size: 540 kb
------------------------------------------------------------------
-----
INTERPRETATION
This targeted analysis was positive for the requested copy
number variant (CNV). Genomic coordinates below correspond to
the region interrogated in this study.

This result indicates that this individual is a carrier of the
CNV identified in the proband. Please refer to the proband
report for the interpretation of this finding. Variants outside
of this targeted region were not evaluated.

Recommendation:
Genetic counseling

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

Cytogenomic Nomenclature (ISCN):
arr[GRCh37] 16p11.2(29651576_30191848)x1

This result has been reviewed and approved by [redacted]
```

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

Background Information: Cytogenomic SNP Microarray, Family-Specific Variant

Technical Information
- This is a targeted microarray test for detection of previously identified familial copy number variants (CNVs). Only requested CNVs that are detectable on this platform will be evaluated.
- Analysis is performed using the CytoScan(TM) HD Suite (Thermo Fisher Scientific) according to validated protocols established within the Genomic Microarray Laboratory at ARUP Laboratories.
- 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 nonpolymorphic probes.
- 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 determination. The general limit of detection for copy number changes across this platform is approximately 25-50 kb.
- 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) will be detected when present at greater than 20 to 30 percent in the sample.
- Genomic coordinates interrogated for targeted CNV analysis correspond to the Genome Reference Consortium human genome build 37/human genome issue 19 (GRCh37/hg19). CNVs reported in other genome builds will be converted to hg19 before interrogation.

Variant Classification and Reporting Criteria
- Interrogated CNV(s) will be reported as "Detected" or "Not Detected."
- In general, requested CNVs will not be classified and an interpretation will not be provided. Reference the original proband report for the interpretation of any interrogated variant.
- Parentage will not be evaluated.
- Inheritance interpretation is based on the familial relationships provided to the laboratory.

Limitations
- This assay only analyzes requested familial CNV(s) and does not rule out alterations outside of the targeted region.
- This analysis cannot provide structural (positional) information associated with genomic imbalance. In some cases, 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 to 30 percent).
- Balanced chromosomal rearrangements (translocations, inversions, and insertions).
- Genomic imbalance in repetitive DNA regions (centromeres, telomeres, segmental duplications, and acrocentric chromosome short arms).

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. 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
Cytogenomic SNP Microarray, Fam Spec Var	24-093-102615	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