UW Medicine

Guide to Prenatal T esting

Learning about your baby's health

This chapter describes prenatal tests that give information about your baby's health. It is your choice whether or not to have these tests done. Talk with your healthcare provider to learn more and to help you decide if any of these tests are right for you.

If you have any of these tests done, you will be asked to read more about each one. You will also be asked to read and sign a consent form for each test.

You can do many things during your pregnancy to keep you and your baby healthy. It is very important to take your prenatal vitamins, eat healthy foods, exercise, and get enough sleep.

But the human body is complex. Even if you do everything "right" during your pregnancy, babies do not always develop normally. Between 3% and 5% of babies (between 3 and 5 out of 100) have some kind of health problem when they are born.

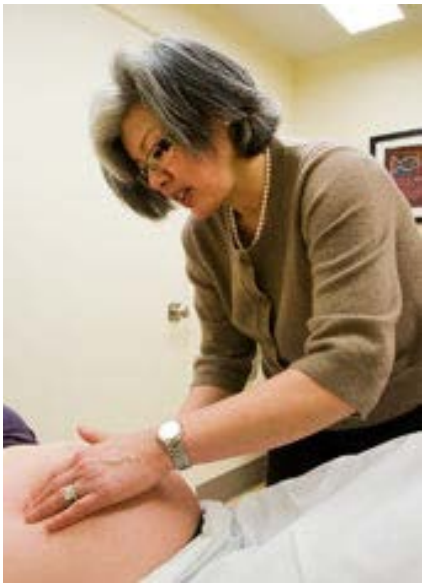
This handout gives some basic information about these tests to help you make the best decision for you.

What are the tests?

There are 2 basic kinds of tests:

- **Screening tests** predict the *chance*, or odds, that your baby has a certain birth defect.
- **Diagnostic tests** tell you if your baby does or does not have a certain birth defect.

The tables on the next page list the tests and when they are done. They also give a brief description of each test and what it will tell you. The rest of this chapter gives more details about these tests, if you would like to read about them before you talk with your provider.



Prenatal tests can provide information about your baby's health.

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Screening Tests

Name of Test	When	Description	What It Tells You
Nuchal translucency (NT) ultrasound	11 to 14 weeks	Abdominal ultrasound to measure small space behind baby's neck	*Chances* your baby has a chromosome problem
Integrated screen	11 to 14 weeks *and* 15 to 22 weeks	NT ultrasound plus 2 separate blood samples	*Chances* your baby has Down syndrome, trisomy 18, or spina bifida
Quad screen	15 to 22 weeks	1 blood sample	*Chances* your baby has Down syndrome, trisomy 18, or spina bifida

Diagnostic Tests

Name of Test	When	Description	What It Tells You
Chorionic villus sampling (CVS)	11 to 14 weeks	Sample of placenta, taken through the vagina or abdomen	*Whether or not* your baby has chromosome problems and sometimes other inherited diseases
Amniocentesis (with ultrasound)	16 to 22 weeks	Sample of fluid from around your baby, taken through your abdomen	*Whether or not* your baby has chromosome problems, spina bifida, and sometimes other inherited diseases

Other Tests

Name of Test	When	Description	What It Tells You
Anatomy ultrasound	18 to 22 weeks	Abdominal ultrasound to check baby's growth and development	*Whether or not* abnormalities are suspected and if further testing is needed

Screening Tests

Nuchal Translucency (NOO-kul trans-LOO-sun-see) or NT Ultrasound

This screening test is done between 11 and 14 weeks of pregnancy. Using ultrasound, your baby's length is measured to confirm your due date. Ultrasound is also used to measure the small space under the skin behind your baby's neck. This space is called the *nuchal translucency* (NT). The larger this space of fluid is, the greater the *chance* your baby has a chromosome problem. An NT ultrasound can be done only by specially trained staff.

Integrated (IN-tuh-grey-tud) screen

This test uses the results of the NT ultrasound and 2 blood tests. The first blood sample is taken between 11 and 14 weeks, usually the same day as the NT ultrasound. The 2nd blood sample is taken between 15 and 22 weeks. The blood tests look for patterns of proteins and hormones that are linked to certain birth defects.

An integrated screen tells you the *chances* that your baby has Down syndrome, trisomy 18, or spina bifida. (See the sidebars on pages 34, 35, and 36 for more details.) It does not diagnose these conditions. Most women who get an abnormal integrated screen result still have a healthy baby.

