Facts About Non-Invasive Prenatal Testing

Non-invasive prenatal testing (NIPT) is a blood test done during pregnancy. This test measures small bits of fetal DNA to check the developing baby for certain genetic conditions.

DNA is the genetic information we inherit from our parents. DNA is present in most cells of the body and is also found in blood. During pregnancy, some of the DNA from the baby crosses into the mother's bloodstream. This DNA carries the baby's genetic information. It is this fetal DNA that is tested and analyzed during NIPT to check for certain chromosome conditions.

Important Facts

- NIPT is a screening test during early pregnancy to detect risk of Down syndrome and other chromosome conditions.
- NIPT is safe and does not pose any risk to mother or baby.
- NIPT, like all prenatal test, is optional.
- The accuracy of NIPT is high but *NOT 100%*.
- NIPT does not test for all chromosome conditions or birth defects.
- Positive NIPT results require additional testing to verify diagnosis of chromosomal conditions.
- NIPT is unable to give a result in about 2-3 % of pregnancies.

Take time to decide about NIPT

- It's up to you to decide if you want this type of information during pregnancy.
- Knowing about a chromosome condition can be useful. You might find the information helpful to prepare for a child with special needs. You might use the information to decide whether or not to continue the pregnancy.
- Not everyone wants to learn about chromosome problems during the pregnancy.

How and when is it done?

Non-invasive prenatal testing is done by taking a sample of blood from the mother early in pregnancy, usually around 10-11 weeks or later.

What can NIPT tell me?

NIPT is able to tell you whether your pregnancy is at low risk or high risk for certain chromosome conditions, including: Down syndrome (Trisomy 21), Trisomy 18, Trisomy 13, and sex chromosome disorders. Fetal sex can also be predicted in most pregnancies.

How accurate in NIPT?

NIPT is highly accurate for the chromosome condition identified by the test result however the accuracy is not 100%. There are also differences in the accuracy, depending on the chromosome conditions tested for. There is also the possibility that no test results are given due to not enough fetal DNA in the mother's blood or difficulty identifying fetal DNA from the mother's DNA.

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Who is NIPT for?

- Women 35 or older
- Previous child born with a chromosome condition.
- Abnormal ultrasound indicating a possible problem with the baby.
- Abnormal prenatal screening test that indicates the baby is at increased risk of having a chromosome condition.

What if NIPT is positive?

If you have a high risk result, you will be offered follow-up testing to verify whether or not the developing baby actually has a chromosome condition.

What follow-up testing will be offered?

You will be referred to a Maternal Fetal Medicine Specialist who will help you decide on recommended testing.

- Chorionic villus sampling (CVS) is done between 10 and 14 weeks of pregnancy. A small sample of the developing placenta is taken by passing a thin needle through the lower abdomen or using a flexible plastic tube through the vagina.
- Amniocentesis is usually done between 15 and 22 weeks in pregnancy. A small amount of the amniotic fluid that surrounds the baby is taken by passing a thin needle through the abdomen.

Both CVS and amniocentesis accurately identify chromosome problems by directly examining fetal cells. However, both of these tests pose a small risk of miscarriage to the pregnancy.

How long does it take to get NIPT test results?

NIPT results usually take about 8 to 14 days. You will get a phone call when your results are ready. In a small number of pregnancies the test is unable to give any results and repeat testing is recommended.

More about……..

Down syndrome is a chromosome condition associated with mild to moderate intellectual disability, a characteristic appearance, and weak muscle tone in infancy. Babies with Down syndrome also have a higher chance to be born with physical birth defects, such as a heart defect or intestinal problems.

Trisomy 13 and 18 are two different chromosome problems associated with severe intellectual disability and medical problems in many parts of the body. For either condition, survival beyond the first year of life is uncommon.

Sex chromosome disorders are a group of conditions that have a difference in the usual number of sex chromosomes. The most common sex chromosome conditions are Turner syndrome, Klinefelter syndrome, Triple X syndrome, and XYY syndrome. These conditions are typically milder than Down syndrome but can include learning difficulties and infertility. Babies with Turner syndrome also have a higher chance for physical birth defects, especially heart defects.