CHROMOSOMAL MICROARRAY

CLINICAL HISTORY

I/V/O missed abortion, the POC sample of Mrs. Aishwarya Arangale was referred for chromosomal microarray for genetic evaluation of chromosomal abnormalities.

ARRAY TYPE

Affymetrix CytoScan 315K array

TEST RESULT

- NO SIGNIFICANT COPY NUMBER VARIANTS DETECTED.
- NO SIGNIFICANT REPORTABLE LCSH REGIONS IDENTIFIED.

INTERPRETATION

The cyto microarray analysis revealed a normal karyoview arr (1-22) ×2 with no signi cant pathogenic and likely pathogenic copy number variants. Clinical correlation is recommended.

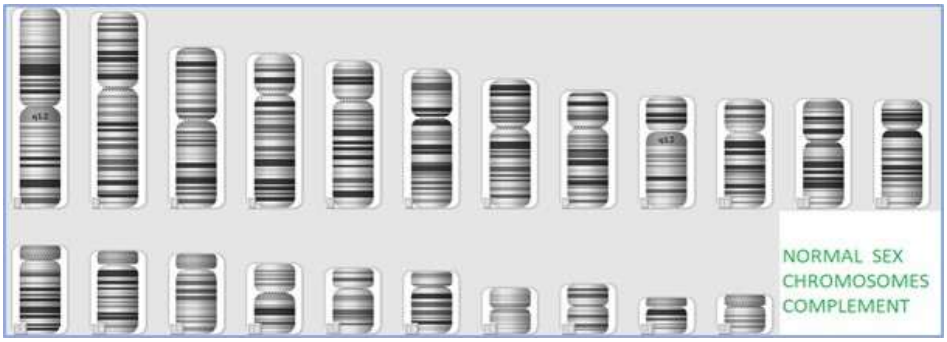

Fig1: Karyoview of sample 11427495

RECOMMENDATIONS

- Genetic Counseling and clinical correlation are strongly recommended.
- Whole Exome sequencing is recommended if there is a strong clinical suspicion of any underlying genetic cause.

PATIENT DETAILS

Reg.No	0662501280003		
Patient Name	POC. AISHWARYA ARANGALE		

TEST METHOD

Chromosomal microarray analysis (CMA) was performed using Affymetrix Cytoscan Optima array. Genomic DNA (250ng) was enzymatically digested and adapter ligated. The ampli ed PCR products of desired size are puri ed using AMP pure beads and fragmented. The fragmented DNA is further labelled to hybridize onto Cytoscan Optima gene chip followed by staining and scanning. Data was analyzed using chromosome Analysis Suite (ChAS) version 4.5 Software developed with ref to Human reference genome (GRCh38).

AMERICAN COLLEGE OF MEDICAL GENETICS (ACMG) GUIDELINES FOR REPORTING

- Copy number changes not containing genes (exclusion: association with controlling elements of the genes with clinical signi cance); regions not containing any controlling elements; and regions smaller than 1 Mb (exception: association of regions with a gene of known clinical signi cance) are not reported. Interpretation of regions of copy neutral LOH (loss of heterozygosity) which can be attributed to uniparental disomy (UPD) or identity by descent(IBD).
- Omit genes related to cancers and late-onset genetic disorders for prenatal/pediatric samples.

LIMITATIONS OF TEST AND OTHER TEST NOTES

Chromosomal Microarray is recommended for the sole purpose of identifying DNA copy number variations (CNVs) associated with chromosomal imbalances and for the detection of absence/loss of heterozygosity (AOH/LOH), regions/runs of homozygosity (ROH), or long contiguous stretches of homozygosity (LCSH).

CMA-ISCA can detect only gross genomic copy number imbalances (aneuploidy, deletions and duplications) and AOH/LCSH in the nuclear genome. It cannot detect balanced chromosomal rearrangements such as inversions, balanced insertions, and reciprocal translocations.

CytoScan® Optima Suite offers minimum resolution of 1 MB for losses, 2 MB for gains, and 5 MB for LOH/AOH and increased coverage density (25 markers/100 kb) in 396 empirically selected regions relevant for prenatal research.

CMA CANNOT DETECT

- Genomic copy number changes in the regions of the genome not represented on the microarray (including regions with repeat sequences such as segmental duplications, repeat sequences in the short arms of acrocentric chromosomes, and heterochromatic regions)
- low levels of mosaicism (<20-25%). point mutations and indels, Imbalances in the mitochondrial genome.
- complete uniparental heterodisomy for the entire chromosome. It can only detect uniparental isodisomy, and segmental heterodisomy. CMA cannot detect imbalances when mosaicism for reciprocal CNVs exist. CMA will fail to detect the presence of the clinically signi cant 45, X line. Failure to detect an alteration at a speci c locus does not rule out the diagnosis of a genetic disorder associated with that locus. Other abnormalities may be present that are undetectable by the microarray design. Failure to detect evidence of UPD/AOH does not exclude the clinical diagnosis of an imprinting or recessive disorder.
- As per the PRE-NATAL DIAGNOSTIC TECHNIQUES (REGULATIONS &PREVENTION OF MISUSE) AMENDMENT ACT2002, sex determination will not be revealed for all pre-conception and prenatal samples.

PATIENT DETAILS

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DISCLAIMER

This CMA test has been developed and validated but has not been approved by the US FDA for diagnostic purposes. Thus, this test is recommended for research use only but not a diagnostic test and hence 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. 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.

REFERENCES

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