

Patient NAME :		Report STATUS :	
DOB/Age/Gender :		Barcode NO :	
Patient ID / UHID :		Sample Type :	
Referred BY :		Report Date :	
Sample Collected :			

## Karyotyping + FISH (5 Probe)

### Chromosome Analysis, Amniotic Fluid

**CLINICAL INDICATION** To rule out chromosome abnormality

**SUMMARY OF RESULTS** **NORMAL KARYOTYPE**

**NOMENCLATURE** **46,--**  
**Sex of the fetus not revealed as per PNDT act.**  
*(As per International System for Human Cytogenomic Nomenclature, ISCN,2020)*

### **CLINICAL INTERPRETATION**

Within the limits of standard cytogenetic methodologies, the chromosomes of the patient showed normal karyotype G-banding patterns with no evidence of aneuploidy or without apparent structural abnormality or rearrangement. The following possibilities, although rare, cannot be ruled out: a) low level mosaicism, b) very subtle rearrangements, c) genetic disorders that cannot be detected beyond the resolution of by standard cytogenetic methods. d) There is a possibility of technical error (2%) in absence of clinical history and sub-optimal quality of sample.

**RECOMMENDATION** Genetic Counseling for the family is recommended.

**SAMPLE DESCRIPTION** The fibroblast culture was setup in AmnioMAX-II (Gibco) tissue culture media in 5% CO<sub>2</sub>. The sample was of optimal quality for conventional cytogenetics culture techniques.

**Disclaimer:-** As per Pre-Natal Diagnostic Techniques (Regulation and prevention of Misuse) Act, 1994", our labs strictly does not determine the sex of the fetus.

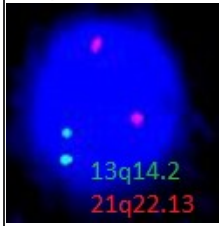


**Dr. Ankur Jindal (Ph.D)**  
**Consultant Cytogenomics**

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**Aneuploidy Five Probes (13, 18, 21, X & Y) By FISH**

**CLINICAL DIAGNOSIS:** Previous two babies have congenital anomaly.  
**METHODOLOGY/KIT :** Interphase Fluorescence in-situ Hybridization (FISH) method; Multicolor DNA Probe Kit for Aneuploidy Detection of Chromosome 13, 18, 21 & X and Y.  
 13q14.2 /21q22.13      cep 18p11.1



First Hybridization



Second Hybridization

**RESULT SUMMARY:** NO ANEUPLOIDY DETECTED for chromosome 13, 18, 21 and Sex chromosomes.(FISH is not intended for use as a standalone test for making clinical decisions)

**NOMENCALTURE:** nuc ish 13q14(RB1X2),18p11.1-q11.1(D18X1X2),21q22.13q22.2(D21S259X2,D21S341X2,D21S342X2) [50].(As per International System of Human Cytogenomic Nomenclature, ISCN, 2020) [Sex of Fetus not Revealed as per PNDT Act]

**INTERPRETATION:** The case is considered as informative NORMAL (NEGATIVE) with normal/disomic signal pattern for chromosome 13, 18, 21 and sex chromosomes observed in more than 95% of the total cells screened.

**Interpretation Guidelines:** Informative Normal: A case is classified as informative normal for specific chromosomes (autosomes/sex chromosome) if more than 90% of the cells showed normal disomic signal pattern.Informative Abnormal: A case is classified as informative abnormal if more than 60% of the nuclei show aberrant/abnormal signal pattern for specific chromosomes (autosomes/sex chromosome)Uninformative: A case is considered as uninformative whenever 10-60% of nuclei shows aberrant signals. In these case additional 200 nuclei are scored and conclusive interpretable results are obtained after routine cytogenetics analysis on more than 20 metaphase **Intended Use:** The AneuVysion (Vysis CEP 18, X, Y alpha satellite LSI 13 and 21) Multicolor probe panel is intended to use CEP18/X/Y probe to detect alpha satellite sequences in the centromere regions of chromosome 18, C and Y and LSI 13/21 probe to detect the 13q14 region and 21q22.13 and 21q22.2 region. The AneuVysion Kit is indicated for identifying and enumerating chromosome 13, 18, 21 X and Y via FISH in metaphase cells and interphase nuclei obtained from amniotic fluid in subjects with presumed high-risk pregnancies. It is not intended to be used as a standalone assay for making clinical decision. FISH Assay is a prenatal genetic test to be used in conjunction with fetal karyotype analysis to provide detection on trisomy 13, 18, 21 (Down's Syndrome) and sex chromosome abnormalities (such as Klinefelter and Turner syndromes).

