

Facts about Non-invasive Prenatal Testing

Non-invasive prenatal testing (NIPT) is a blood test done during pregnancy. This test measures small pieces of fetal DNA to check the developing baby for certain chromosome disorders. It is also called cell-free fetal DNA testing.

DNA is the genetic information we inherit from our parents. DNA is present in most cells of the body and is also found in your blood. When you are pregnant, a small amount of the DNA in your blood comes from the placenta and developing baby. Special blood testing can find pregnancies that seem to have a difference in the usual amount of fetal DNA. This lets you know about your baby's chance to have certain birth defects.

How is it done?

NIPT is done with a single blood test in the first or second trimester. It can be done any time after the 10th week of pregnancy.

What can NIPT tell me?

NIPT can tell you if your pregnancy is at low risk or high risk for common chromosome disorders, including: Down syndrome (trisomy 21), trisomy 18, trisomy 13. NIPT helps find almost all pregnancies with Down syndrome and trisomy 18. It also finds most pregnancies with trisomy 13. NIPT will also screen for sex chromosome disorders, unless you are carrying twins. Your final result includes a risk estimate for each chromosome disorder. Fetal sex can also be predicted in most pregnancies.

Who can have NIPT?

Kaiser Permanente routinely offers NIPT to women who have a higher risk for chromosome disorders, including women who will be 35 or older on their due date. This screening test may also be offered to women who have a screen positive result from the California Prenatal Screening Program, and to women who have a family history that increases the risk for chromosome disorders.

How long does it take to get results from NIPT?

Results from NIPT usually take about 8 to 14 days. You will get your result by a secure email message or a phone call when the result is ready.

What if my NIPT result is low risk?

Most women who have NIPT get a low risk result. A low risk result is very reassuring that your baby does not have Down syndrome, trisomy 18, trisomy 13, or a sex chromosome disorder. Rarely, a baby with one of these conditions will have a low risk result and be missed by this test.

A low risk result does not guarantee that the baby has no health problems or birth defects. Keep in mind that this test **does not** provide information about all chromosome problems and does not test for other types of birth defects or genetic conditions. If you are carrying twins, NIPT does not screen for sex chromosome conditions.

What if NIPT does not give ANY result?

In a small number of pregnancies NIPT is unable to give any result. Most often this is because there was not enough fetal DNA to examine or the sample did not pass all of the quality control steps at the lab. This does not usually mean a problem with the baby. NIPT is more likely to have no result in heavier women, in twin pregnancies, and in some pregnancies with a chromosome disorder. If there is no result, you will need to decide whether to repeat the test or choose different testing.

TAKE TIME TO DECIDE ABOUT NIPT

It's up to you to decide if you want to learn about chromosome disorders during pregnancy.

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.

But not everyone wants to learn about chromosome disorders before the birth of the baby.

IMPORTANT THINGS TO CONSIDER

Non-invasive Prenatal Testing:

- ...has no risk for miscarriage
- ...may not identify all pregnancies with Down syndrome, trisomy 18, trisomy 13, or sex chromosome disorders.
- ...does not test for all chromosome disorders.
- ...does not test for other genetic conditions or other birth defects.
- ...may signal a high risk in a healthy pregnancy (false-positive result).
- ...requires additional testing to verify a diagnosis of a chromosome disorder.
- ...is unable to give a result in a small number of pregnancies

What if my result is inconclusive for fetal sex or sex chromosomes?

Some women are told that a result cannot be provided for the sex chromosome disorders or the fetal sex (or both). This is usually due to the complex biology of X and Y chromosomes and not because of a problem with the baby. Since repeat testing is not likely to give a result, different testing may be considered.

What if my NIPT result is high risk?

A high risk result is reported when there seems to be a difference in the expected amount of DNA. This may be because the developing baby has a chromosome disorder, but there are other reasons for a high risk result. If you have a high risk result, more testing is offered to verify whether or not the developing baby actually has a chromosome disorder.

What follow-up testing will I be offered if my result shows a high risk?

If you have a high risk result you will be offered either CVS or amniocentesis.

- **Chorionic villus sampling (CVS)** is done between 10 and 14 weeks in 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 fluid that surrounds the baby is taken by passing a thin needle through the abdomen.

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

More about...

Down syndrome is a chromosome disorder associated with mild to moderate intellectual disability, and a characteristic appearance. Babies with Down syndrome also have a higher chance to be born with physical birth defects, such as a heart defect or intestinal problem.

Trisomy 18 and trisomy 13 are two different chromosome disorders 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 disorders are Turner syndrome, Klinefelter syndrome, triple X syndrome, and XYY syndrome. These conditions are typically milder than Down syndrome, but can include learning difficulties, differences in growth, and infertility later in life. Babies with Turner syndrome are also more likely to have physical birth defects, like a heart defect or kidney abnormality. NIPT cannot screen for sex chromosome disorders in twin pregnancies.