To: Petals Women Clinic-Rawdan Street
Sanitas Healthcare and Diagnostics Pvt Ltd
125, Hussainpur Main Road, Madurdaha,
Hussaipur, Kolkata
West Bengal
Kolkata - 700107
Contact:
Report Of: Mrs. PAYEL AGARWAL
Pt. Contact:

 Sample ID
 2390016758

 Patient ID
 10023124658

 Collected on
 05/01/2024

 Received on
 06/01/2024 10:50

 Registered on
 06/01/2024 13:55

 Reported on
 09/01/2024 10:01

 Referred by
 DR.SHAMIM KHANDAKER

QF-PCR 13,18,21,X,Y

Patient Name: Mrs. PAYEL AGARWAL
Sex: FEMALE
Age: 39 yrs

Referral Reason: Intermediate risk for Down's syndrome (1:294)

Result Normal

		Analysis Summary	
	Chromosome 21	No chromosomal abnormalities detected.	
			Sex Chromosomes
	Chromosome 18	No chromosomal abnormalities detected.	No chromosomal abnormalities detected.
	Chromosome 13	No chromosomal abnormalities detected.	
Maternal Cel	9	nificant maternal cell contamination was detected in t imit of detection of 10%.	this specimen on PCR-based VNTR/ STR analysis with

Note: Prenatal sex of the fetus cannot be revealed due to central government 2003 (PC-PNDT) act on prenatal diagnosis.

QF-PCR : Quantitative Fluorescent PCR (QF-PCR)

Dr. Ishpreet K Biji Sr. Scientific Officer Dr. Madhavi Pusalkar Technical Head (GM) - Genomics

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Patient Name: Mrs. PAYEL AGARWAL Patient ID: 10023124658

# **Test Methodology**

Fetal DNA is isolated using standard protocol and tested for Chromosomes 13, 18, 21, X and Y aneuploidies by Quantitative Fluorescent PCR. The assay involves amplification of markers using fluorescently labeled primers, followed by size separation and allele peak measurement on a genetic analyzer. The fragments are analyzed using Gene mapper software. Maternal Cell contamination is analyzed by VNTR or STR analysis on peripheral blood and fetal sample.

### **Background Information**

Prenatal diagnosis of chromosomal abnormalities can help prevent the birth of abnormal children. Quantitative Fluorencence Polymerase Chain Reaction (QF-PCR) is a PCR based rapid, cost-effective and sensitive detection technique for detecting chromosomal abnormalities of the most common chromosomal aneuploidies in chromosomes 13, 18 and 21, alongwith X & Y. QF-PCR employs detection of short tandem repeat (STR) markers that varies in either dosage or fragment size and is ususally analyzed on a genetic analyzer machine. The sensitivity of the technique is very high.

#### Disclaimer

This report is based on the sample received in the Lilac Insights laboratory; the analysis is based on the assumption that samples received are representative of the patient mentioned on the test requisition form and the sample. When samples are received from various referral centers, it is presumed that patient demographics are verified at the point of sample collection. Despite all the necessary precautions and stringency adopted whilst performing DNA tests, the currently available data indicates that the technical error rate associated with all types of DNA analysis, is approximately <2%. It is important that all clinicians or persons requesting DNA diagnostic tests are aware of these data before acting upon these results. As with all diagnostic tests, the laboratory report must be interpreted in conjunction with the presenting clinical profile of the patient and evaluation of all reports.

In accordance to the Pre-Conception and Pre-Natal Diagnostic Testing (PCPNDT) Act, 1994- Govt. of India; Lilac Insights Pvt. Ltd. does not disclose the gender of the fetus. (PC-PNDT Registration No.: NMMC/PNDT/168)

### **Test Limitations**

The QF-PCR assay does not quantify sequences other than the tested chromosomes. The test may not detect small segment imbalance and mosaicism for chromosomes tested. The assay may not reflect the fetal chromosome constitution in case of samples contaminated with maternal cells. The limit of detection of chromosomal mosaicism is 20%. Specimens may contain PCR-inhibitors which can inhibit DNA polymerases as well as primer annealing, preventing amplification of the target sequence and the consequence is that the aberration is not detected.

# References:

1. Mann K, Donaghue C, Fox S P, Docherty Z and Ogilvie C M 2004. Strategies for the rapid diagnosis of chromosome aneuploidy. E J Hum Genet 12: 907-915

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