aayushya
DR. SHWETA BHANDARI'S
CENTER FOR FETAL MEDICINE

Non-Invasive Prenatal Testing (NIPT)

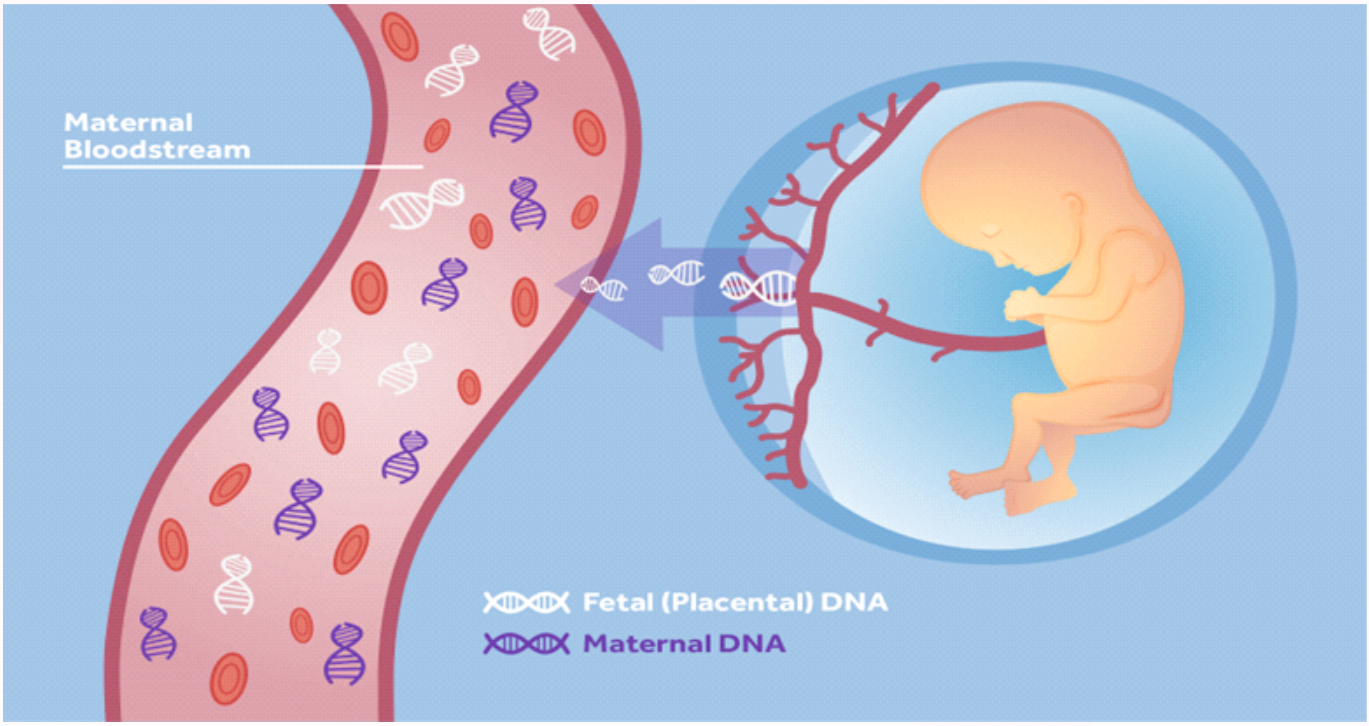
A pregnant woman and her partner have a choice about whether to have any screening and / or diagnostic tests. We are here to help her make decisions throughout the screening pathway. However there are benefits and limitations testing can bring. Having screening tests will provide her with more information.

Introduction to NIPT: Screening Test

Non-invasive prenatal testing (NIPT) is a more accurate screening test than the combined or quadruple test. **NIPT is blood test taken from a pregnant woman to assess the chance of the baby having Down's syndrome, Edwards' syndrome and Patau's syndrome.**

Methodology

During pregnancy the placenta sheds fetal DNA into the maternal bloodstream.
Most of the DNA fragments are from the pregnant woman but some are from the placenta.
These fragments are called cell free fetal DNA, although sometimes we use the term cfDNA.
NIPT analyses DNA from the placenta that is in the mother's blood.



Level of Reliability:

For Down syndrome = 99% I For Edward's syndrome = 96% I For Patau's syndrome = 94%

Analysis of NIPT

Most people will get a low-risk result, meaning your chance of having a baby with the condition is low. You will not be offered a diagnostic test.

If your NIPT result shows a high-risk result then the chance of your baby having the condition is high. You will then be offered a diagnostic test, though it is up to you whether or not to have this. In a small number of cases, NIPT might produce no result. You can then choose between one further NIPT, a diagnostic test or no further testing.

What does it test for?

The NIPT is a safe and very effective way of screening for certain conditions.

These include:
- Down syndrome (also called Trisomy 21)
- Edwards syndrome (Trisomy 18)
- Patau syndrome (Trisomy 13)
- Turner syndrome

When to Offer NIPT?

NIPT is very sensitive. It picks up more than 99% of cases of Down syndrome. But it is a screening test rather than a diagnostic test. It can tell you whether there is an increased chance of having a baby with a genetic condition. It doesn't give you a definitive answer. For some parents, information from screening tests can help them decide about whether to have diagnostic testing.

You might choose to have an NIPT test if:
- **Your first trimester combined screening test shows you have an increased chance of having a baby with Down syndrome (this test combines results from a blood test at 10 to 12 weeks and an ultrasound at 11 to 13 weeks).**
- **You did not have the first trimester combined screening test because it was too late or the test wasn't available in your area.**
- **You want to understand your chance of having a baby with Down syndrome before considering diagnostic tests such as amniocentesis or CVS.**
- **You have an increased chance of having a baby with Down syndrome because you are older.**
- **It's a good idea to consider genetic counseling before you have an NIPT to help you make an informed decision. It's important to understand the risks and benefits of having the test.**

When not to offer NIPT?

- **Risk for Downs syndrome in First trimester screening is in between 1:2 to 1:150.**
- **Strong soft markers or obvious structural abnormality seen on ultrasonography.**

In such a cases an invasive procedure for definitive testing is the recommended test.