The integrated screen can detect:

- 90 out of 100 cases (90%) of Down syndrome
- 90 out of 100 cases (90%) of trisomy 18
- 80 out of 100 cases (80%) of spina bifida

But, it will **not** detect all cases of these birth defects. And, it does not test for any other health problems.

Quad Screen

This screening test involves 1 blood sample that is taken between 15 and 22 weeks. It's like the integrated screen, because it also looks for patterns of proteins and hormones that are linked to certain birth defects.

A quad screen tells you the *chances* that your baby has Down syndrome, trisomy 18, or spina bifida. It does not diagnose these conditions. Most women who get an abnormal quad screen result still have a healthy baby.

What is a chromosome problem?

Chromosomes are packages of genetic instructions. We inherit them from our parents. They control how our bodies grow and develop. They are in every part of our body, and they can only be seen with a microscope.

Most people have 46 chromosomes, but some people have an extra chromosome (47), a missing one (45), or a chromosome that has a piece broken off or an extra piece attached.

A change in a chromosome often causes birth defects and developmental delay, but not always.

The quad screen can detect:

- 85 out of 100 cases (85%) of Down syndrome
- 75 out of 100 cases (75%) of trisomy 18
- 80 out of 100 cases (80%) of spina bifida

But, it will **not** detect all cases of these birth defects. And, it does not test for any other health problems.

A quad screen may be a good test to have if you do not start prenatal care until your 4th month or if an NT ultrasound is not available.

Advanced Aneuploidy (ann-you-PLOY-dee) Screening with Cell-free DNA

You may have heard about a new blood test that can screen for Down syndrome. This test is called a*dvanced aneuploidy screening with cell-free DNA*. It uses a blood sample from the mother, and it is done starting at 10 weeks of pregnancy. It screens for specific chromosome disorders in the baby.

Everyone has some free (not contained within a cell) DNA in their blood. When you are pregnant, most of that cell-free DNA is from you, but some is from your pregnancy. In this test, the total amount of cell-free DNA from chromosomes 21, 18, and 13 is measured in your blood.

Like the other screening tests, this test does not tell you if the baby has, or does not have, a chromosome problem. But if there is an increased amount of DNA from one of these chromosomes in your blood, there is a high chance that the baby has trisomy for that chromosome.

Currently, only women who have a high risk of having a baby with Down syndrome, trisomy 18, or trisomy 13 can have this test. If you have already had a child with one of these trisomies, or if you have another type of screen and the results are abnormal, you may be offered advanced aneuploidy screening with cell-free DNA.

Diagnostic Tests

Anatomy (uh-NAT-uh-mee) Ultrasound

This test is done between 18 and 22 weeks. An ultrasound is used to look at your baby, the amount of fluid around him, your placenta, and your uterus. It checks to see that the baby is growing and that all major organs are formed.

What is Down syndrome?

Down syndrome is also known as trisomy 21. It is caused when a person has an extra copy of chromosome number 21.

Down syndrome affects people in different ways. People with Down syndrome always look different than other members of their family. They always have some developmental delay, but the level of delay differs with each person.

Adults with Down syndrome may be able to play sports, have a basic job, and enjoy friends. But they usually cannot live on their own without help.

Many babies with Down syndrome have a heart defect, which can sometimes be fixed with surgery. Other health problems and birth defects sometimes occur with Down syndrome, but they are rare.

Your baby is developed enough at this age that an ultrasound may find problems such as a severe heart defect, spina bifida, a missing kidney, and severe cleft lip. Although this test will not diagnose chromosome problems, it may show signs of them or other conditions.

Chorionic Villus Sampling (kor-ee-ON-ic VILL-us sam-pling) or CVS

This diagnostic test is usually done between 11 and 14 weeks. The doctor uses either a thin, flexible needle or a thin plastic tube to remove a small sample of the placenta. An ultrasound is done at the same time, so your baby can be seen during the procedure.

The placenta sample is used to diagnose chromosome problems. If an inherited condition such as *muscular dystrophy* or *hemophilia* runs in your family, the sample can be used to test your baby for that condition.

The chance of miscarriage after CVS is 1 to 2 women in 100 (1% to 2%).

Amniocentesis (AM-nee-oh-sen-TEE-sis) or Amnio

This diagnostic test is usually done between 16 and 22 weeks. The doctor uses a thin, flexible needle to take 2 tablespoons of fluid from around your baby. An ultrasound is done at the same time, so your baby can be seen during the procedure.

The fluid is used to diagnose chromosome problems and spina bifida. If an inherited condition like muscular dystrophy or hemophilia runs in your family, the fluid can be used to test your baby for that condition.

