Most news is good news.

Most babies are born without major birth defects.

Early in your pregnancy, you’ll need to make decisions about prenatal testing. Prenatal tests aim to detect the risk or presence of a birth defect or serious disease in your developing baby.

This guide gives you the facts you need to make decisions about testing. Spend some time with this guide. Take it home and read it carefully. At your next prenatal checkup, ask any remaining questions before making your decisions.
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These 2 pages summarize the prenatal tests described in this guide and show you when each happens during pregnancy.

**TESTING TIMELINE**

*When do different prenatal tests occur during pregnancy?*

- **1st-trimester screening**
  - Done at 10 to 13 weeks.

- **2nd-trimester screening**
  - Done at 15 to 22 weeks.

- **Integrated screening and sequential screening**
  - Combine results of 1st- and 2nd-trimester material serum screening.

- **Cell-free DNA (cfDNA) screening**
  - Can be done any time after 10 weeks.

- **CVS**
  - Usually done between 10 and 13 weeks.

- **Amniocentesis**
  - Usually done between 15 and 20 weeks.

- **Carrier screening** for cystic fibrosis (CF), spinal muscular atrophy (SMA), and other conditions
  - Testing can be performed any time.
  - Best time is before pregnancy or during the first 12 weeks.

About 40 weeks before birth.
Maternal serum screening options —
more on pages 6 and 7

- **Maternal serum screening** is a blood test that can be done in the 1st trimester only, the 2nd trimester only, or as a 2-part test (integrated or sequential screening):
  - **1st-trimester screening:** Paired with a specialized ultrasound, results from this blood test calculate your chance of having a baby with Down syndrome or trisomy 18.
  - **2nd-trimester screening:** Results from this blood test calculate your chance of having a baby with Down syndrome, trisomy 18, or a neural tube defect (NTD).
  - **Integrated screening:** This test is a combination of 1st- and 2nd-trimester screening. It looks at the 2 results together to create a clearer picture of your baby’s risk for Down syndrome, trisomy 18, or a neural tube defect (NTD).
  - **Sequential screening:** This test option is a stepwise version of integrated screening. It uses 1st trimester results to help women choose to either continue with 2nd-trimester serum screening or have additional 1st trimester testing.

- These screenings do NOT tell you whether your baby actually has—or will have—Down syndrome, trisomy 18, or a neural tube defect (NTD). Rather, the tests place you in a higher or lower risk category for a pregnancy affected by one of these disorders.

Cell-free DNA (cfDNA) screening —
more on pages 8 to 9

- **Cell-free DNA (cfDNA) screening** is a test used to detect chromosomal abnormalities (genetic changes) such as Down syndrome, trisomy 18, trisomy 13, and Turner syndrome. In high-risk women, the test identifies nearly all cases.
- CfDNA screening is currently offered to women who are known to have a higher risk for chromosome abnormalities in their pregnancies. With time and additional research, this test may be used more broadly to detect even more conditions.

Chorionic villus sampling (CVS) —
more on pages 10 to 12

- **Chorionic villus [KOHR-ee-ON-ik VIL-uhs] sampling (CVS)** requires a small sample of the placenta, which is found in the lining of the uterus. The sample is then tested in a lab.
- Results tell you whether your baby has Down syndrome or trisomy 18. CVS can also check for other genetic disorders such as cystic fibrosis (CF).
- In very rare cases, CVS can result in pregnancy loss (miscarriage).
- CVS is accurate in identifying genetic disorders in 99 out of 100 pregnancies.

Amniocentesis —
more on pages 10 to 12

- **Amniocentesis [am-nee-oh-sen-TEE-sis]** requires a small sample of amniotic fluid (the fluid surrounding your developing baby). The sample is then tested in a lab.
- Results tell you whether your baby has a neural tube defect (NTD) or a genetic disorder such as Down syndrome or trisomy 18. Amniocentesis can also check for other genetic disorders such as cystic fibrosis (CF).
- Amniocentesis has a very low risk of miscarriage (pregnancy loss).
- Amniocentesis is accurate in identifying genetic disorders in 99 out of 100 pregnancies.