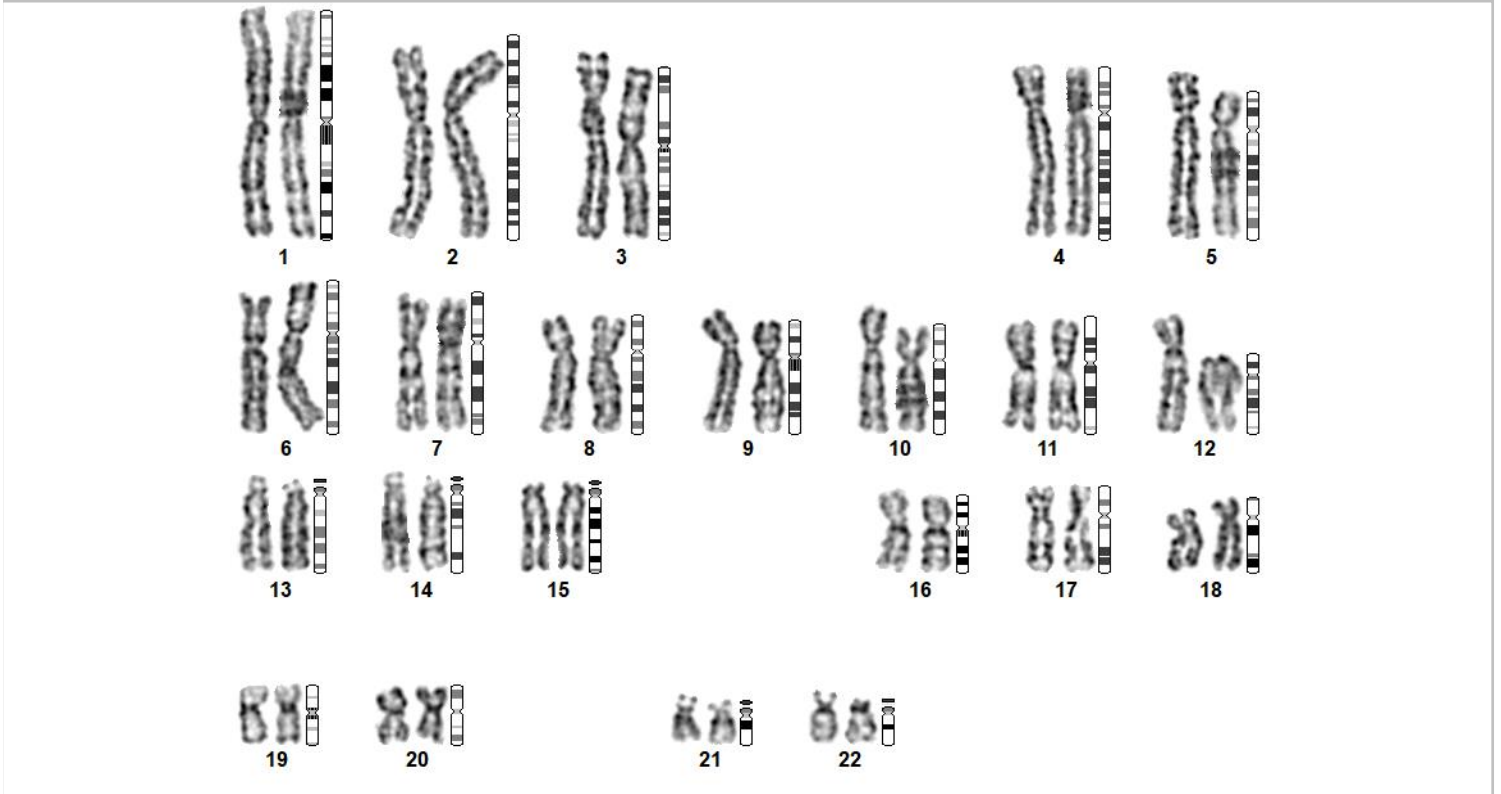
**Limitation of Test:**

1. FISH is used to quickly rule out the common numerical chromosomal abnormalities i.e., Trisomy 13, 18, 21 and sex chromosomes disorder in rapid turn-around time. The FISH test does not provide information about any chromosome other than loci mentioned in this report and kit insert
2. No other numerical and structural chromosome abnormalities like translocations, deletions and abnormalities of other chromosomes cannot be ruled out by FISH. Reliability of the FISH test report is 99%.
3. A negative result does not exclude the presence of chromosome alterations other than the one screened for and imposes
4. The FISH results should be further verified and confirmatory gold standard karyotype test diagnostic tests through conventional culture techniques or through a DNA based chromosome microarray test.
5. The report relates only to the specimen submitted to the lab which was verified and confirmed at the time of specimen collection.

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

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KARYOTYPE IMAGE:



REHANA

Karyotype: 46,...

Barcode No:

**METHOD:** G-BANDING  
**Metaphase Counted:** 20  
**Metaphase Analyzed:** 10  
**Metaphase Karyotyped:** 10  
**Banding Resolution:** 675  
**Metaphase Quality:** Good

*Vikas*

**Mr. Vikas Dixit**  
**(Sr. Cytogeneticist)**  
 Cytogenetics

Reviewed and Signed out on:

**Disclaimer: Sex of Fetus not revealed as per PNDT Act**

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