CHROMOSOMAL MICROARRAY

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SAMPLE TYPE : Product of Conception (POC)
SAMPLE QUALITY : Accepted
CLINICAL HISTORY : -
TEST REQUIRED : Chromosomal Microarray
GESTATIONAL AGE : NA
MATERNAL CELL CONTAMINATION : No maternal cell contamination identified.

RESULTS

MOLECULAR KARYOTYPE (750K CHIP)	CYTOBAND	CNV TYPE	SIZE	COPY NUMBER	CLASSIFICATION
arr(46, X ?)	Normal	Normal	Normal	2N	Normal

INTERPRETATION

Chromosomal microarray analysis showed a Normal Karyotype

KARYOVIEW

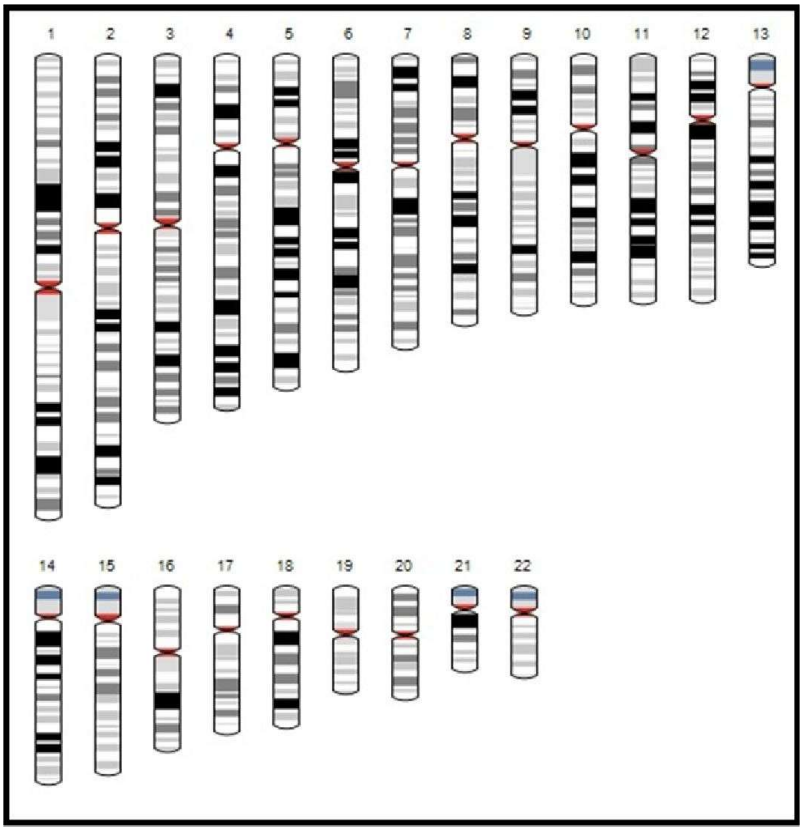

Fig1: Karyoview of MMG023618CMA_POC0325400

List of Syndromes	Result
Autosomal Aneuploidies	
Trisomy 21 (Down syndrome)	Negative
Trisomy 18 (Edwards syndrome)	Negative
Trisomy 13 (Patau syndrome)	Negative
Other autosomal aneuploidies	Negative
Sex Chromosome Aneuploidies	
Monosomy X (Turner syndrome)	Negative
XYY (Jacobs syndrome)	Negative
XXY (Klinefelter syndrome)	Negative
XXX (Triple X syndrome)	Negative
Euploidy	
Triploidy	Negative
Clinically significant Genome-wide copy number variations	
Duplications (Gains)	Negative
Deletions (Losses)	Negative
Loss of heterozygosity	Negative

RECOMMENDATIONS

Clinical correlation is suggested & further genetic counseling is recommended

REFERENCES

1. Gonzales PR, Andersen EF, Brown TR, et al. Interpretation and reporting of large regions of homozygosity and suspected consanguinity/uniparental disomy, 2021 revision: A technical standard of the American College of Medical Genetics and Genomics (ACMG).Genet Med. 2022;24(2):255-261. doi:10.1016/j.gim.2021.10.004

2. Miller, David T et al. Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. American journal of human genetics vol. 86,5 (2010): 749-64. doi:10.1016/j.ajhg.2010.04.006

3. https://medlineplus.gov/genetics/condition/turner-syndrome/.

