

your choices

for prenatal testing





During your pregnancy, you can choose to have prenatal tests that check for birth defects. Some of these birth defects include:

- Down syndrome (a chromosome defect that causes intellectual disability)
- Trisomy 18 (a chromosome defect that causes intellectual disability and other severe problems)
- Spina bifida (an opening in the spine)

Babies can be born with these birth defects even when the mother is healthy and no one else in the family has had them before. Depending on your age, your medical and family history, and how far along you are in your pregnancy, you will have different tests available to you. However, no test can find all defects before birth, and some disabilities do not appear for months or years after birth.

There are two kinds of tests: **screening tests** and **diagnostic tests**.

Screening tests

These include California Prenatal Screening, non-invasive prenatal testing (NIPT), and ultrasound. These tests tell you the chance that your baby has certain birth defects. They cannot tell you for sure if your baby has a problem. If your screening test shows a higher risk, you will have to decide whether you want to have a diagnostic test.

Diagnostic tests

These include chorionic villus sampling (CVS) and amniocentesis. These tests tell you if your baby actually has the specific birth defect for which you were tested.

These tests are optional and voluntary. You do not have to have them if you don't want them. If the results of either the screening or diagnostic tests are abnormal, you will be offered an appointment with a genetic counselor to discuss your options. Please talk to your obstetrician-gynecologist nurse or physician if you want more information about these tests to help you decide.

Genes, Chromosomes, and Birth Defects

Genes are made up of DNA and packaged into chromosomes. Chromosomes can only be seen under a microscope. Genes determine hair color, eye color, and other physical features we can see, as well as those we cannot see, such as blood type. When a baby is conceived, 23 chromosomes come from the mother's egg and 23 come from the father's sperm. Every normal human cell has 46 chromosomes, and each contains many genes.

Birth defects cause abnormalities in the physical or chemical makeup of a newborn. They can affect the child's looks, organs, or metabolic function (the complex system of chemicals that keeps us alive). Some babies are born with more than one birth defect.

Birth defects can be inherited (genetic), acquired (environmental), or a combination of both. Inherited disorders are passed from parents to their children in their genes. Acquired disorders can be caused by infection, certain drugs, chemicals (such as alcohol), or radiation. For many birth defects, a specific cause is not known.



Certain birth defects can be found by looking for abnormalities (changes) in the structure or number of chromosomes in the cells of the unborn baby. These can usually be seen in the cells in the tissue of the developing placenta or within the amniotic fluid (“water” in the sac that surrounds the fetus).

Down syndrome is the most common chromosome abnormality found in prenatal diagnosis. It is one of the most common forms of intellectual disability. Children with Down syndrome may also have other health problems, such as heart defects. Down syndrome does not usually run in families. It occurs because the baby has an extra copy of chromosome 21 (three copies instead of the usual two). Couples of all ages, races, and incomes can have a baby with Down syndrome. Other examples of chromosome abnormalities include trisomy 18 and 13, which are rarer and more severe than Down syndrome.

Neural tube defects, like spina bifida, are another common group of birth defects. They result when the brain or spinal cord does not develop properly. These birth defects occur in about 1 or 2 out of 1,000 newborns. Children with spina bifida may not be able to walk or control their bowels or bladder. They may also have excess fluid in the brain, which may cause problems in the way they develop.

When Is Prenatal Diagnosis Offered?

You and your physician or other health care professional should discuss whether prenatal diagnosis is right for you. Each case is different. It is usually offered:

- To women who will be 35 or older at the time of delivery because they have an increased risk of giving birth to children with Down syndrome and other chromosomal disorders
- When there is an abnormal blood screening test (California Prenatal Screening or NIPT) in the first or second trimester of pregnancy
- When a birth defect is suspected during an ultrasound exam
- If either parent has already had a child with a chromosome disorder
- If a couple has already had a child with a neural tube defect
- When the baby is at an increased risk for a known genetic or chromosomal condition
- To a woman pregnant with twins or more, even if she is younger than 35

Some women choose to have prenatal diagnosis for reassurance, even when they do not have any known risk factors for birth defects.

If you are interested in prenatal diagnosis, you need to meet with a genetic counselor. This gives you a chance to discuss the risks, benefits, limitations, and alternatives with an expert. Genetic counselors are health care professionals specially trained in inherited diseases, birth defects, and testing for these conditions.



Diagnostic Tests

A diagnostic test (chorionic villus sampling or amniocentesis) shows whether or not the fetus actually has a specific birth defect.

Chorionic Villus Sampling

CVS is a test that can find chromosomal defects during the first trimester of pregnancy (at 10 to 13 weeks). The test involves removing a small sample of tissue called “chorionic villi” from the growing placenta.

In rare cases when a tissue sample cannot be taken or results are not clear, your physician will recommend amniocentesis later in pregnancy.

Because CVS does not find neural tube defects, a second trimester blood test should be done between 15 and 20 weeks of pregnancy.

Amniocentesis

Amniocentesis tests the fluid in the amniotic sac around your baby. This test can find certain birth defects during the second trimester of pregnancy, usually between 15 and 20 weeks after the first day of your last menstrual period. Using ultrasound as a guide, the physician inserts a thin needle through the abdomen into the uterus and removes a very small amount of fluid from the amniotic sac to test. The fetus replaces this fluid in less than 24 hours. Amniocentesis is not painful for most women.

Amniocentesis also detects most neural tube defects by measuring the level of alpha-fetoprotein (AFP) in the amniotic fluid.

The CVS and Amniocentesis Procedures

An ultrasound is used to examine the fetus and to guide the physician. It shows the gestational age of the fetus and the location of the fetus and placenta.

