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.
### Screening Tests

<table>
<thead>
<tr>
<th>Name of Test</th>
<th>When</th>
<th>Description</th>
<th>What It Tells You</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Nuchal translucency (NT) ultrasound</strong></td>
<td>11 to 14 weeks</td>
<td>Abdominal ultrasound to measure small space behind baby’s neck</td>
<td><em>Chances</em> your baby has a chromosome problem</td>
</tr>
<tr>
<td><strong>Integrated screen</strong></td>
<td>11 to 14 weeks <em>and</em>  15 to 22 weeks</td>
<td>NT ultrasound plus 2 separate blood samples</td>
<td><em>Chances</em> your baby has Down syndrome, trisomy 18, or spina bifida</td>
</tr>
<tr>
<td><strong>Quad screen</strong></td>
<td>15 to 22 weeks</td>
<td>1 blood sample</td>
<td><em>Chances</em> your baby has Down syndrome, trisomy 18, or spina bifida</td>
</tr>
</tbody>
</table>

### Diagnostic Tests

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

### Other Tests

<table>
<thead>
<tr>
<th>Name of Test</th>
<th>When</th>
<th>Description</th>
<th>What It Tells You</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Anatomy ultrasound</strong></td>
<td>18 to 22 weeks</td>
<td>Abdominal ultrasound to check baby’s growth and development</td>
<td><em>Whether or not</em> abnormalities are suspected and if further testing is needed</td>
</tr>
</tbody>
</table>
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.
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 advanced 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.
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.
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:

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

* Numbers for this group are estimates and may vary depending on exact ethnicity.
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 healthcare provider during office hours.

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