

# Understanding Prenatal Genetic Testing

Are you curious about your baby's health before they're even born? Most babies are born healthy, but some babies have health problems. Some problems are mild, but others can be very serious. Health problems can change how the baby looks, grows, and learns.

Sometimes we can find out about these problems before birth. There are several tests that can help you learn more about whether your baby may have health problems.

It is normal to feel uncertain or anxious about whether testing is right for you, and which test to choose. Your healthcare provider can support you, and help you decide which testing will be most helpful for you.

## Should I have the tests?

Some families feel okay without testing. They feel ready to care for their baby whether or not their baby has health problems.

Some families choose to have some tests, but not others. It is your choice to have the tests. Your healthcare provider will support whatever you decide.

## What are the kinds of tests?

There are two types of tests, screening tests and diagnostic tests.

**Screening tests** take a quick look to check if your baby may have health problems. They can usually be done with a blood draw. They are safe for you and your baby. Screening tests are usually right, but sometimes they can be wrong.

**Diagnostic tests** are more accurate than screening tests. They look for more health problems, but they have a small chance of miscarriage or infection. Anyone can choose to have a diagnostic test instead of a screening test. Families may want diagnostic testing if a screening test has shown their baby may have a health problem, or if they have had a previous pregnancy with a health problem.

Both kinds of tests can help you understand more about how your baby is growing. Sometimes tests show unexpected things about your baby or your family members, and sometimes the test results are not clear. Tests are not perfect, and they cannot promise a baby will be healthy.



### Something to Think About—

What would I do if my baby had a health problem?

### Something to Think About—

Most results are normal, but how will I feel if my results show my baby may have a health problem?



## Screening Tests

### Carrier Screening

Carrier screening is a blood test you can have before or during pregnancy. It looks for hidden health conditions in parents that may affect their children. Parents who carry these hidden conditions are called carriers. They usually do not have any signs or history of the condition. However, sometimes it is possible for their children to be born with health problems, like lung diseases, muscle diseases, or blood disorders.

Sometimes both parents need to carry the same hidden condition for the baby to be affected. Other times, only one parent needs to be a carrier for the baby to be at risk.

If you are found to be a carrier, your partner might need a blood test to see if the baby could have a health problem. You may also talk to a genetic counselor about your results.

Before you have this test, tell your healthcare provider if anyone in your family has a health problem, or if you and your partner are related by blood.

### NIPT (Noninvasive prenatal testing)

NIPT is a blood test you can have around 10 weeks of pregnancy or later. It is a screening test that checks for Down syndrome and a few other rare and serious conditions.

These conditions happen when a baby does not have the right amount of DNA to grow. You cannot control how much DNA your baby gets, and these conditions cannot be cured.

NIPT can check your baby's DNA that predicts their sex. It can also see differences in DNA that might affect their sex. Many times, these differences can have mild effects on your baby.

If the NIPT screening test shows that your baby may have a health problem, you may want to consider a diagnostic test.

### AFP Screening

AFP screening is a blood test done between 15 and 20 weeks of pregnancy. It checks if your baby may have a problem with their brain or spine.

If AFP screening shows that there may be a problem, your provider may recommend an early ultrasound to check your baby's brain and spine.

#### Something to Think About—

What's important for me to know about my baby before birth?

DNA is like a code in the body's cells. Babies inherit DNA from their parents. DNA tells their body how to grow. Having too much or too little DNA can affect your baby's health.



## Ultrasound

Ultrasound is a safe way to see how your baby is growing. It uses sound waves outside of your body to make pictures of the baby. These pictures show how your baby's heart, brain, and other organs are growing. Usually, ultrasound is done around 20 weeks of pregnancy, but your healthcare provider may recommend more ultrasounds at other times.



## Diagnostic Tests

### Chorionic Villus Sampling (CVS)

CVS is a diagnostic test you can choose that is done around 10 to 13 weeks of pregnancy. It is more accurate than a screening test. It can be used to check your baby's DNA by taking a small piece from the placenta (afterbirth). The test is done by placing a tube or a needle through your vagina or belly.

CVS has a small risk of miscarriage and infection. Most people feel cramping during the test. You may want to consider CVS if you have a genetic problem in your family, or if a screening test has shown your baby may have a health problem.

### Amniocentesis (Amnio)

Amnio is a diagnostic test you can choose starting around 16 weeks of pregnancy. It is the most accurate test that can be done in pregnancy. Amnio can be used to test your baby's DNA, and to check for infections that may affect your baby.

The test is done by taking a sample of fluid from around your baby with a needle. Most people feel cramping during the test. It has a small risk of miscarriage and infection. You may want to consider amnio if you have a genetic problem in your family, or a screening test has shown your baby may have a health problem.

#### Something to Think About—

Which of these tests will help me to get the information that is most important to me?



## Should I talk to a genetic counselor?

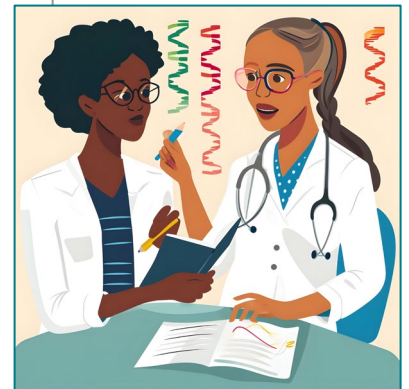
A genetic counselor is a healthcare provider who helps patients get and understand information about health conditions running in their family. They also help families adapt to what this information means to them. They support families in making decisions with this information.

Ask your provider about talking to a genetic counselor if—

- You have questions about your test results
- You would like to consider more testing
- Ultrasounds have shown differences in your baby
- You are worried about a health condition affecting your baby

## Questions to ask yourself and your healthcare provider—

- What is important for me to know about my baby before they are born?
- Most babies are born healthy, but what would I want to do if my baby had a health problem?
- Most results are normal, but how will I feel if my results show my baby may have a health problem?
- Which of these tests will help me get the information that is important to me?



## Find Out More

Read more about prenatal genetic testing here.

<https://medlineplus.gov/prenataltesting.html>

<https://medlineplus.gov/spanish/prenataltesting.html>