Clinical Genomics Laboratory

Phone: (206) 616-4062
Lab Genetic Counselor: (206) 598-8684

UW Medicine
LABORATORY MEDICINE & PATHOLOGY

Patient: [redacted]
Sex: female, DOB: [redacted]
MRN: [redacted]

SNP Microarray, Constitutional (Final result)

Indication for Testing
Abnormal fetal ultrasound: Echogenic intracardiac focus

RESULTS SUMMARY
Normal constitutional chromosomal SNP microarray results from amniotic fluid

Interpretation and Comments
No pathogenic or likely pathogenic variants were detected in this amniotic fluid sample, including deletions, duplications, and unusually large or numerous regions of copy number-neutral absence of heterozygosity.

Results were consistent with one copy of the X and one copy of the Y chromosome.

Benign, likely benign, and copy number variants of unclear significance (VUS) were not reported. If desired, an amended report can be issued that includes any VUS that were detected. Please call the lab genetic counselor at 206-598-8684 to request this service.

Recommendations:
1. Chromosomal SNP microarray analysis results should be interpreted in the context of the patient's clinical and family history.

2. Genetic counseling can be helpful to patients in understanding their test results.

References:
ClinGen: https://www.clinicalgenome.org/
ClinGen Dosage Sensitivity Map: https://dosage.clinicalgenome.org/
ClinVar: https://www.ncbi.nlm.nih.gov/variation/view/
DECIPHER: https://decipher.sanger.ac.uk/
DGV: http://dgv.tcag.ca/dgv/app/home
Ensembl: https://uswest.ensembl.org/index.html
GeneReviews: http://www.ncbi.nlm.nih.gov/books/NBK1116/
gnomAD: https://gnomad.broadinstitute.org/
MANE: https://www.ncbi.nlm.nih.gov/refseq/MANE/
MedGen: http://www.ncbi.nlm.nih.gov/medgen
OMIM: http://omim.org/
UCSC genome browser: http://genome.ucsc.edu/
UniProt: https://www.uniprot.org/

Test Limitations: The Illumina Infinium CytoSNP-850K BeadChip is used in this test for the sole purpose of identifying genomic chromosomal abnormalities. This microarray will detect aneuploidy as well as copy number gains (duplications or amplifications), copy number losses (deletions), and copy-neutral regions of homozygosity (ROH) for the loci represented on the microarray. Analysis in our laboratory is limited to detecting copy number variants (deletions and duplications) that include at least 10 SNPs and regions of ROH that include at least 500 SNPs. Abnormalities below these levels of resolution may not be detected. Uniparental disomy is reported when a telomeric region of ROH is >5 Mb in size or an interstitial region of ROH is >10

Mb in size for imprinted chromosomes or >15 Mb in size for non-imprinted chromosomes. Possible identity by descent is reported when regions of ROH >3 Mb in size comprise ≥1.5% of the autosomal genome. Copy number variants (CNVs) are interpreted according to technical standards established by the American College of Medical Genetics and Genomics (PMID: 31690835). Pathogenic and likely pathogenic CNVs are reported, regardless of size, including deletions conferring probable carrier status for autosomal recessive conditions. CNVs ≥400 kb that are classified as of uncertain significance are reported per provider request; those <400 kb are not reported. Benign and likely benign CNVs are not reported.

Chromosomal SNP microarray analysis will not detect imbalances in the regions not represented on the microarray, low-level mosaicism (<20%), tetraploidy, balanced alterations (e. g. reciprocal translocations, Robertsonian translocations, inversions, balanced insertions), methylation anomalies and other epigenetic events, single nucleotide variations, or small insertions or deletions. The failure to detect an alteration at any locus does not exclude the diagnosis of any of the disorders represented on the microarray. The laboratory can assist the ordering provider in determining whether other types of testing are appropriate. This discussion should be considered in the context of the clinical indication for testing.

The performance characteristics of this test have been validated by the Clinical Genomics Laboratory in the University of Washington School of Medicine as required by CLIA '88 regulations. It has not been cleared or approved for specific uses by the U.S. Food and Drug Administration. Pursuant to the requirements of CLIA '88, this laboratory has established and verified the test's accuracy and precision. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research.

Methods Summary: Genomic DNA extracted from the patient sample was amplified, fragmented, and hybridized to the Illumina Infinium CytoSNP-850K BeadChip v1.2. This microarray contains 850,000 probes for SNP markers. The approximate distance between probes covering genes of known relevance in either constitutional or neoplasia settings is 1 kb; the spacing over the remainder of the genomic backbone is about 5 kb (www.illumina.com/content/dam/illumina-marketing/documents/products/datasheets/datasheet_CytoSNP850K_POP.pdf). After hybridization, the microarray was washed manually, extended and stained by Illumina Automation Tecan 8-tip Robot, and scanned with an Illumina iScan. Allele and intensity ratio data of the fluorescent signals were generated. Microarray data were visualized and analyzed using Illumina BlueFuse Multi 4.5 and NxClinical 6.1 to identify chromosomal copy number variants and copy-neutral regions of homozygosity. Genome build GRCh38 was used

ISCN
arr(X,Y)x1,(1-22)x2

Chip ID
[redacted]

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ID:	[redacted]	Test:	SNP Microarray, Constitutional
Type:	Amniotic fluid	Source:	Amniotic fluid
Ordered by:		Authorized by:	[redacted]
Collected:	[redacted]	Last Received:	1/3/2023 2:57 PM
Verified On:	1/18/2023 5:33 PM	Verifying User:	Liu, Yajuan J, PhD

Resulting Labs

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