

Booking Date 20/09/2024

Patient ID : 012409200333

Printed on 07/10/2024

Name Janvi

Maternal Age : 23 Years

Sex - Female

Ref By RLC Path Lab

DIAGNOSIS: Normal by FISH only

METHODOLOGY: Fluorescence *in situ* Hybridization (FISH)

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

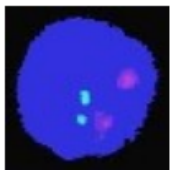
ICSN: nucish (DXZ1x-, DYZ3x-, D18Z1x2),(RB1,D21S259/D21S341/D21S342)x2

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:

There is no evidence of aneuploidy for chromosomes 13, 18, 21 and sex chromosomes by FISH in the fetus. This FISH analysis provides information only on aneuploidy for the chromosomes tested. This test does not detect abnormalities of all other chromosomes or regions not targeted by the probe panel. This probe set detects most common aneuploidies observed in live births. 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 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.