Non – Invasive Prenatal Testing (NIPT)

Introduction:

- ✓ Non-invasive prenatal screening (NIPS) is a new method of determining the risk that the foetus/ baby may be born with certain genetic abnormalities.
- ✓ This test is performed on peripheral blood sample of the pregnant woman and analyses the cell-free DNA (cf DNA) of the foetus that is circulating in a pregnant woman's blood

Who Should be Screened?

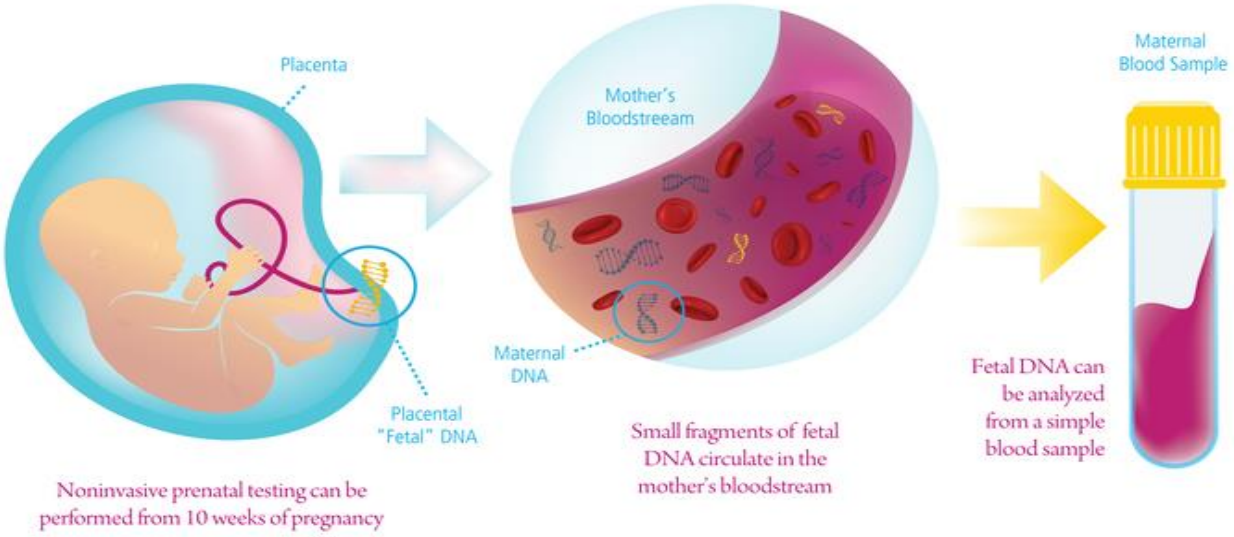
- ➢ Pregnant Woman who are 35 or older (advanced maternal age)
- ➢ Abnormal prenatal screening test that indicates the baby may at increased risk of having a chromosome condition.
- ➢ Abnormal ultrasound indicating a possible problem with the baby.
- ➢ Partners having family history of chromosomal abnormalities.
- ➢ Multiple miscarriages.
- ➢ Already having a child with a genetic disorder.
- ➢ Women who receive IVF or previously suffered from recurrent abortion.

Benefits

- ➢ The test is done by using maternal peripheral blood. (Completely safe for mother and baby)
- ➢ It can be conducted as early as 10 weeks post implantation; hence the patient can get most accurate screening information at an earlier gestational age, enhancing informed decision making.
- ➢ Only NIPS screen test that can identify Triploidy.
- ➢ Sensitivity is >99% with a false positive rate of <0.05%.

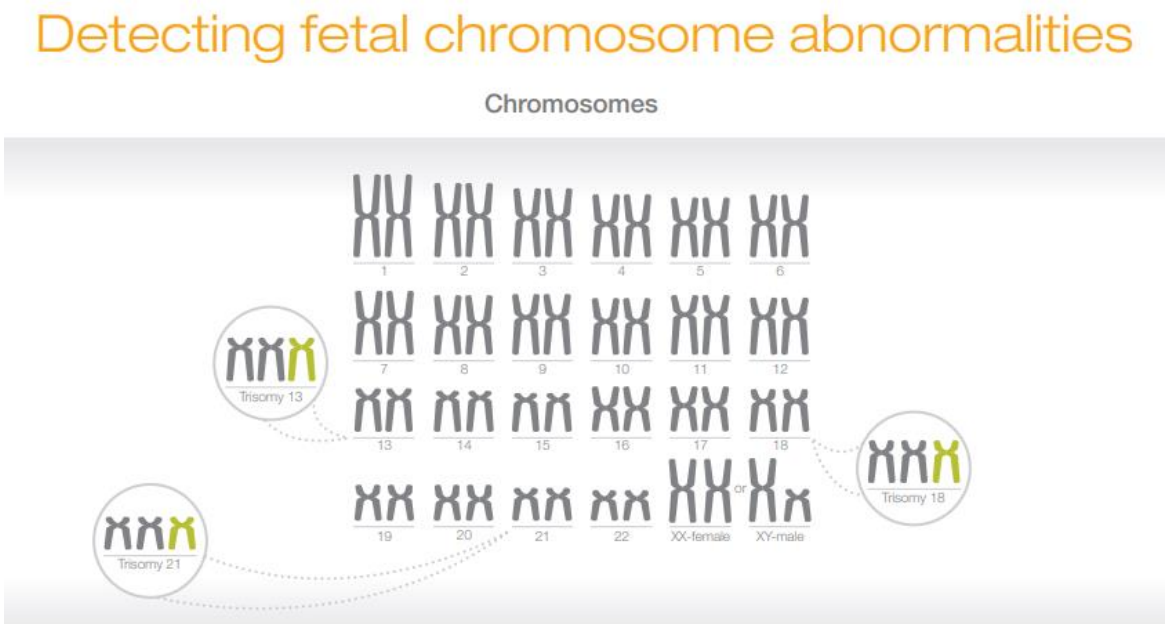
How does NIPS work?