Carrier screening for cystic fibrosis (CF), spinal muscular atrophy (SMA), and other conditions —
more on pages 13 and 14

- **Carrier screening** involves testing a sample of your blood or saliva. It can tell whether you carry an altered gene (piece of DNA) that causes a genetic condition.
- If you are found to be a carrier, your partner will also be offered carrier testing. Results from both partners allow your doctor or genetic counselor to determine the risk to your baby.
- Results apply not only to this pregnancy, but also to any future pregnancy that you and your partner have together.
RISK GROUPS — not certainties
Screenings assess the risk of having a baby with birth defects, not the actual presence of birth defects.

• Low risk means your baby is less likely to have the problem. It does not mean the baby absolutely will not have it.

• High risk means your baby is more likely to have the problem. It does not mean the problem is certain. If the screening shows a high risk and the baby is born without the problem, it is called a false positive.

Serum screening is a popular choice for many pregnant women even though results of the test can be uncertain. Integrated screening, in particular, has a high detection rate for Down syndrome and a fairly small false positive rate.

Tests

Options for screening and testing
This section gives more information about the prenatal testing options listed on the previous pages. As you read, keep in mind that you won’t need to have every test described here. (Some of them check for the same conditions.) Also, remember that no test is perfect. No test is 100% accurate, nor is it possible to test for every condition.

Maternal serum screening options
Maternal serum screening is a blood test to determine whether there’s a high chance or a low chance that your pregnancy is affected by a condition such as Down syndrome, trisomy [TRY-soh-mee] 18, or an open neural tube defect (NTD) such as spina bifida [SPY-nuh BIF-i-duh]. See pages 18 and 19 for more information about these conditions.

There are several types of maternal serum screening. The best option for you depends on when you want the results and how important accuracy is to you. Maternal serum screening can be done in the 1st trimester only, in the 2nd trimester only, or as an integrated screen that combines information from the 1st- and 2nd-trimester screenings. See the table on the next page to compare the accuracy and timing of these 3 options.

If you choose the 1st-trimester screen or the integrated screen, your doctor or midwife may refer you to a Maternal-Fetal Medicine office for the ultrasound. (The 1st-trimester screen requires a specialized ultrasound that includes a measure of the clear tissue at the back of the baby’s head, called the nuchal translucency.) You may also meet with a genetic counselor to discuss the screening. If you choose the 2nd-trimester screening by itself — called the quad screen — you can have the blood drawn at your doctor’s or midwife’s office.

What if I have a screen positive result?
A screen positive result places you in a higher risk category for having a baby with a birth defect. However, it’s important to know that most pregnancies that “screen positive” from maternal serum screening result in healthy, unaffected babies. Additional testing can determine if your baby is one the few that will truly have a problem.

After further ultrasound evaluation of your pregnancy, you may choose to have additional screening with a new blood test called cell-free DNA (cfDNA) screening or to have diagnostic testing via chorionic villus sampling (CVS) or amniocentesis. Each of these tests is explained in the following pages.

Whether or not you decide to have more testing, it’s a good idea to speak with your doctor, midwife, and/or a genetic counselor about your screening results. They can help you better understand the meaning of your results and which options would work best for your family.
What is a pregnancy ultrasound?

In pregnancy, ultrasound (sonography) is a way to capture images of your developing baby. It creates pictures by bouncing special sound waves off body structures. Doctors use ultrasound to measure the baby’s size, check how far along the pregnancy is (gestational age), and look for some types of problems.

