

# Noninvasive prenatal screening: What is it?



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Maternal-Fetal Medicine

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### What are the concerns regarding noninvasive prenatal testing?

Our doctors have some concerns about this new test:

- It is a new, complicated test that has not been as fully reviewed by medical experts as other more established tests.
- You may have to wait up to two weeks for results.
- It's possible that your test would not show any conclusive results. In this case, you would need another blood sample or another type of test.
- If NIPS shows a high chance for a chromosome abnormality, your doctor may recommend more testing. More testing means more time until you have an answer.
- We don't know how this test works in unusual situations. It may not work in cases of mosaicism. Talk to your doctor/midwife or request a consultation in our office to learn more.

### Does insurance cover NIPS?

NIPS is new and is not always covered by insurance companies. You may find that it is difficult to get an accurate answer about coverage and cost for patients, even when insurance is verified ahead of time.

Most people whose insurance companies cover noninvasive prenatal screening are billed around \$200–500 for the test. However, some people pay significantly more or less than this amount.

Talk with your doctor or genetic counselor about your options and the risks and benefits of noninvasive prenatal screening.

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# A testing option during pregnancy

**You may have heard** of new, noninvasive ways to screen for Down syndrome during pregnancy. These tests use a blood sample taken from the mother to check DNA for a few chromosomal abnormalities. While there are several brand names for these new tests, they are generally referred to as “noninvasive prenatal screening (NIPS).”

There are benefits to NIPS. However, this type of testing is not the best choice for every patient. This brochure answers some questions about noninvasive prenatal screening.



## Whom is this test for?

Noninvasive prenatal screening is an option for pregnancies where an increased risk for at least one of three specific chromosomal conditions has already been identified: Down syndrome, trisomy 18 or trisomy 13. It is not recommended for pregnancies with twins or other multiples.

## How does it work?

When a woman is pregnant, some of the DNA in her blood is from the pregnancy. NIPS captures this DNA in order to screen for Down syndrome, trisomy 18 and trisomy 13 in the baby.

DNA is the hereditary material that instructs our bodies how to grow and develop. DNA is packaged into structures called chromosomes. Every person has 23 pairs of chromosomes, for a total of 46.

These pairs of chromosomes come from our parents. One set of each pair comes from the mother in her egg, and the other set comes from the father in his sperm. Sometimes a baby inherits an extra chromosome, resulting in three copies of a chromosome instead of the typical two.

The extra chromosome generally leads to changes in growth and development of the baby to varying degrees:

- Down syndrome occurs when there is an extra copy of chromosome 21.
- Trisomy 18 occurs when there is an extra copy of chromosome 18.
- Trisomy 13 occurs when there is an extra copy of chromosome 13.

## What are the benefits?

This new technology is more accurate than other screening tests. In addition, noninvasive prenatal screening:

- Does not increase the risk of miscarriage; other more invasive prenatal tests carry this risk
- Carries less chance for a false-positive result as compared to other blood-screening tests
- Can be offered any time after 10 weeks gestation, along with a first trimester ultrasound

## What are the limitations of NIPS?

NIPS is not a diagnostic test like amniocentesis or chorionic villus sampling (CVS). These tests are more accurate and can also detect other conditions. However, because these tests are invasive, they have a risk for miscarriage. Other limitations of NIPS:

- Currently, NIPS does not reliably test for anything other than extra copies of chromosomes 13, 18 and 21. It is only recommended for pregnancies specifically at risk for these conditions.
- NIPS is not yet proven effective for low-risk pregnancies or when there is more than one baby.
- Both “false-positive” and “false-negative” results have been reported using NIPS. A false-positive is a test result that says your baby has the condition when he or she does not. A false-negative is when the test says your baby does not have the condition when he or she does.
- It has a higher failure rate than other tests.
- Even if NIPS results are normal, your doctor may still recommend additional testing.