

## **KOS Diagnostic Lab** (A Unit of KOS Healthcare)



Dr. Vinay Chopra MD (Pathology & Microbiology) Chairman & Consultant Pathologist

Dr. Yugam Chopra MD (Pathology) CEO & Consultant Pathologist



Mrs. LATIKA CHOPRA

AMBALA Ambala City H.O.. 9874474455 Tel No: PIN No:

134003 PID NO: P39423518377887

Age: 34 Year(s) Sex: Female Reference: SELF

Sample Collected At:

Dr Vinay Kumar Chopra Kos Diagnostic Lab, 6349/i, Nicholson Road, Ambala Cantt, Hry 133001. Processing Location: - Metropolis Healthcare Ltd, Unit No409-416, 4th Floor, Commercial Building-1, Kohinoor

Mall, Mumbai-70

VID: 230333504365817

Registered On: 08/02/2024 08:15 PM Collected On: 08/02/2024 8:11PM Reported On: 16/02/2024 09:37 PM



FISH for Aneuploidy in POC - Chromosomes 13,18,21, X,Y

INTERNAL LAB NO.

392-24-F

Tests marked with NABL symbol are accredited by NABL vide Certificate no MC-2139; Validity till 01-06-2024

Dr. Talat Khan MD PATHOLOGY

Ms. Gauri Pradhan

Sr Analyst, Cytogenetics & FISH Medical Genetics, Metropolis-Mumbai.

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NOTE:

This Sample was outsourced



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

INTERNAL LAB NO.

**SPECIMEN** 

**FISH INVESTIGATION FOR** 

**METHOD** 

: 392-24-F

: Products of conception

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

: 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.

ZytoLight Aneuploidy Panel SPEC 13/CEN 18/SPEC 21/ CEN

RESULTS:	
TEST	RESULT
CEP 18/X/Y	Normal
LSI 13/21	Normal

#### INTERPRETATION

PROBE(S) USED

FISH RECOMMENDATION(S)

- : FISH studies revealed Normal diploid status for chromosomes 13, 18, 21 and Sex chromosome (XX) in 100% of the cells analysed.
- : Kindly correlate clinically. In view of history provided and present findings, couple Karyotype (if not done) is suggested. For any queries please feel free to contact at grlsupport@metropolisindia.com.



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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
202G	50	Normal
Signal pattern showing 2 Orange, 2 Green signals indicative of normal diploid status for chromosomes 13 and 21.		

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
2A2G	50	Normal
	Signal pattern showing 2 Aqua, 2 Green signals indicative of normal diploid status for chromosome 18 and sex chromosome (XX).	

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 numerical disorders.
- 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.

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-- End of Report --

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KOS Central Lab: 6349/1, Nicholson Road, Ambala Cantt -133 001, Haryana

KOS Molecular Lab: IInd Floor, Parry Hotel, Staff Road, Opp. GPO, Ambala Cantt -133 001, Haryana