<table>
<thead>
<tr>
<th>OPTIONS for maternal serum screening</th>
<th>DETECTION rates (the number of times that the screening finds when a problem exists)</th>
<th>FALSE POSITIVE rates (the number of times that screening suggests “high risk” when the baby is actually unaffected)</th>
<th>TIMING of the screening</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>1st-trimester screen</strong></td>
<td>• Down syndrome: 85 out of 100&lt;br&gt;• Trisomy 18: 80 out of 100</td>
<td>6 out of 100</td>
<td>10–13 weeks of pregnancy</td>
</tr>
<tr>
<td>Requires a single draw of your blood and an ultrasound of your baby. The ultrasound allows for a measure of the fluid behind the baby’s neck (nuchal translucency). This measurement, when combined with information from the chemical markers in your blood, helps provide a risk estimate for certain birth defects in your pregnancy.</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>2nd-trimester screen</strong>&lt;br&gt;(also called the quad screen when done on its own)</td>
<td>• Down syndrome: 81 out of 100&lt;br&gt;• Trisomy 18: 80 out of 100&lt;br&gt;• Open NTDs: 80 out of 100</td>
<td>4 to 5 out of 100</td>
<td>15–22 weeks of pregnancy</td>
</tr>
<tr>
<td>Requires a single draw of your blood, which is then analyzed for 4 chemical markers.</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Integrated screen and sequential screen</strong></td>
<td>• Down syndrome: 87 out of 100&lt;br&gt;• Trisomy 18: 90 out of 100&lt;br&gt;• Open NTDs: 80 out of 100</td>
<td>1 to 2 out of 100</td>
<td>• part 1: 10–13 weeks&lt;br&gt;• part 2: 15–22 weeks</td>
</tr>
<tr>
<td>Results are obtained from information from both 1st- and 2nd-trimester screening tests.</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**for more information…**

See pages 18 and 19 to learn more about Down syndrome, trisomy 18, and neural tube defects (NTDs).
Cell-free DNA (cfDNA) screening

Cell-free DNA (cfDNA) screening, also called non-invasive prenatal testing (NIPT), is a recently developed test to help detect certain genetic problems in a pregnancy. It tests for Down syndrome, trisomy 18, trisomy 13, and Turner syndrome.

This test requires a sample of your blood. It works by examining small fragments of DNA—from your own cells as well from the pregnancy—that are normally present in your blood. Pregnancies affected with chromosome abnormalities have an abnormally high amount of fetal DNA in the maternal blood sample. This test also looks for DNA from the Y chromosome, which can determine whether the baby is male or female.

CfDNA screening uses blood drawn any time after 10 weeks gestation and gives results in about 7 to 10 days.

Who can have cfDNA screening?

This test is available to any pregnant woman. However, cfDNA screening is less accurate in women younger than 35 years. These women are low-risk and a screen positive is more likely to be a false positive. Women older than 35 (or those with another risk discussed below) are higher risk and a screen positive result is more likely to be accurate. At this time, most insurance companies will only cover cfDNA screening in women:

- Who will be 35 years or older when their baby is born
- Who have had a “screen positive” result from maternal serum screening or certain findings in a pregnancy ultrasound
- Whose previous pregnancy was affected by a chromosome abnormality.

Experts think that with time and further development of cfDNA screening, insurance coverage will increase to all women regardless of risk.

How accurate is cfDNA screening?

For singleton (1 baby) pregnancies that are known to be at higher risk for chromosome abnormalities, cfDNA screening is highly accurate. CfDNA screening can detect 99% of pregnancies affected by Down syndrome. The detection rate is also very high for trisomy 13, trisomy 18, and Turner syndrome. The test doesn’t detect open neural tube defects.

The accuracy of cfDNA screening for twin pregnancies is not as well understood, but the results can tell if there’s a higher risk for one or more of the babies to have a disorder. Currently, no information is available about the test’s accuracy for pregnancies with triplets or other multiples.
When thinking about cfDNA screening results, keep in mind that although a positive result means a very high chance that the pregnancy is affected—and a negative result decreases the chance to less than 1 out of every 100—this test is not 100% accurate. It’s a screening test, not a diagnostic test. Results that show a high risk for one of these disorders can only be confirmed with CVS or amniocentesis.

Since cfDNA screening is a new technology that is rapidly developing, it may soon be able to look for conditions other than the major chromosome abnormalities. Ask your doctor or genetic counselor if you are interested in discussing other conditions included in this test.