LIMITATIONS

1. Chromosomal microarray analysis will not detect imbalances in regions of the genome not represented on the microarray, low-level mosaicism (<20%), tetraploidy, balanced alterations (e.g. reciprocal translocations), methylation anomalies and other epigenetic events, or point mutations that may be responsible for a clinical phenotype. Copy number changes that do not contain any genes or regions with no controlling elements are not reported.
2. Regions smaller than 0.2 Mb are reported only if the region is associated with a known clinical significance.
3. Chromosomal rearrangement: Inversions, balanced insertions, balanced translocations, and certain cases of polyploidy are not detected by CMA.
4. Small sequence changes (point mutations) and/or epigenetic changes are not detected by CMA.
5. The error rate even with the best possible precautions is 2%. The test will not elucidate the chromosomal mechanism of a genetic imbalance.
6. CMA may detect UPD but does not distinguish between UPD caused by heterodisomy or isodisomy.

DISCLAIMER

This CMA test has been developed and validated by Illumina Inc. but has not been approved by the US FDA for diagnostic purposes. Thus this test is recommended for research use only. This is not a diagnostic test and so is not to be considered as a purpose of diagnosis of any diseases. This test is meant for only understanding chromosomal aberrations and their clinical relevance. Clinicians should use their own clinical judgment and not base clinical decisions solely on this document. This report must be given only in the presence of a medical professional to explain the findings and implications. The company will not be liable for any direct, indirect, consequential, special, exemplary, or any other damages. This test detects chromosomal abnormalities only under its limit of resolution. As per the PRENATAL DIAGNOSTIC TECHNIQUES (REGULATIONS AND PREVENTION OF MISUSE) AMENDMENT ACT 2002, sex determination shall not be for all the samples at MAPMYGENOME INDIA LTD

METHODOLOGY

Chromosomal microarray analysis is done using the Illumina Infinium™ Global Screening Array-24 v3.0 BeadChip, which contains ~ 700604 probes for SNP markers, that include exonic, intronic, synonymous, missense, nonsense, mitochondrial, indels, and sex chromosome markers. It can detect copy number variations (CNVs) upto ~2.5Kb with a maximum median marker spacing of 2.53 Kb and covers 245 high-value cytogenetic regions containing ~9000 genes (higher density in ~447 disease-associated genes) commonly screened for cytogenetic abnormalities. Copy number changes containing no genes or found commonly in the general population are not reported. UPD is reported for telomeric regions of LOH 5 Mb in length or interstitial regions of LOH 15 Mb in length. Possible identity by descent is reported when regions of LOH 3 Mb comprise 1.5% of the autosomal genome. Deletions and duplications of 400 kb are reported, even if clinical significance is unclear, as per the provider's request. Smaller pathogenic or likely pathogenic deletions or duplications in regions of known microdeletion/microduplication syndromes or in clinically relevant genes will also be reported. This microarray and associated software (Karyostudio v1.4) is designed by Illumina. DNA for the experiment was isolated from the provided sample using a commercial kit that works on silica membrane-based DNA purification.

The hg19(GRCh37) version of the genome reference is used for the analysis. The laboratory follows the ACMG guidelines (South et. al., Constitutional Microarray Guidelines, Genetics in Medicine, Volume 15, Number 11, November 2013) for reporting Microarray findings. All findings are correlated with clinical history before reporting. All VOUS (variants of unknown significance) are reported if they are found relevant to clinical history. An unrelated pathogenic or likely pathogenic finding is reported if there is sufficient empirical evidence for its involvement in a disorder. MCC is detected by the pattern of SNP markers on the microarray or in selected instances it is also confirmed by performing VNTR-based analysis.

END OF REPORT