Booking Date	29/06/2023	Patient ID 012306290220	Printed on 12/07/2023
Name	Rajbala	Maternal Age 34 Years	Sex - Female
Ref By	Atlas Hospital Palwal		

DIAGNOSIS: Abnormal by FISH only

METHODOLOGY: Fluorescence *in situ* Hybridization (FISH)

PROBE NAME: AneuVysion (Abbott Mol., Inc.)

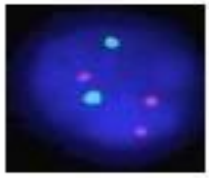
ICSN: nucish (DXZ1x-, DYZ3x-, D18Z1x2),(RB1x2),(D21S259/D21S341/D21S342)x3

Fluorescence *In Situ* Hybridization (FISH) on uncultured cells was performed using probes specific for chromosomes 13, 18, 21, X and Y due to karyotype culture failure.

INTERPRETATION:

Fluorescence In Situ Hybridization (FISH) on uncultured cells showed no evidence of aneuploidy for chromosomes 13 and 18 in this specimen, and normal sex chromosome complement. Three copies of chromosome 21 were observed in all the cells analyzed, which is consistent with Trisomy 21 in this specimen. Trisomy is found in approximately 40% of spontaneous abortions. However, birth defects due to submicroscopic chromosomal rearrangements, low level mosaicism, or maternal cell contamination, as well as other genetic disorders not detected by this test, cannot be ruled out.

FISH:



Interphase cell showing two copies of chromosome 13 (green) and three copies of chromosome 21 (orange).

Interphase cell showing two copies of chromosome 18 (Aqua).

RECOMMENDATION:

Chromosomal Microarray Test on the fetal DNA is recommended to rule out small copy number variants (microdeletions and duplication) which cannot be detected by FISH or karyotype. Genetic counselling is recommended.

Please Note: Interphase analysis may not detect structural abnormalities for the chromosomes tested. In addition, chromosome abnormalities from other regions of the genome, which do not involve the probes tested, cannot be detected by this FISH analysis. Failure to detect an aneuploidy for the chromosomes tested does not exclude the diagnosis of other chromosome abnormalities and any other genetic disorders.

Although the methodology used in this analysis and interpretation is highly accurate, it does not detect small rearrangements and very low-level mosaicism, which are detectable only by molecular methods. Failure to detect an alteration at any locus does not exclude the diagnosis of any of the disorders.