**What are the risks?**

There is no risk of miscarriage from cfDNA screening. There is a small chance that the lab will not be able to report a result from the sample and that they will request another one. In this case, you may send another blood sample or opt for a different kind of testing.

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**GENETIC BASICS:**

**terms and facts related to genes and genetic disorders**

- Almost every cell in a person’s body contains DNA. DNA is the chemical that makes up genes.
- **Genes** act as instruction manuals to tell our bodies how to grow, develop, and stay healthy. Genes also determine our personal characteristics.
- Genes are packaged together into structures called chromosomes. Most people have 23 pairs of chromosomes, which are numbered 1–22. The last pair consists of the sex chromosomes (X and Y), which determine whether the baby is a boy or a girl.
- Babies usually inherit one copy of each chromosome pair from each parent. Sometimes, a mistake in this process can lead to an extra or missing chromosome in the developing baby. Babies that have extra or missing chromosomal material often have birth defects or developmental problems. Common chromosomal disorders include Down syndrome, trisomy 18 and 13, and Turner syndrome.
- Some **genetic disorders** are caused by a mutation (change) in a single gene, which is usually inherited from parents who are carriers. Cystic fibrosis (CF), Tay-Sachs disease, and sickle cell anemia are all inherited genetic conditions. Knowing about these conditions in pregnancy or in the newborn period can help families make good decisions and may allow for specialized care.
Chorionic villus sampling (CVS) and amniocentesis

Chorionic [KOHR-ee-on-ik] villus [VIL-uh-s] sampling (CVS) and amniocentesis are similar in many ways. Both are highly accurate ways to check for genetic disorders. They are both called invasive tests because they require going inside the mother’s womb (uterus) to get the test sample. This section describes CVS and amniocentesis, their similarities, and their differences.

What is CVS?

CVS is a prenatal test that’s usually done between 10 and 13 weeks of pregnancy. It involves testing a sample of the placenta, the tissue that joins you and your developing baby.

CVS routinely checks for genetic disorders like Down syndrome and trisomy 18. It can also test for other genetic disorders like cystic fibrosis (CF). Talking to your doctor or genetic counselor will help determine whether CVS is right for you and what specific conditions it should test for.

How is CVS performed, and what does it tell me?

During CVS, a doctor inserts a hollow needle into the placenta. (To reach the placenta, the doctor can either go through your abdomen into your uterus or through the vagina and cervix.) Ultrasound shows the doctor where to safely put the needle. Once the needle is in place, the doctor withdraws a small sample of the placenta. The needle is then taken out, and the sample is sent to a laboratory for testing.

It usually takes about 7 days to get your results from CVS—though some results are returned within 2 days. The results are highly accurate. With more than 99% accuracy, CVS can tell you whether your baby has a genetic disorder.

What are the risks of CVS?

CVS can cause cramping, bleeding, and infection. It also has a very low rate of pregnancy loss (miscarriage). Miscarriage happens in about 1 out of every 300 to 500 procedures done.
What is amniocentesis?

Amniocentesis is a test usually performed between 15 and 20 weeks of pregnancy. It involves testing a sample of the amniotic fluid that surrounds your developing baby.

Amniocentesis routinely checks for neural tube defects (NTDs) and genetic disorders such as Down syndrome and trisomy 18. It can also test for other genetic disorders like cystic fibrosis (CF). Talking to your doctor or genetic counselor will help determine whether amniocentesis is right for you and what specific conditions it should test for.

How is amniocentesis performed, and what does it tell me?

During amniocentesis, a doctor inserts a hollow needle through your abdomen into your uterus. Ultrasound shows the doctor where to safely put the needle. Once the needle is in place, the doctor withdraws a small amount of fluid. The needle is then taken out, and the sample is sent to a laboratory for testing.

It takes about 10 to 14 days to get your results from amniocentesis. The results are highly accurate. With more than 99% accuracy, amniocentesis can tell you whether your baby has a genetic disorder.

