Mrs. SONIA
AMBALA, AMBALA
Tel No : 8607344999
PIN No: 133001
PID NO: P542100754744
Age: 34.0 Year(s) Sex: Female

Reference: Dr.VINAY CHOPRA

Sample Collected At:
Dr vinay kumar chopra
Kos diagnostic lab, 6349/i, nicholson road, ambala cantt, hry 133001.
PROCESSING LOCATION:- Metropolis Healthcare Ltd, Unit No. 409- 416, 4th Floor, Commercial Building-1, Kohinoor Mall, Mumbai-70

VID: 54213320015904
Registered On:
10/11/2021 04:03 PM
Collected On:
12/11/2021 3:03AM
Reported On:
27/11/2021 08:53 PM

Karyotyping by G-Banding Reflex FISH (Chr 13,18, 21, X and Y), POC

INTERNAL LAB NO.	2672-21-F
CULTURE METHOD	Short term and long term fibroblast cultures
BANDING METHOD	GTG-Banding with Trypsin & Giemsa Stain
INTERPRETATION	Cytogenetic analysis with short and long planned cultures could not yield any analyzable metaphases probably due to contamination causing lack of spontaneous mitosis.
COMMENTS	FISH studies carried out to rule out aneuploidy of chromosomes 13, 18, 21, X and Y.

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Trisomy for Chromosome 13, 18, 21, X, Y By FISH POC

INTERNAL LAB NO. : 2672-21-F

SPECIMEN : Products of conception

FISH INVESTIGATION FOR : Aneuploidy detection of chromosome 13,18,21, X and Y.

METHOD : Fluorescence in situ hybridization (FISH) was performed using fluorescent probes on cells obtained from short planned cultures. The analysis was done on an Olympus BX43 fluorescent microscope with appropriate filters using the Applied Spectral Imaging Software.

PROBE(S) USED : ZytoLight Aneuploidy Panel SPEC 13/CEN 18/SPEC 21/ CEN X/Y.

RESULTS:

TEST	RESULT
CEP 18/X/Y	Normal
LSI 13/21	Abnormal

INTERPRETATION : FISH studies revealed Trisomy for chromosome 21 and normal diploid status for chromosomes 13, 18 and Sex chromosome (XY) in 100% of the cells analysed.

FISH RECOMMENDATION(S) : Kindly correlate clinically.
If there is any other significant contributory family/medical/obstetric history couple Karyotype (if not done) is suggested. For any queries please feel free to contact at Department of Medical Genetics on 022-50560767.

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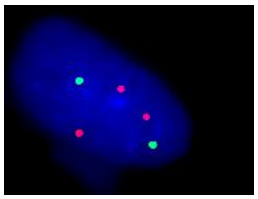
RESULTS:

Probe Name: LSI 13/21

The probe hybridizes to chromosome region 13q14 (Green) and 21q22.13-22.2 (Orange) with a normal signal pattern of two orange and two green, and 3 green or 3 orange in cells with trisomy of 13 and 21 respectively.

SIGNAL PATTERN	NO.OF CELLS	RESULT TYPE
3O2G	50	Abnormal
	Signal pattern showing 3 Orange, 2 Green signals indicative of Trisomy for chromosome 21 and normal diploid status for chromosomes 13.	

O=orange (chromosme 21 signal); G=green (chromosome 13 signal)

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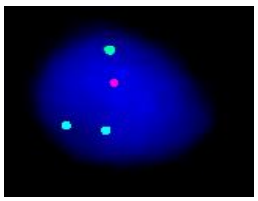
Probe Name: CEP 18/X/Y

CEP 18 (Aqua) / X (Green) / Y (orange) probe hybridizes to centromeric regions of chromosomes 18, X and Y and aid in identification and enumeration of these chromosomes. Apart from the signal patterns mentioned, other variant patterns may also be observed.

SIGNAL PATTERN	NO.OF CELLS	RESULT TYPE
2A1O1G	50	Normal
	Signal pattern showing 2 Aqua, 1 Orange, 1 Green signals indicative of normal diploid status for chromosome 18 and sex chromosome (XY).	

O=orange (chromosme Y signal); G=green (chromosome X signal); A= aqua (chromosome 18 signal)

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Note:

- FISH is performed on the tissue that has been provided to the laboratory.
- FISH is used as an adjunct to conventional karyotyping and rules out the most common chromosomal abnormalities i.e. Trisomy of chromosomes 13, 18, 21 and numerical sex chromosome disorders.The accuracy of this test is 99%.
- The report should be correlated with the clinical history and routine fetal scan by USG.
- The results are not to be used as sole means for clinical diagnosis or patient management decisions.
- Whenever the FISH results show presence of XX sex chromosome pattern then there is likely possibility of presence of maternal decidua. Possibility Of placental mosaicism/ maternal contamination though small cannot be ruled out. Maternal cell populations may be increased in culture and therefore result in misdiagnosis (Rodgers et al, 1996).

Test Indication: Aneuploidy detection by FISH on products of conception sample is indicated in cases of bad obstetric history, recurrent pregnancy loss, abnormal prenatal diagnosis results, and/or abnormal USG findings or as deemed by the referring physician.

Cut Off Values: The cut off for numerical and structural abnormalities for chromosomes 13, 18, 21 and sex chromosomes in normal individuals is 5%.

Limitation of Assay:

- FISH is a rapid and precise molecular diagnostic technique that identifies only probe specific numericaldisorders.
- Structural abnormalities like translocations, deletions, duplications and abnormalities of other chromosomes, single gene disorders related to birth defect cannot be ruled out by FISH.
- Turbid and contaminated samples, samples with very low cell count and poor cell viability may yield either poor quality hybridization or unsuccessful test results.

References:

- Human Chromosome Preparation- Essential Techniques, Ed:- Rooney D E and Czepulkowski B H, 1997.
- Rodgers CS, Creasy MR, Fitchett M, Maliszewska CT, Pratt NR, Waters JJ. Solid tissue culture for cytogenetic analysis: a collaborative survey for the Association of Clinical Cytogeneticists. J ClinPathol (1996); Aug;49(8):638-41.
- Zhang T, Sun Y, Chen Z, Li T. Traditional and molecular chromosomal abnormality analysis of products of conception in spontaneous and recurrent miscarriage. BJOG (2018);125:414–420.
- Shearer BM, Thorland EC, Carlson AW, Jalal SM, Ketterling RP. Reflex fluorescent in situ hybridization testing for unsuccessful product of conception cultures: a retrospective analysis of 5555 samples attempted by conventional cytogenetics and fluorescent in situ hybridization. Genet Med (2011)Jun;13(6):545-52.

-- End of Report --

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