The chance that having an amniocentesis will cause a miscarriage is 1 in 400 women (0.25%).

Ancestry-Based Carrier Screening

Your ancestry, or ethnicity, is one clue to help learn if your baby could have a rare genetic disease. Each ancestral group has conditions that can be inherited that are more common in that group compared to other ethnic groups. The conditions that are linked with each ancestral group are listed in the table on page 36.

Most times, a couple can have a child with one of these disorders only when *both* parents are "carriers" for the *same* disorder. **Carriers usually have no symptoms of the disease.** Also, most carriers have no family history of the disease. If someone in your family has one of these conditions, tell your healthcare provider.

What is trisomy 18?

Trisomy 18 is also known as Edwards syndrome. It occurs when a person has an extra copy of chromosome number 18.

Most babies with this condition do not survive the pregnancy. Children with trisomy 18 have severe brain damage and usually other problems, such as heart defects and clubfoot.

What is spina bifida?

Spina bifida is a condition in which part of the baby's spine does not form normally and the nerves in the spine are damaged. This happens within the first few weeks of pregnancy.

Spina bifida affects people in different ways. Some people have trouble walking and may need to use braces or a wheelchair. Some have trouble controlling their bladder or bowel. Sometimes, spina bifida can cause brain damage and developmental delay.

If you and your partner are both carriers for the same genetic condition, then your baby could inherit that condition. If you want to know for sure before birth, an amniocentesis or a CVS can be done. The integrated screen, quad screen, and ultrasound will **not** diagnose these disorders.

To see if you are a carrier for these hereditary conditions, you will need to give a small blood sample. It is your choice whether or not to have any or all of these tests.

This table is adapted from "Ancestry Based Carrier Screening," published by the National Society of Genetic Counselors, Inc., 2005:

Ancestral Group	Hereditary Condition	Chance of Being a Carrier
African-American	Beta Thalassemia	10% (10 out of 100)
	Sickle Cell Disease	11% (11 out of 100)
Eastern European (Ashkenazi) Jewish	Canavan Disease	2.5% (2 to 3 out of 100)
	Cystic Fibrosis	3% to 4% (3 to 4 out of 100)
	Familial Dysautonomia	3% (3 out of 100)
	Tay-Sachs Disease	3% (3 out of 100)
European Caucasian	Cystic Fibrosis	3% (3 out of 100)
Mediterranean	Beta Thalassemia	3% to 5% (3 to 5 out of 100)
	Sickle Cell Disease	2% to 30% (2 to 30 out of 100)
East and Southeast Asian*	Alpha Thalassemia	5% (5 out of 100)
	Beta Thalassemia	2% to 4% (2 to 4 out of 100)
Hispanic*	Beta Thalassemia	0.25% to 8% (fewer than 1 to 8 out of 100)
	Sickle Cell Disease	0.6% to 14% (fewer than 1 to 14 out of 100)
Middle Eastern and South Central Asian*	Beta Thalassemia	0.5% to 5.5% (fewer than 1 to 6 out of 100)
	Sickle Cell Disease	5% to 25% (5 to 25 out of 100)

** Numbers for this group are estimates and may vary depending on exact ethnicity.*

A genetic counselor can help you and your partner make decisions about prenatal tests.

Deciding Whether to Do These Tests

Choosing whether to have any of these tests, or deciding which ones are best for you, can be hard. There is no "right" choice. Some women choose only an anatomy ultrasound and no other tests. Others may choose an integrated screen and anatomy ultrasound. And, if one of these tests is abnormal, they may have amniocentesis. Some women prefer a CVS or amniocentesis without any of the screening tests.

Making an Informed Decision

Our goal at UW Medicine is to partner with patients and families in making decisions about their care. We encourage you to ask questions to help you to make your decisions.

These are some questions you may want to ask yourself as you think about having genetic testing:

- ❑ Do I want to have any of this information?
- ❑ How would learning about these birth defects before my baby is born help me and my healthcare provider prepare and plan?
- ❑ How would this information help me make choices about my pregnancy if a birth defect is found?
- ❑ Will taking these tests help me feel more reassured?

Your healthcare provider can talk more with you about your choices. Or, you can schedule an appointment in the Prenatal Diagnosis Clinic. Genetic counselors are specially trained to help people think through these questions. They can help you make the decision that is best for you.

Questions?

Your questions are important. If you have questions about prenatal testing, call your health care provider during office hours.

You may also call the Prenatal Diagnosis Clinic: 206-598-4072

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