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Noninvasive Prenatal Screening Patient Brochure

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What is noninvasive prenatal screening or NIPS?

Noninvasive prenatal screening, or NIPS, is a test you may have while you are pregnant. The test screens your developing baby for chromosome disorders and can determine your baby's sex. If your healthcare provider determines that you have a high risk of having a child with chromosome disorders, then you may want to have the NIPS test.

The NIPS test looks at your baby's DNA, the unique genetic code inside the cells, packaged in structures called chromosomes. Your placenta releases fragments of your baby's genetic code, and it circulates in your blood. NIPS purifies a sample of your blood to look only at your baby's DNA to see if there are chromosome abnormalities or mutations.

Both the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG) recommend NIPS.

What makes NIPS different from other prenatal tests?

The NIPS test is noninvasive, which means it does not require an invasive procedure or exam. There is no risk to you or the baby during this test. Other prenatal tests such as amniocentesis and chorionic villus sampling (CVS) require a sample of fluid from around the baby. By eliminating the need to collect this fluid, the risk of miscarriage and other complications is lower. NIPS only requires a blood draw from the mother.

NIPS also has a lower false-positive rate and a higher detection rate for genetic disorders compared to other screening tests. This means results are more accurate, so the likelihood of needing further invasive procedures is lower.

The NIPS test has a quicker turnaround time for results. Other genetic tests require seven to 10 days, but NIPS results are delivered in only three to seven days from the time the lab receives the blood sample.

Who should have the NIPS test?

Your healthcare provider will discuss your risk factors for having a child with chromosome disorders to determine if you need NIPS.

Common risk factors for pregnant women include:

- You are 35 or older.
- You have an ultrasound during your pregnancy that indicates a risk for chromosome disorders in your baby.
- You have a family history of chromosome disorders.
- You had a previous pregnancy or child with chromosome disorders.
- You are a known carrier of chromosome abnormalities.

You may have this test if you are carrying one child or twins, but it does not work for triplets or more. You may also have NIPS if you are having an in vitro fertilization (IVF) or egg-donor pregnancy.

When should you have the NIPS test?

You can have the test starting from the 10th week of your pregnancy. If you wait longer, there will be more cell-free DNA from your baby in your blood, so the results may be more accurate. Usually, women have this test during their first trimester.

What does the NIPS test include?

The NIPS test screens for chromosome disorders in your baby. It can also show the baby's sex.

The test can show if your baby has a high or low risk of having the following genetic disorders:

- Trisomy 21 (Down syndrome)
- Trisomy 13 (Patau syndrome)
- Trisomy 18 (Edwards syndrome)
- 45,X (Turner syndrome)
- 47,XYY (XYY syndrome)
- 47,XXY (Klinefelter syndrome)
- 47,XXX (Triple X syndrome)
- 48,XXYY (XXYY syndrome)

How do you prepare for the NIPS test?

Since the test is noninvasive, you do not have to do anything special to prepare for it. However, you may want to talk about any concerns you may have with your healthcare provider.

How does the test work?

First, your healthcare provider will describe the NIPS test and offer genetic counseling. Then, you will have a blood draw from a vein in your arm, and two test tubes of your blood will go to the laboratory for screening. The lab will run tests on the blood sample and will send the results to your doctor. Your doctor will then meet with you to discuss the results and decide the next steps, if necessary.

What happens to my blood sample in the lab?

The lab purifies your blood sample to isolate your baby's cell-free DNA. Then, the DNA goes through sequencing to look for mutations. The lab focuses on the DNA for chromosomes 13, 18, 21, X and Y to increase efficiency and save time. These are the most common chromosomes to have mutations that lead to genetic disorders.

What are the limitations of the NIPS test?

It is important to remember that NIPS is a screening test and not a diagnostic test. It can only show if your baby has a high or low risk of having a specific chromosome disorder. It cannot determine with 100 percent accuracy if your baby has the condition.

NIPS does not screen for every genetic disorder that your baby may have. It only looks at the most common ones in the population.

This may not be the right test for you if:

- You are obese.
- You had a bone marrow transplant.
- You are pregnant with triplets or more.
- You had vanishing twin syndrome during this pregnancy.
- Your DNA has chromosome abnormalities that can be confused with your baby's DNA.
- Your blood has a low amount of your baby's cell-free DNA.
- You are not able to give a large enough sample of blood for testing.
- Your blood is more than seven days old when it arrives in the lab.

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How long does it take to get the results?

Turn-around time is usually three to seven days after the lab gets the blood sample. Your healthcare provider should contact you upon receiving the results.

What do the results mean?

Positive test results show a high probability of your baby having a chromosome disorder that is greater than 99/100 or 99 percent. The recommendation for positive results is genetic counseling and diagnostic testing. A positive result does not guarantee that your baby has a specific genetic disorder.

Negative results show a low probability of less than 1/10,000 or 0.01 percent of your baby having a chromosome disorder. The recommendation for negative results is genetic counseling, if necessary. A negative test result does not guarantee that your baby does not have chromosome abnormalities.

In some cases, you may not get positive or negative results because of insufficient data or other problems. Ask your healthcare provider about repeating the test at a later date or having a different screening procedure.

How can you get more information?

Talk to your healthcare provider if you need more information about the NIPS test or the results. Your doctor can also recommend genetic counseling and other resources.

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