ALTUM 750K - Chromosomal Microarray Report

Name: VIJETHA CHANABASANAVAR (60247)	Age/Gender: 29 Years/Female
Patient ID: 1002496823	Sample ID: 2400223332
Specimen type: CVS	Sample Quality: Optimum
Referring Doctor: DR.PRATHIMA RADHAKRISHNAN	Referring Center: Bangalore Fetal Medicine Centre
Test: ALTUM 750K - Chromosomal Microarray	Collection Date: 05/11/2024
Receiving Date: 07/11/2024	Reporting Date: 13/11/2024
Indication: Non-consanguineous couple, Primigravida - USG at 13 weeks shows increased NT 2.85 mm at CRL of 66.2 mm, absent/ hypoplastic nasal bone and occasional tricuspid regurgitation. FTS test shows increased risk for Trisomy 21 (1:8).	

SUMMARY

No clinically relevant copy number gains or losses were found in the targeted regions of the given specimen.

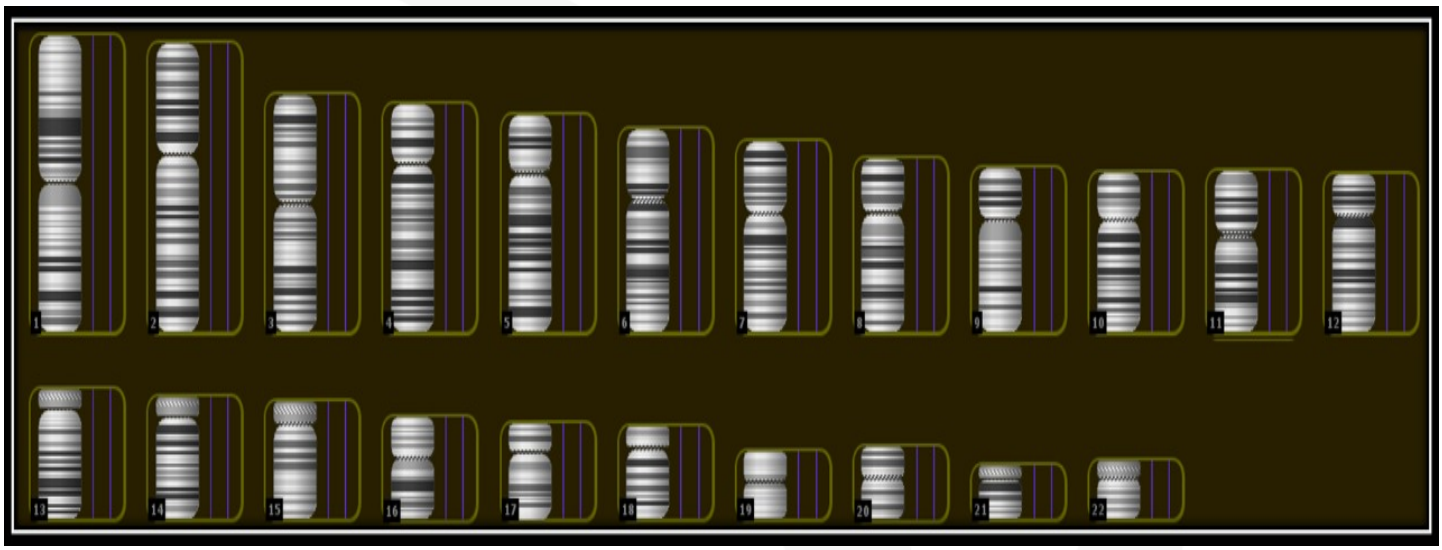
RESULT

Clinically relevant Copy Number Variations: **Not Detected**
SNP Array Result (ISCN 2016): **arr[GRCh38](1-22)×2**

Sex of the fetus has not been disclosed in accordance with the PCPNDT Act 2003

INTERPRETATION

Chromosomal microarray analysis did not reveal any aneuploidy or copy number variations of clinical significance.



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MATERNAL CELL CONTAMINATION

No significant maternal contamination detected on STR analysis with a lower limit of detection of 20%.

Lilac Insights Pvt. Ltd. 301-401, Building A-1, Rupa Solitaire Millennium Business Park, MIDC Industrial Area, Sector-1, Navi Mumbai, Maharashtra 400710.
Phone: **+91 22 41841438**; Website: **www.lilacinsights.com**; For queries or complaints, please email: **info@lilacinsights.com** | CIN - U85191MH2011PTC217513

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RECOMMENDATION

1. Genetic counselling is recommended for further genetic evaluation based on clinical correlation.

TEST METHODOLOGY

This microarray consists of 750K oligonucleotide probes across the genome, including 550K unique non-polymorphic probes, and 200K bi-allelic SNP (single nucleotide polymorphism) probes. The minimum resolution for detection is ~200 kb for losses, ~400 kb for gains and >5 Mb for LOH. However, LOH will be reported depending upon chromosomal location, significance and likelihood of imprinting disorder.

Genomic DNA was digested with Nsp1 and then ligated by Nsp1 adaptor followed by PCR amplification. Amplified PCR products were then purified and fragmented. The fragmented products were labelled w ith biotin and hybridised overnight onto the array. The array was washed using a fluidics station and then scanned on an Affymetrix GeneChip scanner. The data file generated was analysed using Chromosome Analysis Suite (ChAS). The analysis is based on the Human reference genome (GRCh38/hg38). 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.

LIMITATIONS

- The test can only detect gross genomic copy number imbalances (aneuploidy, deletions and duplications) and LOH in the nuclear genome.
- It cannot detect balanced chromosomal rearrangements such as balanced translocations, inversions and balanced insertions.
- Low grade mosaicism (<15%) for chromosomal abnormalities cannot be detected.
- The test cannot detect point mutations. This test detects the chromosomal abnormalities only under its limit of resolution.

DISCLAIMER

- The above analysis is based upon the sample received in the laboratory. This is not a diagnostic test and hence should not be considered as a purpose of diagnosis of any diseases.
- Lilac Insights follows a policy of not reporting Variations of Unknown Clinical Significance (VOUS) for prenatal cases. An unrelated pathogenic or likely pathogenic finding is reported if there is sufficient empirical evidence for its involvement in a disorder.
- Clinical decisions should not be taken solely on the basis of Chromosomal Microarray Results and correlation with other clinical findings like patient history, ultrasound findings is necessary.
- The test was validated and its performance characteristics have been determined by Lilac Insights Pvt. Ltd as required by the ACMG guidelines.
- MCC is tested on the basis of the distribution of SNP markers in this assay or in selected instances it is also confirmed by performing VNTR based analysis.
- Although all precautions are taken during DNA tests the currently available data indicates that the technical error rate for all types of DNA analysis is approximately 2%. There is a slight chance of failure due to degraded DNA or contaminated sample.
- Each sample received at Lilac Insights' processing centre is handled with the utmost sensitivity and care. All samples received on Sundays and National holidays are stored as per specific guidelines for the respective specimens and processed on the next day.

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REFERENCES

1. South *et. al.* , Constitutional Microarray Guidelines, Genetics in medicine, Volume 15, Number 11, November 2013.
2. CytoScan Suite- Data sheet.

Checked by
Dr. Ashish C Trivedi, PhD
Sr. Scientific officer, Level 5

Verified by
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Head Clinical Cytogenetics

- End of Report -

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