What are the risks of amniocentesis?

Amniocentesis can cause cramping, bleeding, and infection. It can cause leaking of the amniotic fluid. Amniocentesis has a very low rate of miscarriage (pregnancy loss). Miscarriage happens in about 1 out of every 300 to 500 procedures done.
Who usually has CVS or amniocentesis?

Women who choose CVS or amniocentesis are often those who:

- Are older at the time of the pregnancy, as the risk of genetic disorders increases with the mother’s age
- Have a family history of a disorder that can be detected by CVS or amniocentesis
- Have had prenatal screening results suggesting an increased risk of a genetic disorder
- Seek reassurance about their pregnancies

How do you choose between these 2 tests?

Many women are choosing between CVS and amniocentesis. Both tests offer reliable information about whether a pregnancy is affected by a genetic disorder.

One possible advantage of CVS is timing. (See the sidebar at left.) An advantage of amniocentesis is that it can help detect neural tube defects.

What if I get abnormal results?

If CVS or amniocentesis reveals a birth defect, try to learn as much as you can about the condition and your options. Talk to your doctor or genetic counselor. You may also want to talk to parents of children with the same disorder. Learning as much as you can will help you make the best decision for the future.
Carrier screening for cystic fibrosis (CF), spinal muscular atrophy (SMA), and other conditions

Carrier screening tells you whether you “carry” a gene (DNA) mutation that would mean an increased risk for your children to inherit a disease. The 2 most common carrier screens done in the U.S. are for cystic fibrosis (CF) and spinal muscular atrophy (SMA). Other carrier screening may be recommended based on your family history and ethnic background. Expanded carrier screening is also available and can give you information about the possibility of many other inherited conditions. These tests are explained below.

Carrier screening for CF and SMA

Carrier screening looks at the risk of you and your baby’s father having a child with CF or SMA. These are serious genetic disorders which can shorten your baby’s life and require extra treatments and medical care. There is no cure for these conditions. For more information on CF and SMA, and your risk for having a child with these conditions, see pages 15 and 16.

How is the test performed, and what does it tell me?

CF and SMA carrier screening is done in a laboratory using a sample of your blood or saliva. It can be done at any time, even if you’re not pregnant. If you choose this test during pregnancy, it’s best to have it during the 1st trimester. That way, you’ll have more time to make decisions about the pregnancy. The carrier test has no risks to you or to the pregnancy.

The carrier test looks to see if you carry the gene mutations that cause CF and SMA. If the test shows that you do, the baby’s father is tested as well. Your baby is at greater risk of having this condition only if both you and the father are carriers. In this case, there is a 1 in 4 chance of your baby having the condition. (Your own health is not affected.)

What if results say that we’re both carriers?

If you and the baby’s father are both carriers, you can choose to test your baby during pregnancy. Amniocentesis and CVS—prenatal tests described earlier in this guide—can tell whether the developing baby actually has CF or SMA. Or, you may choose to wait until after birth for this information. All newborns are routinely screened for certain disorders, including CF. These screenings detect most, but not all, cases of CF. For a diagnosis of CF in a newborn, further testing is required. SMA is not included in routine newborn screenings.

MORE FACTS

• Since the test is about you and the father’s status as carriers, the results apply to any pregnancy you have together—not just this one. For example, if you’re both carriers of the same condition, any children you have in the future will face the same 25% chance of inheriting that condition.

• The carrier test will find most—but not all—carriers. The test’s accuracy may be lower in certain ethnic groups.

• You may want to check with your health insurance provider to find out if carrier screening is covered.
Who might choose CF carrier testing?
As with any testing, the decision to have carrier screening is personal. You might be influenced by your view of the risk based on ethnic background or family history (see page 15).

Expanded carrier screening
CF and SMA carrier screening only tests for those individual conditions. An expanded carrier screening test can tell you about the possibility of many different inherited conditions in your children. It’s available to all women of reproductive age and may be valuable for couples with different ethnic backgrounds, a family history of a genetic condition, or when limited information about family history is known (such as when a parent is adopted).

