ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Gender: Unknown

Specimen Collected: 13-Sep-21 12:29

Cytogenomic MIP Array FFPE POC |Received: 13-Sep-21 14:53 Report/Verified: 14-Sep-21 09:17

Procedure	Result	Units	Reference Interval
Cytogenomic MIP Array FFPE,POC	Normal f1 i1		Normal
EER Cytogenomic MIP Array FFPE,POC	EERUnavailable		

Cytogenomic MIP Array FFPE POC |Received: 13-Sep-21 14:53 Report/Verified: 14-Sep-21 09:44

Procedure	Result	Units	Reference Interval
Block ID	SP21-1234 1B		

Result Footnote

f1: Cytogenomic MIP Array FFPE, POC
Test Performed: Cytogenomic Molecular Inversion Probe Array, FFPE Tissue- Products of Conception (CMA PFFPE)
Specimen Type: Products of Conception (Villi / Fetal Tissue)
Estimated Villi/Fetal Content: _ percent
Indication for Testing: _

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.

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 OncoScan(TM) CNV Assay (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) 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 OncoScan CNV array contains over 220,000 SNP probes with a median probe density (kb/probe) of 16-19 kb
- In general, the genome-wide resolution is approximately 300-400 kb for copy number changes and approximately 5 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 30 percent in the sample
- Genomic coordinates correspond to the Genome Reference Consortium human genome build 37/human genome

Result Footnote

f1: Cytogenomic MIP Array FFPE, POC
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, likely pathogenic or variant of uncertain significance, are generally reported, based on information available at the time of review
- 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 and gains greater than 500 kb are generally reported, dependent on genomic content
- ROH are generally reported when a single terminal ROH is greater than 5 Mb and a single interstitial ROH is greater than 10-15 Mb (dependent upon chromosomal location and likelihood of imprinting disorder) or when total autosomal homozygosity is greater than 3 percent (only autosomal ROH greater than 5 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 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

Test Information

i1: Cytogenomic MIP Array FFPE, POC

INTERPRETIVE INFORMATION: Cytogenomic Molecular Inversion Probe Array, FFPE Tissue – 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.