

North Carolina Women's Hospital Non-Invasive Prenatal Testing (NIPT) for Down Syndrome, Trisomy 18 and Trisomy 13

What is Non-Invasive Prenatal Testing?

Non-Invasive Prenatal Testing (NIPT) is one of several screening and testing options available to women during pregnancy. Results from this test are used to determine if a pregnancy has an increased chance of being affected with one of three conditions, Down syndrome, trisomy 18, or trisomy 13. This test is also sometimes called cell-free fetal DNA screening.

NIPT is a new technology measuring DNA that comes from the pregnancy and found in the mother's blood. DNA is the genetic material present in humans and is found in every cell of our bodies. It contains the instructions for how we grow and develop. DNA is packaged together in structures called chromosomes. Typically, a healthy individual has 23 pairs of chromosomes, for a total of 46 chromosomes.

Is NIPT available to all pregnant women?

At this time, NIPT is only being offered to women who have an increased chance of having a pregnancy with Down syndrome, trisomy 18, or trisomy 13. All pregnancies have some chance of being affected with these conditions. However, there are certain factors that increase a woman's chance of having a pregnancy with Down syndrome, trisomy 18, or trisomy 13. These factors can include the mother's age or her previous pregnancy history. Additionally, women who have had an abnormal screening test or ultrasound finding may be at increased risk for having a pregnancy with these conditions.

What is Down syndrome?

Down syndrome is the most common genetic condition seen in newborns. About 1 in 700 babies will be born with the condition. Down syndrome is also called trisomy 21, because individuals with Down syndrome have an extra copy of chromosome 21. Down syndrome typically happens by chance and does not run in families. Individuals with Down syndrome usually have developmental delays and intellectual disabilities. They are also at increased risk for heart problems and other health concerns. All people with Down syndrome are unique. In most cases, it is not possible to predict how a child with Down syndrome will be affected before they are born.

What are trisomy 18 and trisomy 13?

Trisomy 18 is the result of an extra chromosome 18 and trisomy 13 is the result of an extra chromosome 13. Both of these conditions are less common than Down syndrome. Babies with trisomy 18 or trisomy 13 usually have severe birth defects and often do not live very long after birth. Many pregnancies with trisomy 18 or trisomy 13 result in miscarriage or stillbirth. Like Down syndrome, trisomy 18 and trisomy 13 typically happen by chance and do not run in families.

How is NIPT done?

NIPT is a blood test that is done using a sample of a pregnant women's blood. The test specifically looks at information from chromosomes 21, 18, and 13. Since the mother's blood contains genetic material from the pregnancy and from herself, the test will measure the total amount of genetic material.

Do I need to have an ultrasound before having NIPT?

An ultrasound is recommended before the screening is completed. The ultrasound is needed to determine how far along a woman is in her pregnancy, as this screening can only be done after 10 weeks of pregnancy. It is also important to know if the woman is carrying more than one baby. Additionally, if any abnormalities are found on ultrasound, then other testing options will likely be recommended.

How will the results of this screening come back?

The results will either be reported as "negative" or "positive" for each of the three conditions. In about 1% of samples, no result is reported due to test failure. Results typically take about two weeks, but in some cases may take longer.

What does it mean to have a "negative" result?

A "negative" result means that extra chromosome 21, 18, or 13 material was not detected in the blood sample. This result would mean that it is very unlikely that the pregnancy has Down syndrome, trisomy 18, or trisomy 13. However, it cannot exclude the possibility that the pregnancy has these conditions. It is also important to remember that this test does not detect all chromosome or genetic conditions in the pregnancy.

What does it mean to have a “positive” result?

A “positive” result means that extra material from chromosome 21, 18, or 13 was detected in the blood sample. This means that Down syndrome, trisomy 18, or trisomy 13 is very likely. The detection rate for this screening is reported to be at least 99% for Down syndrome, approximately 99% for trisomy 18, and from 79-92% for trisomy 13. As this test is only considered to be a screening test, diagnostic testing will be recommended to confirm a “positive” result. This will include the option of CVS or amniocentesis.

What are CVS and amniocentesis?

Chorionic villus sampling (CVS) and amniocentesis are tests used to diagnose conditions like Down syndrome, trisomy 18, or trisomy 13 in pregnancy. The CVS is done by taking a small sample of cells from the placenta. It is performed between 10-14 weeks of pregnancy. Amniocentesis is done by taking a small sample of the amniotic fluid. It is performed after 15 weeks of pregnancy. Both of these tests have a chance of causing pregnancy complications or miscarriage. The chance for miscarriage as a result of the CVS is about 1 in 200 pregnancies (0.5%) and the chance for miscarriage as a result of amniocentesis is about 1 in 300 to 1 in 500 pregnancies (0.2-0.3%).

What is the cost of NIPT?

The cost and billing for this screening depends on the woman’s specific insurance. The laboratories offering this screening currently have financial assistance available on a sliding scale and may be able to assist with out of pocket costs.

Will I be offered other screening tests in my pregnancy?

You may be offered a screening test in the second trimester to screen for neural tube defects, such as spina bifida, in the pregnancy. Screening for neural tube defects can be done through a blood test called AFP, or alpha-fetoprotein, test. A targeted ultrasound in the second trimester can also detect most cases of neural tube defects.

What if I have more questions?

Ask your healthcare provider to refer you to UNC Women’s Hospital or UNC Specialty Women’s Center at Rex for a genetic counseling appointment. A genetic counselor will review all testing and screening options and help you choose the option that is best for you.

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Notification to Users

These algorithms are designed to assist the primary care provider in the clinical management of a variety of problems that occur during pregnancy. They should not be interpreted as a standard of care, but instead represent guidelines for management. Variation in practices should take into account such factors as characteristics of the individual patient, health resources, and regional experience with diagnostic and therapeutic modalities. The algorithms remain the intellectual property of the University of North Carolina at Chapel Hill School of Medicine. They cannot be reproduced in whole or in part without the expressed written permission of the school.

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