Laboratory Investigation Report

Patient Name	Centre
Age/Gender	OP/IP No
Max ID/Mobile	Collection Date/Time
Lab ID	Receiving Date
Ref Doctor	Reporting Date/Time
Passport No.	

NON INVASIVE PRENATAL ANEUPLOIDIES AND MICRODELETION SCREENING

1. SCREENING RESULTS

Chromosomes	Risk	Z score	Test Results	Reference interval
Chromosome 21		0.60	Low Risk	-6<Z score<2.8
Chromosome 18		0.32	Low Risk	-6<Z score<2.8
Chromosome 13		4.5	High Risk – Further Investigation Recommended	-6<Z score<2.8
Other Chromosomes		Part II	Low Risk	-6<Z score<6
Microdeletion Syndromes		Part III	Low Risk	-4<Z score

FETAL FRACTION - 18%

Sex Chromosome Aneuploidies	Risk	Test Results	Remarks
SCA		Low Risk	As per PCPNDT act, sex chromosomal aneuploidies will only be provided in case an aneuploidy is detected.

Low Risk Group

Borderline Group

High Risk Group

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2. OTHER CHROMOSOMES

Chromosome		Risk	Z score	Test Results	Reference interval
Chromosome 1			0.41	Low Risk	-6<Z score<6
Chromosome 2			0.10	Low Risk	-6<Z score<6
Chromosome 3			0.22	Low Risk	-6<Z score<6
Chromosome 4			0.35	Low Risk	-6<Z score<6
Chromosome 5			0.11	Low Risk	-6<Z score<6
Chromosome 6			0.26	Low Risk	-6<Z score<6
Chromosome 7			0.16	Low Risk	-6<Z score<6
Chromosome 8			0.38	Low Risk	-6<Z score<6
Chromosome 9			0.05	Low Risk	-6<Z score<6
Chromosome 10			0.19	Low Risk	-6<Z score<6
Chromosome 11			0.28	Low Risk	-6<Z score<6
Chromosome 12			0.35	Low Risk	-6<Z score<6
Chromosome 14			0.13	Low Risk	-6<Z score<6
Chromosome 15			0.32	Low Risk	-6<Z score<6
Chromosome 16			0.26	Low Risk	-6<Z score<6
Chromosome 17			0.09	Low Risk	-6<Z score<6
Chromosome 19			0.19	Low Risk	-6<Z score<6
Chromosome 20			0.27	Low Risk	-6<Z score<6
Chromosome 22			0.34	Low Risk	-6<Z score<6

SIN [redacted] Test Performed at :910 - Max Hospital - Saket M S S H, Press Enclave Road, Mandir Marg, Saket, New Delhi, Delhi 110017

Booking Centre :794 - Max Hospital - Vaishali, W-3, Sector-1, Vaishali, Ghaziabad-201012, U.P, 0120418800

