

Booking Date 29/06/2023  
Name Rajbala  
Ref By Atlas Hospital Palwal

Patient ID 012306290220  
Maternal Age 34 Years

Printed on 11/07/2023  
Sex - Female

**DIAGNOSIS:** Abnormal by FISH only

**METHODOLOGY:** Fluorescence *in situ* Hybridization (FISH)

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

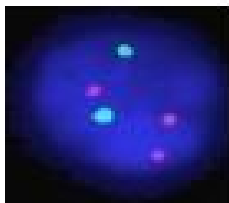
**ICSN:** nucish (DXZ1x-, DYZ3x-, D18Z1x2),(RB1x2),(D21S259/D21S341/D21S342)x3

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.

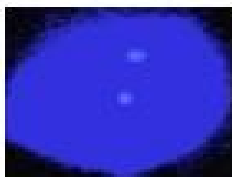
## INTERPRETATION:

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:

Genetic counselling is recommended.

**Please Note:** 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.