How is the test performed, and what does it tell me?
Like CF and SMA carrier screens, expanded screening is done in a laboratory, using a sample of your blood or saliva. It can be done at any time, even if you’re not pregnant. The test poses no risk to you or your pregnancy.

The expanded carrier test looks to see if you carry the gene mutations that can cause genetic conditions in your child. The test will typically include screening for genes related to CF, SMA, fragile X syndrome, Tay-Sachs disease, sickle cell anemia, and many other hereditary diseases. If you’re interested in expanded carrier screening, your doctor or genetic counselor can explain more information about the types of conditions that are likely to be included on your panel.

Keep in mind that most of these conditions are extremely rare. However, it’s fairly common to identify carriers of these conditions with this screening test. If you’re found to be a carrier, your partner will also be screened if he wasn’t screened earlier.

What if results say that we’re both carriers?
If both you and the baby’s father carry the genetic change for the same disease, your doctor or genetic counselor will review the results and your options with you. A positive carrier test result for both of you could mean there is a higher chance for your baby to be affected, but only further testing could tell if this particular pregnancy is affected. (Remember that being a carrier doesn’t affect your own health.)

Finally, keep in mind that expanded carrier screening doesn’t test for every health condition or birth defect. So although normal results from this test are reassuring, they don’t completely eliminate the chance for a genetic condition or birth defect in your baby.
Conditions

Diseases and disorders discussed in this guide

This section summarizes some of the conditions that prenatal testing may detect. It also discusses some of the factors that affect your risk of having a baby with one of these conditions.

Cystic fibrosis (CF)

Cystic fibrosis (CF) is one of the most common genetic diseases in families of European descent (Caucasian families). CF causes severe problems with breathing and digestion. It cannot be cured. It requires daily, lifelong treatment.

In the past, children with CF rarely lived to adulthood. Today, newer treatments allow people with CF to live longer and more comfortably.

What affects my chances of having a baby with CF?

Your chances of having a baby with CF depend on whether you and the baby’s father carry the gene mutation that causes CF. This, in turn, may depend on your ethnic background. See the table below to get an idea of how common CF is in your ethnic group.

Family history is also a factor. If someone in your family has CF—or is known to be a carrier of the CF gene—you have an increased risk of having a baby with CF. Still, most couples who have a child with CF have no known family history of the disease.

<table>
<thead>
<tr>
<th>Ethnicity / Race</th>
<th>Chance of being a CF carrier</th>
<th>Babies born with CF in the US each year</th>
</tr>
</thead>
<tbody>
<tr>
<td>European Caucasian</td>
<td>1 in 29</td>
<td>1 out of 3,300</td>
</tr>
<tr>
<td>Hispanic American</td>
<td>1 in 45</td>
<td>1 out of 9,000</td>
</tr>
<tr>
<td>African American</td>
<td>1 in 65</td>
<td>1 out of 15,300</td>
</tr>
<tr>
<td>Asian American</td>
<td>1 in 90</td>
<td>1 out of 32,000</td>
</tr>
</tbody>
</table>

Regardless of ethnicity, both parents must be carriers of the CF gene for their baby to have CF. If both parents are carriers, their baby has a 1 in 4 chance of having CF.
Spinal Muscular Atrophy (SMA)

Spinal muscular atrophy (SMA) is a disease in which certain nerves in the brain and spinal cord die, leading to muscle weakness and limiting movement. SMA cannot be cured. Some babies with SMA will pass away in the first months or years of life. In other people with SMA, muscle weakness may not begin until adulthood (20s to 30s) and lifespan can be normal. It’s not always possible to predict the type of SMA a child could have based on the SMA testing.

What affects my chances of having a baby with SMA?

In the U.S., 1 in every 6,000 to 10,000 people develop SMA. It’s the most common cause of infant death. Just as with CF, your chances of having a baby with SMA depend on whether you and the baby’s father carry the gene mutation that causes SMA. The condition is more common in certain ethnic backgrounds, although it has been found in people of every ethnicity. See the table below to get an idea of how common SMA is in your ethnic group.

