

Booking Date 19/02/2024

Patient ID 012402170044

Printed on 06/03/2024

Name RITU SHRESHTHI

Maternal Age 32 Years

Sex - Female

Ref By Shivam Hospital

**DIAGNOSIS:** Abnormal 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

This fetal tissue failed to grow successfully in culture, therefore karyotype was not possible. Fluorescence In Situ Hybridization (FISH) on uncultured cells was performed using probes specific for chromosomes 13, 18, 21, X and Y.

## INTERPRETATION:

There is no evidence of aneuploidy for chromosomes 13, 18 and 21 in this specimen. This specimen **showed monosomy X** sex chromosome complement in all the cells analyzed. **Monosomy X**, is associated with the features of Turner Syndrome in live births and is found in approximately 7% of spontaneous abortions.

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.

## FISH:



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



Interphase cell showing two copies of chromosome 18 (Aqua) and one copy of chromosome X (green).

## 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.