

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 4/18/2017
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 01/01/2017 12:34

Cytogenomic Molecular Inversion Probe Array, FFPE Tissue - Oncology

ARUP test code 2010229

Cytogenomic MIP Array, FFPE

Abnormal * (Ref Interval: Normal)

Specimen Received
Specimen Type: FFPE Tumor
Estimated Tumor Burden: 70 - 80 percent
Reason for Referral: Wilms tumor
Test Performed: FFPE ARRAY

TEST RESULT

ABNORMAL MICROARRAY RESULTS

The FFPE microarray analysis performed on tumor tissue showed the following abnormalities present in approximately 100 percent of the sample:

- Terminal deletion of 16q11.2q24.3 (43.7 Mb)
- Gain of 1q21.1q32.2 (65.7 Mb) and 1q32.2q44 (terminal, 38.8 Mb)
- Trisomy 12

The FFPE microarray analysis also showed a deletion of 3q27.3 (748 kb; involving EIF4A2 gene) present in approximately 60 percent of the sample, as well as a deletion of 3q26.32 (263 kb; involving TBL1XR1 gene), gain of 4q34.2q34.3 (314 kb; involving VEGFC gene) and 7q36.3 (871 kb) present in approximately 30 percent of the sample:

Sex chromosome complement: XY (male)

Interpretation:
This genomic microarray analysis showed recurrent abnormalities observed in Wilms tumor. The microarray patterns observed for deletion of 16q and gain of 1q suggest the presence of a derivative chromosome 16 resulting from an unbalanced translocation between chromosomes 1 and 16 - der(16)t(1;16). der(16)t(1;16)(q10-21;q10-24) has been reported to be associated with tumor progression. Trisomy 12 is observed in 25% of Wilms tumor cases. Please correlate these results with clinical and other laboratory findings in this patient.

No other significant DNA copy number changes or copy neutral loss of heterozygosity was detected.

SUMMARY OF ABNORMALITIES DETECTED (PATHOGENIC, ACQUIRED, hg19):
arr[GRCh37] 1q21.1q32.2(144009052_209712087)x3
arr[GRCh37] 1q32.2q44(210381256_249212878)x3
arr[GRCh37] 3q26.32(176696110_176959505)x1-2
arr[GRCh37] 3q27.3(186296303_187043868)x1-2
arr[GRCh37] 4q34.2q34.3(177416241_177730455)x2-3
arr[GRCh37] 7q36.3(155124599_155995315)x2-3
arr[GRCh37] 12p13.33q24.33(189399_133818115)x3

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

Unless otherwise indicated, testing performed at:

arr[GRCh37] 16q11.2q24.3(46461308_90158005)x1

Formalin-Fixed Paraffin-Embedded (FFPE) Molecular Inversion Probe 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 loss of heterozygosity (LOH). Chromosome Analysis Suite, manufactured by Affymetrix, was used for the data analysis.

Patient hybridization parameters are normalized to a reference set derived from over 300 FFPE samples from unaffected tissues. Detected gains, losses and LOH are reported when found to have clear or suspected clinical relevance. Gains, losses and LOH devoid of relevant gene content or commonly detected in the general population may not be reported. Genomic linear positions correspond to the NCBI Genome Reference Consortium Human Build 37 (GRCh37/hg19).

The functional resolution of this assay varies across different samples and across the genome, dependent upon the size of the abnormality, probe density in the region, tumor content and quality of the DNA obtained. The limit of detection will range from approximately 400 kilobases genome-wide, with higher resolution in targeted regions containing cancer genes for samples with high tumor content (generally greater than 70 percent); to several megabases for samples with lower tumor content (30-40 percent). The limit of detection for LOH is approximately 3 megabases.

This test is used by ARUP Laboratories for the purpose of identifying DNA copy number gains and losses as well as copy-neutral LOH. This analysis will not detect all forms of polyploidy, balanced rearrangements (e.g., inversions and balanced chromosomal translocations), small deletions, point mutations, and some mosaic conditions. 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. Validation of this assay was performed according to ACMG guidelines [American College of Medical Genetics and Genomics technical standards and guidelines: microarray analysis for chromosome abnormalities in neoplastic disorders. Cooley LD, Lebo M, Li MM, Slovak ML, Wolff DJ; Working Group of the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. Genet Med. 2013 Jun;15(6):484-94]. While extensive efforts are made to analyze a variety of genomic alterations that may be encountered during clinical testing, analysis of all potential genomic aberrations is not practically feasible in a validation study.

This result has been reviewed and approved by [REDACTED]

INTERPRETIVE INFORMATION: Cytogenomic Molecular Inversion Probe Array, FFPE Tissue
- Oncology

For detection of copy number alterations and loss of heterozygosity in FFPE specimens.

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

EER Cytogenomic MIP Microarray, FFPE See Note

Block ID SSW20-6531 A13

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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 20-085-400051
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Cytogenomic MIP Array, FFPE	20-085-400051	3/18/2020 4:36:00 PM	3/25/2020 4:53:23 AM	4/8/2020 7:06:00 PM
EER Cytogenomic MIP Microarray, FFPE	20-085-400051	3/18/2020 4:36:00 PM	3/25/2020 4:53:23 AM	4/8/2020 7:06:00 PM
Block ID	20-085-400051	3/18/2020 4:36:00 PM	3/25/2020 4:53:23 AM	3/25/2020 7:30:00 AM

END OF CHART

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

Unless otherwise indicated, testing performed at:

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