<table>
<thead>
<tr>
<th>Ethnicity / Race</th>
<th>Chance of being an SMA carrier</th>
</tr>
</thead>
<tbody>
<tr>
<td>European Caucasian</td>
<td>1 in 35</td>
</tr>
<tr>
<td>Hispanic American</td>
<td>1 in 117</td>
</tr>
<tr>
<td>African American</td>
<td>1 in 66</td>
</tr>
<tr>
<td>Asian American</td>
<td>1 in 53</td>
</tr>
</tbody>
</table>

Regardless of ethnicity/race, both parents must be carriers of the SMA gene for their baby to develop SMA. If both parents are carriers, their baby has a 25% chance (1 in 4) of having SMA.
Turner syndrome

Turner syndrome is a chromosome abnormality that affects girls who inherit only 1 copy of the X chromosome instead of 2. Girls with Turner syndrome may be born with heart defects requiring surgery, and generally, they are short compared to their family members. They may have learning disabilities, but most girls with Turner syndrome have normal intelligence. When a woman is pregnant with a baby with Turner syndrome, the pregnancy has a very high chance of ending in miscarriage.

What affects my chances of having a baby with Turner syndrome?

Turner syndrome is one of the most common chromosome abnormalities in girls. Researchers estimate that the syndrome occurs in about 1 in every 2,000 to 2,500 live female births. The actual occurrence is unknown since girls with a mild form of the condition may not be diagnosed, or may be diagnosed later in life. Turner syndrome occurs at similar rates in all ethnic groups and in different countries.
Down syndrome

Down syndrome is a genetic disorder and the most common form of mental disability. It’s caused by an extra copy of chromosome 21 in a developing baby. Children with Down syndrome tend to reach developmental milestones more slowly than their peers and have a higher risk of being born with a heart or intestinal problem requiring surgery. Some adults with Down syndrome are able to hold a job and live independently. However, others with more severe disabilities require significant assistance. Life expectancy for people with Down syndrome is close to that of the general population.

What affects my chances of having a baby with Down syndrome?

Your risk for having a baby with Down syndrome is linked to your age during pregnancy. The older you are, the greater your chance of having a baby with Down syndrome (see the table below). Having a family history of Down syndrome may also increase the chance.

<table>
<thead>
<tr>
<th>Mother’s age</th>
<th>Chance of having a live baby with Down syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>20</td>
<td>1 in 1,667</td>
</tr>
<tr>
<td>25</td>
<td>1 in 1,250</td>
</tr>
<tr>
<td>30</td>
<td>1 in 952</td>
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<tr>
<td>33</td>
<td>1 in 602</td>
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<tr>
<td>35</td>
<td>1 in 378</td>
</tr>
<tr>
<td>37</td>
<td>1 in 224</td>
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<td>40</td>
<td>1 in 106</td>
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<td>1 in 82</td>
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<td>1 in 63</td>
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<td>43</td>
<td>1 in 49</td>
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<tr>
<td>44</td>
<td>1 in 38</td>
</tr>
<tr>
<td>45</td>
<td>1 in 30</td>
</tr>
</tbody>
</table>

Trisomy 18 and trisomy 13

Trisomy 18 and trisomy 13 are disorders caused by an extra chromosome (chromosome number 18 and number 13, respectively). Both disorders are much more severe than Down syndrome. Children affected by trisomy 18 or trisomy 13 are severely disabled, and most don’t survive their first year of life.

What affects my chances of having a baby with trisomy 18 or 13?

As with Down syndrome, the chance for this disorder increases with the mother’s age. However, the risk numbers are lower overall for these more rare conditions.
Neural tube defects (NTDs)

**Neural tube defects (NTDs)** are caused by problems in the development of the baby’s brain, skull, backbone, or spinal cord during the early part of pregnancy. Two more common NTDs are **spina bifida** and **anencephaly** [an-en-SEF-uh-lee]:

- **Spina bifida.** The effects of spina bifida vary a lot from person to person. Some people have only mild problems. Others have leg paralysis and lack of bladder or bowel control. Some people with spina bifida are intellectually disabled.