- ➢ The maternal blood sample contains mixture of both maternal and foetal cfDNA.
- ➢ Both maternal and foetal fragments are counted and analyzed.



- Aneuploidies are detected by comparing the amount of chromosomal material against a set of reference chromosomes.

- **Down syndrome (Trisomy 21),** which is caused by an extra copy of chromosome 21. Mild/moderate mental retardation and risk of some birth defects (affects 1 in 1,000 live births).
- **Edwards syndrome (Trisomy 18),** which is caused by an extra copy of chromosome 18. Severe mental retardation and risk of multiple birth defects (affects 1 in 3,000-6,000 live births).
- **Patau syndrome (Trisomy 13),** which is caused by an extra copy of chromosome 13. Severe mental retardation and risk of multiple birth defects (affects 1 in every 5,000 live births).

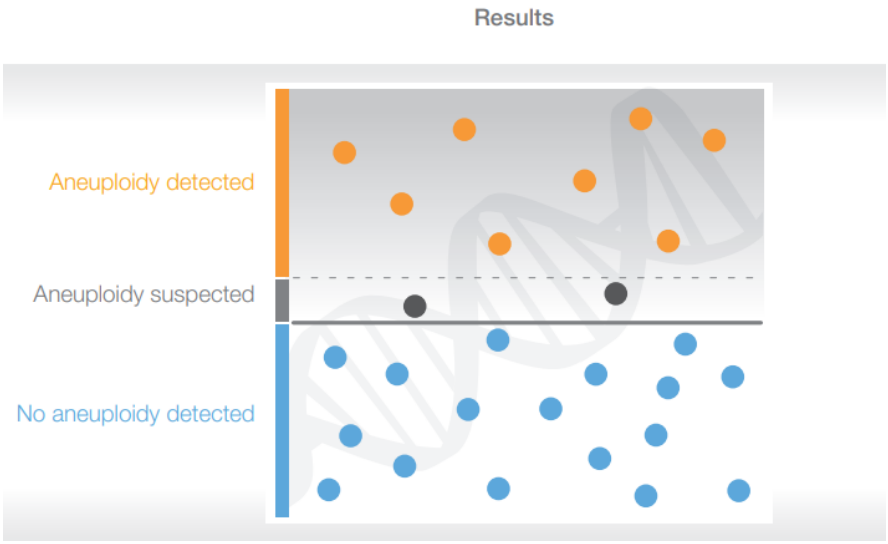


What does the result mean?

There are three possible results for the autosomes:

- Aneuploidy Detected,

- Aneuploidy Suspected (borderline value),
- Aneuploidy Not Detected.



- If the aneuploidy is not detected, the result is '**negative' or 'low risk'**, the baby is unlikely to have any of the chromosomal disorders tested.
- If the aneuploidy is suspected, the result is **'likely to be positive' or 'borderline risk'**, this means the baby is likely to be affected.
- If the aneuploidy is detected, the result is **'positive' or 'high risk'**, this means the baby is affected.

(Patients who receive an Aneuploidy Detected or Suspected result should be offered a follow-up discussion of results and options of invasive testing (CVS or amniocentesis) for confirmation.)

Significance of NIPS Tests over others

Method	**Diagnosis ability**	**False positive**	**Pregnancy age**	**Nature of method**
Screening with help of chemical markers of mother's blood	70%-90%	5%	$11\text{-}13^{+6}$ and $15\text{-}20^{+6}$	Non-invasive
NT ultrasonography	60%-80%	5%	$11\text{-}13^{+6}$	Non-invasive
CVS	>99%	Zero	10-13	Invasive (1-4 percent risk of abortion)
Amniocentesis	>99.9%	Zero	16-21	Invasive (0.5-1 percent risk of abortion)
NIPT	>99.9%	<0.05%	From week 10	Non-invasive

Recommenders:

Gynecologists and obstetricians

IVF specialist

Test Details:

Test Name	Test Code	MRP	Test Detail	Tech	Specimen	Temp	TAT
Non-invasive Pre-natal test (NIPT)	SMO10455	18000	13, 18 & 21 Chromosomes	DNA Sequencing	10 mL Whole blood in a special tube. Ship refrigerated. DO NOT FREEZE. Valid between 10-24 weeks of gestation. Give clinical history on Maternal Serum Screen request form	2-8°C	8th Working day by 7:00 p.m.
Panorama (NIPT) for Twins/Egg Donor/Surrogate Pregnancy	SMO10382	40000		SNP array	20ml Blood drawn with 21G or thicker needle in 2 Streck tubes provided in the Kit	2-8°C	25th working day by 7:00 p.m.