Chromosome Microarray-Product of Conception

Test Description

This array detects copy number changes and absence of heterozygosis (AOH), which may be due to uniparental disomy (UPD) or identity by descent (IBD). The array detects gains and losses at a minimum of 5Mb and 5Mb, respectively, across the genome, for clinically relevant deletion/duplication syndromes, sub-telomere and peri-centromere region or targeted genes. Significant AOH is reported for long stretches of DNA on a single chromosome suggestive of UPD or on multiple chromosomes, suggestive of IBD. Benign copy number changes are not reported.

Patient Demographic

Name: Ms. Suman Mahender Malani
Sex: Female
Date of Birth/Age: 27 years

Clinician

Clinician Name: Dr Amit Verma
Medical Facility: Dr AV Institute of Personalized Therapy and Cancer Research (IPTCR)
Pathologist: Not Provided

Specimen

Booking ID: 012202080102
Site: NA
Sample Type: POC Tissue
Date of Collection: 15-01-2022
Date of Booking: 15-01-2022

CLINICAL INDICATION

Spontaneous expulsion of fetus. Anomalous baby with intrauterine growth restriction (IUGR) with BPV.

RESULTS

Chr14q32.2q33.33(97,057,997-107,285,437) x1 [hg19]
Abnormal, Pathogenic

INTERPRETATION

Chromosomal microarray analysis on the DNA from this specimen identified an approximately 10,227Mb deletion in the long arm of chromosome 14.

A deletion of this size and genomic content is expected to be the cause of miscarriage.

RECOMMENDATIONS

- To further evaluate clinical relevance, parental microarray studies are recommended to determine whether this deletion is a de novo change or resulting from a familial chromosome rearrangement.
- Genetic counseling is recommended for this family.

Jatinder Kaur, PhD
Head, Molecular Biology & Genomics

Dr. Gulshan Yadav, MD
Head, Pathology

APPENDIX 1: TEST METHODOLOGY

Method

Whole genome oligonucleotide array analysis was performed with following specifications:
Microarray chip: CytoScan-Optima (Affymetrix), Whole Genome Array
Type: SNP
Human Genome Build: hg19

DISCLAIMER

- This test has not been cleared or approved for specific uses by the U.S. Food and Drug Administration or any Indian Governmental regulatory or medical Institution(s)/agency/department /association.
- The report shall be generated within turnaround time (TAT), however, such TAT may vary depending upon the complexity of test(s) requested. MolQ Laboratory under no circumstances will be liable for any delay beyond afore mentioned TAT.
- This test was performed at reference laboratory. This is a technical report about the findings of the array result as interpreted by reference laboratory to their best knowledge based on the information available at the time of reporting. Clinical correlation and correlation with alternate testing are recommended before taking any further medical decisions.
- Although the methodology used in this analysis and interpretation is highly accurate, failure to detect an alteration at any locus does not exclude the diagnosis of any of the disorders. MolQ can assist the physician in determining the appropriate test in the context of clinical findings.
- It is hereby clarified that the report(s) generated from the test(s) do not provide any diagnosis or opinion or recommends any cure in any manner. MolQ Laboratory hereby recommends the patient and/or the guardians of the patients, as the case may be, to take assistance of the clinician or a certified physician or doctor, to interpret the report(s) thus generated. MolQ Laboratory hereby disclaims all liability arising in connection with the report(s).
- The test results are subject conditions of reporting.
  - Results pertain to the specimen submitted at the lab. It is presumed that the specimen belongs to the patient's name provided at the time of submission and the verification of the identity has been carried out at the point of generation of the said samples. Results relate only to the standards/procedures indicated in the report.
  - The laboratory may request additional or repeat sample to complete testing/analysis.
  - The laboratory assures that all the tests have been carried out with reasonable technical integrity however machine and human errors cannot be completely rule out. The results are subject to the quality of the specimen and pre/post analytical variations. Hence verification of test results with alternate methods and correlation with clinical indications is recommended before arriving at any conclusion. The laboratory shall not be responsible for any adverse effects, loss or damage caused by any clinical decisions taken on the basis of this report.
  - In case of any clerical errors or discrepancy with clinical observations, kindly notify the laboratory immediately for verification and rectification.
  - This report is not valid for medio-legal purposes.
  - Partial reproduction of this report is not permitted.
  - Any sample which remains after testing, will be retained in accordance with company retention policies, unless otherwise agreed by and between the client and company in writing.

LIMITATIONS

- This test will only detect gain or loss of entire chromosomes, deletions and duplications of the loci represented on this array.
- Balanced rearrangements such as Robertsonian translocations, reciprocal translocations, inversions and balanced insertions will not be detected by this test.
- This test will also not detect DNA mutations, deletions or duplications below the resolution of this array or diseases associations based on linkage analysis.
- SNP analysis can detect some cases of uniparental isodisomy but not all.
- The detection of mosaicism by this array is variable and is dependent on the size of the chromosomal imbalance, the type of the array used and the quality of array results.
- Results of this test are for investigational purposes only as per the assay's manufacturer.