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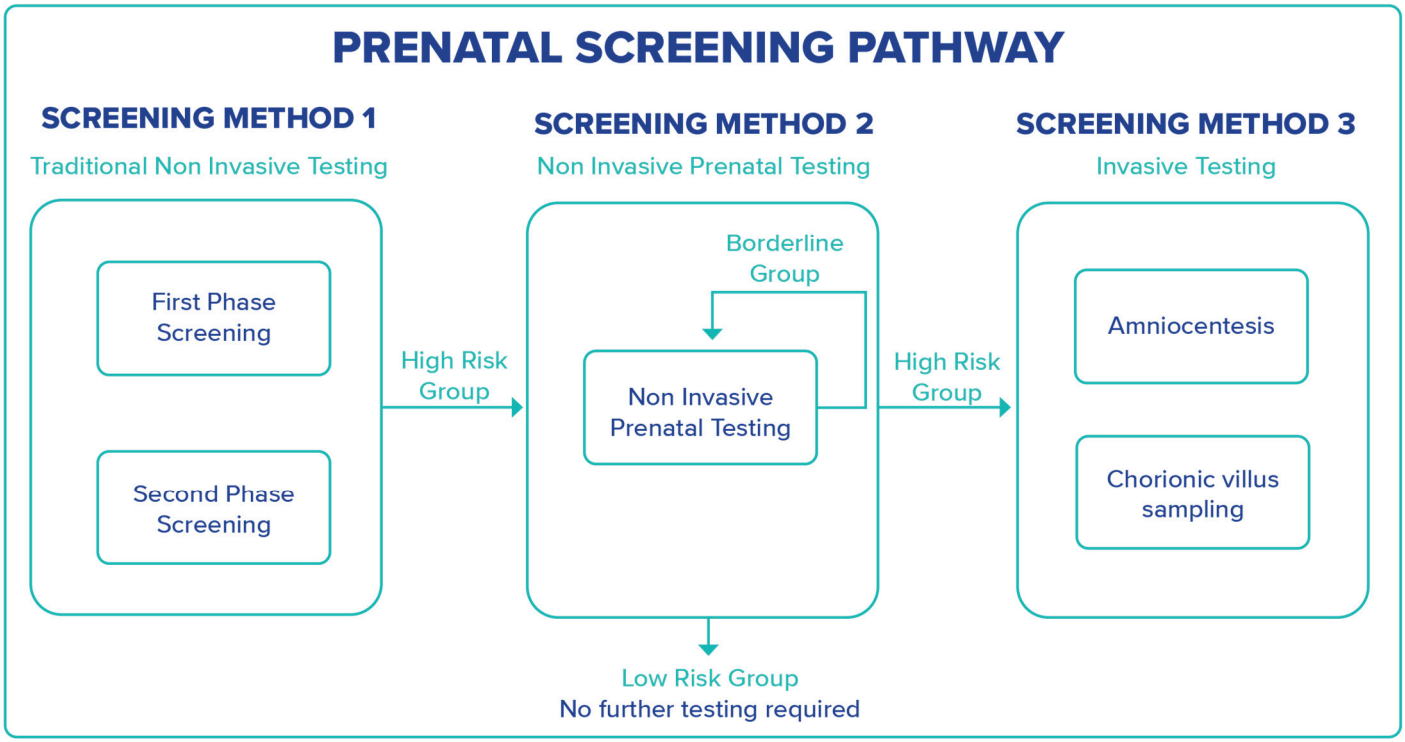
3. MICRODELETION SYNDROMES

Microdeletion Syndromes	Risk	Z score	Test Results	Reference interval
DiGeorge syndrome		0.34	Low Risk	-4<Z score
1p36 deletion syndrome		0.21	Low Risk	-4<Z score
Angelman syndrome / Prader-Willi syndrome		0.18	Low Risk	-4<Z score
Cri-du-Chat syndrome		0.22	Low Risk	-4<Z score
Wolf-Hirschhorn syndrome		0.36	Low Risk	-4<Z score



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Test Methodology

1. The NIPS test screens a maternal blood sample for chromosome aneuploidy in placental DNA using the following methodology:
   (1) Extraction of cell-free placental DNA from the maternal blood sample
   (2) High throughput sequencing of the extracted cell-free placental DNA
   (3) Calculation of molecular mass of placental DNA in all chromosomes
2. The method is intended for use in pregnant women who are at least 10 weeks of pregnancy. The method is suitable for both singleton and twin pregnancies. The accuracy may be slightly lower in twin pregnancies due to multiple sources of fetal DNA.
3. Based on the scope, the NIPS test can detect the following:
   Whole Genome - 23 pairs of human chromosomes
   Microdeletions - 5 specific disorders including: DiGeorge syndrome, 1p36 deletion syndrome, Angelman syndrome/Prader-Willi syndrome, Cri-du-Chat syndrome and Wolf-Hirschhorn syndrome
4 The test is capable of genome-wide aneuploidy detection over the whole fetal genome and gives the results for 23 pairs of chromosomes. This test confers an accuracy of up to 99% on the detection of fetal aneuploidy for chromosomes 13, 18 and 21.

Results are indicated for screening, NOT diagnosis. – (Results should be reviewed and discussed with your healthcare provider.)

Limitations of the Test

Non invasive prenatal is a screening test and all high-risk results should be confirmed through further investigation which may include tests such as amniocentesis or Chorionic Villus Sampling (CVS). Pregnant women with a high-risk result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results. Pregnant women with a negative test result do not ensure an unaffected pregnancy. While results of this testing are highly accurate, not all chromosomal abnormalities may be detected due to placental, maternal or fetal mosaicism, or other causes (micro-deletions, chromosome re-arrangements, translocations, inversions, unbalanced translocations, uniparental disomy). The test is not reportable for known multiple gestations, or if the gestational age is less than 10 weeks.

References:

1. Obstet Gynecol 2012;119:890-901.
2. BMJ 2011;342:c7401.
3. Prenat Diagn 2012;32:c7401.
4.ACOG/SMFM Joint Committee Opinion No. 545, Dec 2012.

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