Chromosomal Microarray 315K , *PERIPHERAL BLOOD*

Sample Type:	PERIPHERAL BLOOD
Clinical Indication:	
To Rule Out Chromosomal Abnormality	
NEGATIVE No clinically significant copy number variant (CNV) detected.	
Result	arr(X,1-22)x2 Normal chromosome complement
Sample Description:	
Sample quality is optimum for the test. DNA conc.: 168.66 ng/ul	
Karyoview:	

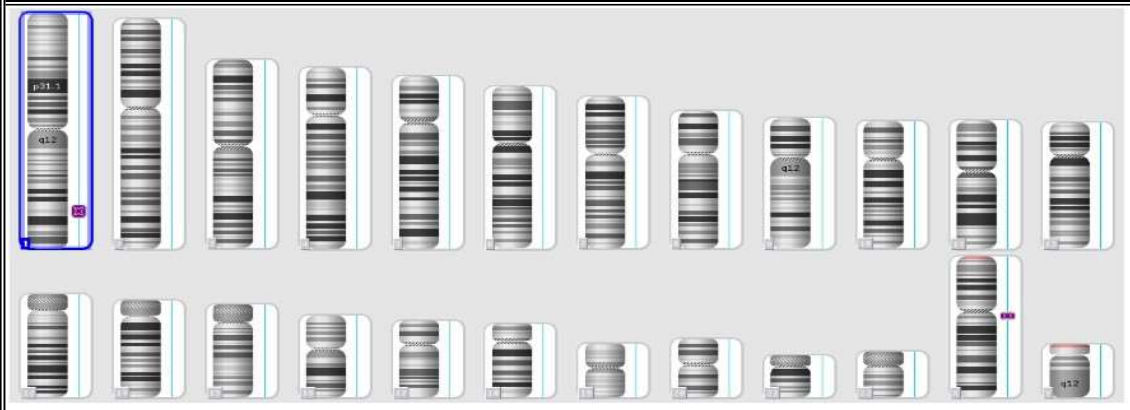

Fig 1: Genomic View of Duplication/Deletion in the analyzed DNA sample

SIN NO.:CG00010958

ADDITIONAL FINDINGS: LOH DETECTED IN THE ANALYSED SAMPLE

LOH

arr[GRCh38] 1q32.1q41(203,164,524_216,026,742)x2 hmz

Test Methodology:

- Chromosomal microarray analysis (CMA) was performed using an Affymetrix CytoScan™ 315K array.
- CytoScan® Optima Array content has been empirically selected from CytoScan® HD Array and consists of a total of

315,608 features covering control, copy number (CN), and single-nucleotide polymorphism (SNP) probes.

- There is a total of 18,018 CN and 148,450 SNP markers uniformly spaced over the genome with enhanced interrogation of 396 regions of prenatal interest. Cumulatively, through the collection of SNPs and nonpolymorphic probes, the application provides the ability to support detection of CNVs, enable the elucidation of allelic imbalance, identify copy number neutral abnormalities such LOH, and characterize unbalanced translocation events in the samples of interest.
- Genomic DNA extracted from Peripheral Blood, Saliva, Amniotic fluid, CVS, POC, Cord Blood or any other standard source is used for further protocol of Affymetrix CytoScanTM Optima 315k assay.
- Data was analyzed using Chromosome Analysis Suite (Hg19).

Limitations:

- CMA is limited to detection of gain or loss of genomic material. It does not detects low level mosaicism (<20%), balanced translocations, inversions or point mutations that may be responsible for the clinical phenotype.
- This assay can detect a minimum resolution of 1 MB for losses, 2 MB for gains, and 5 MB for LOH.
- This assay has increased coverage density (25 markers/100 kb) in 396 empirically selected regions relevant for prenatal research.

Variant classification as per ACMG guidelines:

Variant	A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).
Benign	A variant which is known not to be responsible for disease has been detected. Generally no further action is warranted on such variants when detected.
Likely Benign	A variant which is very unlikely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of Pathogenicity
Pathogenic	A disease causing variation in a gene which can explain the patient's symptoms has been detected. This usually means that a suspected disorder for which testing had been requested has been confirmed.
Likely Pathogenic	A variant which is very likely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity.
Variant of Uncertain Significance	A variant has been detected, but it is difficult to classify it as either pathogenic (disease causing) or benign (non-disease causing) based on current available scientific evidence. Further testing of the patient or family members as recommended by

	your clinician may be needed. It is probable that their significance can be assessed only with time, subject to availability of scientific evidence.

References:

1. Levy B., et al. Genomic imbalance in products of conception: single-nucleotide polymorphism chromosomal microarray analysis. Obstetrics and Gynecology 124(2 Pt 1):202–209 (2014).
2. Wang B. T., et al. Abnormalities in spontaneous abortions detected by G-banding and chromosomal microarray analysis (CMA) at a national reference laboratory. Molecular Cytogenetics 7:33 (2014). eCollection 2014. doi:10.1186/1755-8166-7-33

Disclaimer:

This test was developed and its performance characteristics have been determined by Lupin Diagnostics. Cytogenetic testing generates technical reports and these should not be used as medical certificates without a clinical re- validation and or interpretation of test result by a registered clinician.The microarray is designated according to International system for Human Cytogenomic Nomenclature (ISCN 2020).

*** Marked values are the critical values.**

***** End Of Report *****