ARUP Laboratories
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*****Example Report*****

Patient Age/Gender: Unknown Unknown
Printed: 16-Dec-19 11:40:02

Procedure	Result	Units	Ref Interval	Accession	Collected	Received	Reported/ Verified
Cytogenomic MIP Array FFPE, POC	Normal f		[Normal]	19-343-900154	09-Dec-19 12:32:00	09-Dec-19 12:32:00	12-Dec-19 13:40:33
EER Cytogenomic MIP Array FFPE, POC	EERUnavailable			19-343-900154	09-Dec-19 12:32:00	09-Dec-19 12:32:00	12-Dec-19 13:40:33
Block ID	MKS23-111996 1A			19-343-900154	09-Dec-19 12:32:00	16-Dec-19 10:20:00	16-Dec-19 10:20:34

09-Dec-19 12:32:00 Cytogenomic MIP Array FFPE, POC:

Specimen Received
Specimen Type: FFPE Villi
Estimated Villi Content: 95 percent
Reason for Referral: Holoprosencephaly
Test Performed: CMA PFFPE

NORMAL MICROARRAY RESULT
ISCN: arr(1-22)x2,(XY)x1 (hg19)(normal male result)

Interpretation:
The FFPE microarray analysis showed no clinically significant DNA copy number changes or long contiguous stretches of homozygosity (LCSH), and is consistent with a male chromosome complement.

Recommendations:
Genetic counseling

If you would like additional information, please contact an ARUP genetic counselor at (800) 242-2787 x 2141. ARUP genetic counselors are available to help health care providers with test selection, result interpretation and identifying local clinical genetic services.

Formalin-Fixed Paraffin-Embedded (FFPE) Molecular Inversion Probe (MIP) Array Methodology:
FFPE MIP Array was performed using the Affymetrix OncoScan FFPE Assay. This technology contains 220,000 probes across the genome for detection of copy number changes and long contiguous stretches of homozygosity (LCSH).

Patient hybridization parameters are compared to data derived from over 300 FFPE samples from unaffected tissues. Detected gains, losses and LCSH are reported based on genomic content. Gains, losses and LCSH devoid of relevant gene content or commonly detected in the general population may not be reported. Gains and losses less than 500 kilobases may not be investigated. Long contiguous stretches of homozygosity (LCSH) may indicate either a uniparental disomy (UPD) if it occurs within a chromosome or regions identical by descent (IBD) if it occurs across numerous independent regions. Haplotype structure within populations allows for normal stretches of homozygosity, usually under 8 to 10 Mb in size. Therefore, a size filter for a LCSH is important to distinguish between normal homozygosity for a region versus homozygosity due to potential UPD or IBD. LCSH on imprinted chromosomes (6, 7, 11, 14 and 15) less than 8 Mb (telomeric) or 10 MB (interstitial) may not be investigated or reported. LCSH less than 10 Mb (telomeric) or 15 Mb (interstitial) on non- imprinted chromosomes may also not be investigated or reported. Homozygosity of approximately 3 percent of the genome or greater may be reported as it suggests an increased risk for a recessive condition. Genomic linear positions correspond to the NCBI Genome Reference Consortium Human Build 37 (GRCh37/hg19).

Chromosome Analysis Suite, manufactured by Affymetrix, was used for the purpose of identifying DNA copy number gains and losses and LCSH. This analysis will not detect balanced rearrangements, such as translocations, inversions and balanced insertions. Although triploidy can be detected due to the associated allelic imbalance, tetraploidy is not detectable by this method. Additionally base pair mutations, genomic imbalances below the resolution of this array platform, and aberrations in regions of the genome not represented on the array platform will not be detected. Low-level mosaicism (<25 percent) may also be undetectable, and in some cases, mosaicism will result in discrepancies between the karyotype and microarray analysis. This technology cannot determine positional information regarding the genomic location of copy number alterations and may not be able to distinguish between mechanisms of origin for certain genomic aberrations. The failure to detect an alteration at any locus does not exclude the diagnosis of any of the disorders represented on the microarray. Validation of this assay was performed according to ACMG guidelines (American College of Medical Genetics standards and guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013. South ST, Lee C, Lamb AN, Higgins AW, Kearney HM; Working Group for the American College of Medical Genetics and Genomics Laboratory Quality Assurance Committee. Genet Med. 2013 Nov;15(11):901-9.)

This result has been reviewed and approved by Erica F. Andersen, Ph.D., FACMG

***** Abnormal, **#** = Corrected, **C** = Critical, **f** = Footnote, **H** = High, **L** = Low, **t** = Interpretive Text, **@** = Reference Lab

09-Dec-19 12:32:00 Cytogenomic MIP Array FFPE, POC:
INTERPRETIVE INFORMATION: Cytogenomic Molecular Inversion Probe Array, FFPE Tissue – Products of Conception

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS