Fluorescent in situ Hybridization (FISH)   [ISCN2020]

Sample Type	Clinical Indication	Test Requested	Method
Peripheral blood	Not Provided	FISH for chromosome 21	FISH
Sample Quality	**Probe Details**	**Ref Range**	**No Of cells**
Good	SPEC(Wuhan Health care) FISH for five markers (13and 21 chromosomes)	chromosome 21 = 3% chromosome 13 = 2%	50

Results Table

S.No.	Result (ISCN2020)	Chromosome Loci/Color	No. of Cells	Result
1	nuc ish (RB1)x2,(D21S1917/D21S341/D21S 339)x3	Red (chromosome 21) - 3 Green (chromosome 13) - 2	50/50	Positive for Trisomy 21

FISH Images
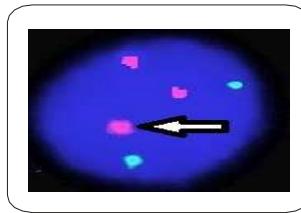
LSI 21q22.1-q22.2- spectrum Red
LSI 13q34.2 (RB1 gene)-spectrum

INTERPRETATION

Fluorescence In Situ Hybridization (FISH) of direct harvesting of peripheral blood sample revealed the presence of an additional copy of chromosome 21, suggestive of trisomy 21(Down Syndrome) and no evidence of aneuploidy of chromosome 13 in any of the interphase cells scored.

References:

1. An International System for Human Cytogenetic Nomenclature (2020). Karger Publishers.
2. Human Cytogenetics: Constitutional Analysis. A Practical Approach. Third Edition, Edited by Denise Rooney.

Note: Kindly note that aneuploidy of other chromosomes, structural abnormalities have not been ruled out by this analysis.

Disclaimer

1. FISH is a spatial detection and quantification of cellular DNA only and not meant for cell free DNA.
2. FISH is limited to labeling of the complete set of 24 different chromosomes and derived from the 24 chromosomes, and detection of structural and numerical aberrations.
3. The clinical interpretation of any test results is made in conjunction with other diagnostic procedures and patient medical history.
4. This test is not going to provide information about any other chromosomes /loci not listed in the report.
5. A negative report does not exclude the presence of other chromosomal alterations other than that tested.
6. FISH results should be evaluated in the context of the patient's medical history and other lab reports.