What is NIPT?

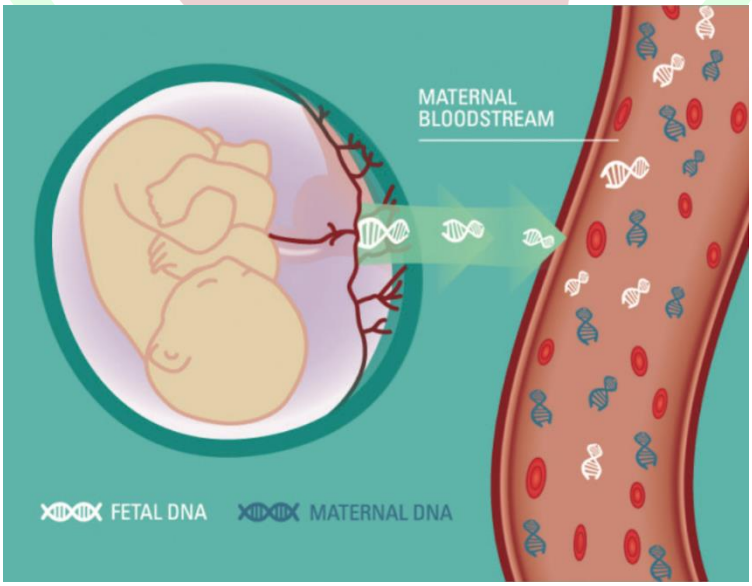
Non Invasive Prenatal Test (NIPT) or cell free DNA test (cfDNA) is a safe and simple blood test done to determine the baby's risk of having some chromosomal disorders including Down's Syndrome.

How is the test done?

During pregnancy some of the fetal DNA circulates in the maternal blood. NIPT analyses these small fragments of DNA circulating in the maternal blood. So we take a sample of maternal blood and send it to a certified laboratory for analysis. The test is considered non-invasive since only the maternal blood is taken and does not pose any risk to the baby.

What is cell free DNA?

DNA as some of you might know is often found in the nucleus of the cells. But those of the fetal DNA are free floating in the maternal blood pool and hence are called cell free DNA. Placenta is the connection between the mother and the baby which provides nutrition to the baby. It sheds some of the fetal DNA into maternal blood throughout pregnancy and the DNA of placental tissue is similar to the DNA of the fetus.



When can the test be performed?

It is usually performed after the 11-13 weeks nuchal translucency scan

What are the disorders that NIPT can detect?

Everyone of us are made of 23 pairs of chromosomes , one set from the mother and one set from the father which makes a total of 46 chromosomes. An addition or deletion in any of these chromosomes can result in various abnormalities. NIPT is a screening test for chromosomal abnormalities like trisomy 21/ Down's syndrome(caused by an extra pair of chromosome 21), trisomy 13, trisomy 18 and addition or deletion in X and Y chromosome.

How to interpret the result?

I. Screening results

Chromosomes	Risk	Z score	Test Results	Reference interval
Chromosome 21		0.74	Low Risk	-6<Z score<2.8
Chromosome 18		-0.54	Low Risk	-6<Z score<2.8
Chromosome 13		-1.17	Low Risk	-6<Z score<2.8
Sex Chromosomes		Please See Part III	Low Risk	Part III
Other Chromosomes		Please See Part IV	Low Risk	Part IV
Fetal fraction				
9.83%				

NIPT result can be either a Positive/High risk, Negative/low risk or inconclusive.

How accurate is NIPT in detecting Down's syndrome?

NIPT is said to be 99% accurate in screening for Down's syndrome and 98% for Trisomy 13 and 18. Also it can detect sex chromosomal abnormalities and triploid where the baby has 3 pairs of chromosomes in each cell instead of two.

Is NIPT 100% confirmatory?

No. NIPT is a screening test and hence if the result comes a 'high risk' the result has to be confirmed with an Invasive test (Amniocentesis/ chorionic villus sampling).

What are the reasons for an inconclusive result?

As NIPT detects the cell free fetal DNA that is circulating in the maternal blood, it requires a minimum fetal fraction to accurately detect the abnormalities. Hence if fetal fraction is below 4% NIPT fails to provide a result.