Allina Health

# What You Should Know About Prenatal Tests

## Birth Defects

Most babies in the United States are born healthy. But all pregnancies face a 3 to 4 percent chance of having a birth defect and a 2 to 3 percent chance of developing a cognitive (thinking, learning) delay.

Some pregnancies may be at a higher risk because of maternal age, family history, ethnicity or other factors. If you have a special concern, talk with your health care provider before deciding about a prenatal test.

There are a variety of prenatal tests available to you. Choosing the right test can be difficult and confusing. The following information is an overview about prenatal testing to help you decide if it is right for you. It may also be helpful to talk with your health care provider or a genetic counselor.

## Types of Tests

There are two types of tests available to all pregnant women:

- **Screening tests:** A screening test is used to identify pregnancies that have a "high" risk of having a specific birth defect. It does not tell you if your baby has a birth defect. If a screening test is abnormal, it means you are in a higher-risk category and a diagnostic test will be offered. Most women who have an abnormal screening test result have healthy babies.
- **Diagnostic tests:** A diagnostic test can determine if there is a specific problem. Most diagnostic tests are more than 99 percent accurate.

No test can guarantee that you will have a healthy baby. There are no tests that can identify all types of birth defects and developmental conditions. However, prenatal screens may help predict if you have an increased risk of carrying a baby with some specific problems. Prenatal diagnostic tests can say "yes" or "no" about most of the concerns raised by the screening tests.

## Screening Tests

There are several different screening test options available to you. The most common birth defects addressed by prenatal screens are:

- Down syndrome, a condition that causes mild to moderate delays in mental development
- trisomy 18, a condition that causes severe developmental disabilities
- spina bifida, an opening into the baby's spine.

### First Trimester Screening

First trimester screening is done between 10 and 14 weeks of pregnancy. This screening test includes a blood test and an ultrasound. The blood test measures proteins made by your baby. The ultrasound (called nuchal translucency or NT) is used to measure the thickness of the fluid at the back of the baby's neck. These tests will tell you if your baby is at "high" risk for having Down syndrome or trisomy 18. These tests do not screen for spina bifida.

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First trimester screening has an 83 to 85 percent detection rate for Down syndrome. If the screen is abnormal, a diagnostic test such as an amniocentesis or chorionic villi sampling (CVS) will be offered.

### Second Trimester Screening

Second trimester screening is done between 15 and 20 weeks of pregnancy. This screening test is a blood test that measures proteins made by your baby.

Second trimester screening has about an 80 percent detection rate for Down syndrome. If the screen is abnormal, an amniocentesis will be offered. The second trimester screen can also detect most pregnancies affected with spina bifida. If your provider has a concern about spina bifida, a detailed ultrasound or amniocentesis will be offered.

### Sequential or Integrated Screening

A sequential or integrated screening test combines the results from the first and second trimester screens. These screens have the highest detection rate for Down Syndrome. If these screens are abnormal, an amniocentesis would be offered. Ask your health care provider or genetic counselor about this option.

## Diagnostic Tests

Diagnostic tests can determine if there actually is a specific birth defect. These tests are relatively safe, but there is a small increased risk of miscarriage.

The earliest diagnostic test that is available is chorionic villi sampling (CVS). This test is usually done between 10 and 12 weeks of pregnancy. A CVS uses an ultrasound to guide a catheter (thin plastic tube) into your vagina, through the cervix, into your uterus and to the placenta. A small sample of chorionic villi will be removed by the catheter.

The most common type of diagnostic test is an amniocentesis. This test is done after 15 weeks of pregnancy. An amniocentesis uses an ultrasound to guide a thin needle into your lower abdomen and into the amniotic sac. A small sample of amniotic fluid is removed.

Both of these tests are more than 99 percent accurate in finding most abnormalities in chromosomes such as Down syndrome. There may be a slightly lower risk of miscarriage with an amniocentesis than with CVS.

## Making Decisions

When making a decision about prenatal testing, keep in mind the following:

- If you want to know if there is or isn't a problem, a diagnostic test is best.
- If you want to know if you are at a high or low risk for a birth defect before having a diagnostic test, a screening test is best.
- If you feel that a screening or diagnostic test is something you do not want to have, you can choose not to have any prenatal testing.

During pregnancy you will be asked to make many health care decisions. In many cases there isn't a clear "good" or "bad" choice. The best decision is one that feels right for you and your circumstances.

When you face a tough decision, it may be helpful to talk with your health care provider or a genetic counselor. He or she can review your risk factors and talk with you about the different options you have for prenatal testing.

For more in-depth information about birth defects and prenatal tests, visit The American College of Obstetricians and Gynecologists (ACOG) website at acog.org and choose the *ACOG PATIENT PAGE*.





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