- **Anencephaly.** With this severe defect, the brain and head don’t develop normally. Nearly all babies with anencephaly are stillborn or die soon after birth.

What affects my chances of having a baby with an NTD?

Anyone can have a child with an NTD. Since NTDs are probably caused by many factors, it’s difficult to know the risk for any single pregnancy. Still, scientists have learned that:

- Most babies with NTDs are born to women without a family history of the disease. However, having this history does increase your baby’s risk for an NTD. Having diabetes or a seizure disorder treated by certain medications also increase your baby’s risk.

- Women who take a vitamin with 400 mcg (micrograms) of folic acid before and during their pregnancy have a much lower risk of having a baby with an NTD or another birth defect.

Folic Acid Lowers Risk

Take a vitamin with at least 400 mcgs (micrograms) of folic acid every day...

- Before you’re pregnant
- During your pregnancy
- After you’re pregnant
- Always—if you have any chance of getting pregnant, on purpose or accidentally

Folic acid is important to help prevent NTDs in a developing baby. It’s also good for your health. So if you have any chance at all of becoming pregnant, take a pill!

For more information...

- National Down Syndrome Society
  [www.ndss.org](http://www.ndss.org)
  800-221-4602

- Support Organization for Trisomy 18, 13 and Related Disorders
  [www.trisomy.org](http://www.trisomy.org)
  1-800-716-7638

- Spina Bifida Association of America
  [www.sbaa.org](http://www.sbaa.org)
  800-621-3141

- Utah Fetal Center
  [intermountainhealthcare.org/locations](http://intermountainhealthcare.org/locations)
  844-693-3825
Considerations

**Things to think about as you decide about testing**

The tips on this page can help you weigh your options and find more information if you need it.

**Who you can talk to**

It often helps to talk to others about your prenatal testing options. Your doctor or midwife can probably answer many of your questions. But you may also want to talk to a genetic counselor.

A **genetic counselor** is a medical professional who can analyze your family’s medical history and help you understand your risk of having a baby with a particular disorder. A counselor can also clarify the pros and cons of different prenatal tests and help you interpret your results. Talk to your doctor if you're interested in meeting with a genetic counselor.

**Questions to ask yourself**

As you make decisions about testing, consider the questions below.

- What do I hope to learn from this test? Can the test give me what I'm looking for?
- How will I cope with waiting for results, or with any uncertainty in the results?
- Does the value of the test (the information it gives me) outweigh any risks?
- How do I interpret my risk for having a child with this disease or disorder? (Risk can be based on such things as your age, ethnic group, family history, etc.)
- How serious does this condition seem to me?
- What if the test identifies problems? What decisions might the news affect, and what choices might I make? For example, I may need to make decisions about:
  - Further testing or monitoring during the pregnancy
  - Continuing the pregnancy
  - Creating a delivery plan that works for me
  - Arrangements for special care or treatment at delivery
  - Plans for caring for a child with special needs
  - Meeting with a specialist to help coordinate care
Questions for my doctor

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Verification of Education

It’s important that you understand the information in *A Guide to Prenatal Testing.*

If you have any questions about the tests described in this guide, please ask your healthcare provider or genetic counselor before signing this form.

- I have read and I understand the information in *A Guide to Prenatal Testing.*

I plan to choose the following:

- Maternal serum screening
  - [ ] 1st trimester
  - [ ] 2nd trimester
  - [ ] integrated / sequential
- Cell-free DNA (cfDNA) screening
- Chorionic villus sampling (CVS) (*separate consent form required*)
- Amniocentesis (*separate consent form required*)
- Cystic fibrosis (CF) carrier testing
- Spinal muscular atrophy (SMA) carrier testing
- Expanded carrier screening
- None of the above

__________________________________________
Signed

__________________________________________
Date