

Dr. Vinay Chopra
 MD (Pathology & Microbiology)
 Chairman & Consultant Pathologist

Dr. Yugam Chopra
 MD (Pathology)
 CEO & Consultant Pathologist

LABORATORY REPORT



Name : Mrs RAJKA	Sex/Age : Female/31 Years	Case ID : 40821601997
Ref By :	Dis. Loc. :	Pt ID :
Bill. Loc. : WALK IN SECTOR 62		Pt. Loc. :
Registration Date & Time : 13-Aug-2024 10:34	Sample Type : Product Of Conception Material	Ph # :
Sample Date & Time : 13-Aug-2024 10:34	Sample Coll. By :	Ref Id :
Report Date & Time : 30-Aug-2024 18:51	Acc. Remarks :	Ref Id 2 :

PRODUCT OF CONCEPTION ANEUPLOIDY SCREENING BY NGS

Clinical History

Aneuploidy Screening Analysis requested from POC.

Results

Aneuploidy	Not Detected
Final Result	NORMAL

Note: If there is a high clinical suspicion of the chromosomal abnormalities then a product of conception microarray (RapidSure Constitutional) test is recommended for the presence or absence of micro deletions.

Interpretation

This POC sample contains **46,XX chromosome complement** as depicted in the image below.

For specimens received from non NCGM locations, it is presumed that it belongs to the patient as identified on the labels of the container/Test Requisition Form and it has been verified as per GCLP (Good Clinical Lab Practices) by the referrer at the tPage 1 of 3 collection of the specimen. NCGM's responsibility is limited to the analytical part of the assay performed.

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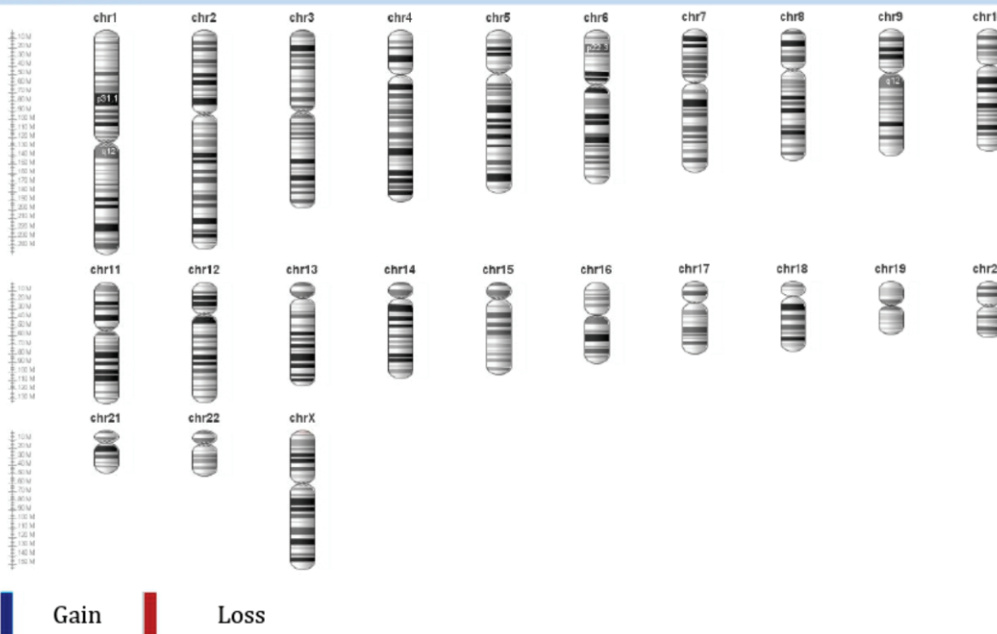
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Karyoview



The karyoview provides details of the analysis of the Samples in a pictorial format using the image of a Karyotype as a template. Please note that these images are only a crude representation of the virtual Karyotype. Balanced translocations and certain other abnormalities cannot be deciphered by this methodology and this may result in an image that is not representative of the actual Karyotype.

Test Information

- This test is performed on product of conception for aneuploidy detection. Samples sent to the lab are first processed through Whole Genome Amplification. After being subject to several Quality Control checks, the sample is processed through a Next Generation Sequencing Workflow by Semiconductor Sequencing. Results are provided in graphical as well as annotated manner. The analysis is based on the human reference genome (GRCh37/hg19).
- The exact Nomenclature can be requested by contacting the lab.

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Limitations

- Although all precautions are taken while conducting these tests, there is a standard error rate of approximate 1% in all genetic tests and this should be taken into consideration before any clinical decision is made on the basis of these reports. The accuracy and quality of the test may be affected by improper sample collection, storage and transportation.
- Although Negative controls are processed with every run, it is strongly advised that routine cleaning be performed using DNAase and RNAase solutions. Any DNA or RNA contamination may lead to erroneous results and Neuberger Center for Genomic Medicine will not be responsible for the reported results in event of such contaminations.
- In cases where any of the QC parameters fails during the process, the referring institute will be informed. Every attempt will be made to ensure the QC checks are within acceptable limits, but in cases where on account of technical difficulties or other causes, this is not possible, testing for those samples will be ceased.
- This test cannot analyze triploidy, balanced translocations, individual gene mutations, low level mosaicism, inversions, translocations, small indels, epigenetic alterations. Also certain high complexity regions, which are likely to be Variants of Unknown Significance, may not be covered.
- In case of Klinefelter Syndrome, mosaic maternal contamination cannot be ruled out.
- The theoretical and proven experimental limit of this assay is to detect a deletion of up to 10 MB. In cases where smaller deletions (<10 MB) are detected, they will be reported on an experimental basis only. Interpretation of the genomic copy number changes which have unknown clinical significance can be complicated to conclude any result.
- This test has been approved by NABL but not approved by CAP or FDA, due to lack of available proficiency testing materials. Its validity has been established by this laboratory and concordance established by using results from standard techniques like Microarray, Karyotyping and available standard DNA.
- The test results should be interpreted only in conjunction with the patient's clinical history and should be interpreted only by a qualified physician.

References

- Xu J, Chen M, Liu QY, et al. Detecting trisomy in products of conception from first-trimester spontaneous miscarriages by next-generation sequencing (Ngs). *Medicine (Baltimore)*. 2020;99(5):e18731. doi: 10.1097/MD.00000000000018731. PMID: 32000376.
- Kato T, Miyai S, Suzuki H, et al. Usefulness of combined NGS and QF-PCR analysis for product of conception karyotyping. *Reprod Med Biol*. 2022;21(1):e12449. doi: 10.1002/rmb2.12449. PMID: 35386384.
- Zhou Y, Xu W, Jiang Y, et al. Clinical utility of a high-resolution melting test for screening numerical chromosomal abnormalities in recurrent pregnancy loss. *The Journal of Molecular Diagnostics*. 2020;22(4):523-531. doi: <https://doi.org/10.1016/j.jmoldx.2020.01.005>.

----- End Of Report -----

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