Cells from the sample are grown in the laboratory. The chromosomes are studied under a microscope to look for any abnormalities. Results are available in about two weeks. CVS and amniocentesis can find more than 99 percent of chromosome defects. When a couple is known to be at risk for having a child with certain conditions, other tests may be done.

In most cases, the results of CVS and amniocentesis are normal and reassuring. If the test finds an abnormality, you will have the choice whether or not to continue the pregnancy.

Complications After Amniocentesis and CVS

Minor complications, such as cramping, spotting, slight leakage of amniotic fluid, and infection, can occur. More serious complications occur in between 1 in 300 and 1 in 500 procedures. They can include miscarriage, bleeding, infection, leakage of amniotic fluid, or premature birth.

Because of the higher rate of miscarriage that naturally occurs between 9 and 16 weeks of pregnancy, the overall miscarriage rate after CVS is slightly higher than the rate after amniocentesis.

Screening Tests

You can start with a screening test to help you decide whether you want to have a diagnostic test. Screening tests estimate the chance that the baby has certain birth defects. The tests are optional and voluntary.

If a screening test shows that follow-up is recommended, you will meet with a genetic counselor to discuss diagnostic testing (CVS and amniocentesis).



Sometimes the screening result is positive, but the baby **does not** have Down syndrome or another birth defect. Sometimes the screening result is negative, but the baby **does** have Down syndrome or another birth defect.

*California Prenatal Screening Test (10 to 13 weeks 6 days of pregnancy and 15 to 20 weeks)**

All pregnant women in California are offered testing for certain birth defects through the California Prenatal Screening Program. This program is considered the standard of care for women who want a screening test for certain chromosome problems and birth defects.

*Refer to the California Prenatal Screening Program booklet for more information.



This test combines the results of a blood test in the first trimester (between 10 weeks 0 days and 13 weeks 6 days) with the results of a blood test in the second trimester (between 15 weeks 0 days and 20 weeks 0 days).

If you have a first trimester ultrasound measuring the fluid-filled area at the back of the baby's neck (nuchal translucency, or NT), you will get a preliminary result for Down syndrome and trisomy 18. If you do not have an NT measurement, your results will not be available until after the second blood test.

If you did not have your blood drawn in the first trimester, you may have second trimester screening (quad marker).

The California Prenatal Screening test detects:

- About 80 to 90 percent of Down syndrome
- About 67 to 81 percent of trisomy 18
 - The detection rates for Down syndrome and trisomy 18 depend on your age and which test(s) you have.
- About 80 percent of spina bifida and about 85 percent of abdominal wall defects
- About 60 percent of Smith-Lemli-Opitz syndrome (a rare inherited form of intellectual disability and birth defects)

Non-Invasive Prenatal Testing (10 to 24 weeks)

NIPT is a newer prenatal screening test offered to women who are at higher risk for having a baby with a chromosome problem. It is usually discussed for these situations:

- You will be 35 or older at delivery
- Some types of positive California Prenatal Screening test results
- Some types of ultrasound abnormalities
- Some types of chromosome problems in a prior pregnancy or in one of the parents

This blood test can be done after 10 weeks of pregnancy. It checks the mother's blood for extra DNA pieces from the developing baby. Extra DNA is a sign that the pregnancy has a much higher chance for certain birth defects. NIPT finds almost all pregnancies with Down syndrome and trisomy 18, and most pregnancies with trisomy 13. If you have NIPT, you will be offered quad marker screening for spina bifida and abdominal wall defects.



What to Consider When Making a Decision

Is a Screening Test Right for You?

- Would you like more information than just your age-related risk of having a baby with a genetic birth defect like Down syndrome?
- Are you comfortable knowing that screening tests cannot detect all birth defects and will miss 10 to 20 percent of babies with certain genetic birth defects?
- Do you understand that if you have a positive screening test, the only way to know for sure if your baby has certain birth defects is to have a diagnostic test like CVS or amniocentesis?
- Although most women with positive screening test results have normal babies, a positive result may still cause you anxiety.

Is a Diagnostic Test Right for You?

- Do you need reassurance that your baby does not have specific genetic birth defects?
- Are you willing to accept a small risk of miscarriage to get this information?

- Would knowing that your baby has certain genetic birth defects help you prepare for a child with special needs?
- Would you consider ending the pregnancy if the baby has a birth defect?

Is Not Having Any of These Tests Right for You?

- Would you rather wait until the baby is born to find out if he or she has a chromosome defect or spina bifida?
- It can be helpful for you and your physician to know if your baby has a birth defect before you deliver. Your physician may need to change your prenatal care plan or even change where or how your baby will be delivered.

Conclusion

There is no test that can show all possible birth defects before a baby is born. Prenatal screening or prenatal diagnosis can be useful or reassuring for most couples. However, it can be an emotional experience and may cause anxiety in some people.

The decision to have prenatal screening or prenatal diagnostic tests requires careful thought along with a discussion with your physician or a genetic counselor. All the choices for prenatal testing are optional. Some people choose to decline all testing.

If you have other questions about prenatal screening, amniocentesis, or CVS, contact your prenatal care team or a genetic counselor in your area.

Summary of Prenatal Testing Choices

Diagnostic Tests

- Generally offered to all women at increased risk for birth defects and/or women who are 35 or older. Also available if you do not have an increased risk but want information.
- There is a chance of pregnancy complications, including miscarriage, with these procedures.

TEST	DESCRIPTION
Chorionic Villus Sampling (10 to 13 weeks of pregnancy)	<ul style="list-style-type: none">• Finds more than 99 percent of chromosome abnormalities, including Down syndrome• Does not find neural tube defects, so a second trimester blood test is recommended at 15 to 20 weeks• Chance of complications is similar to amniocentesis, though recent data is limited
Amniocentesis (15 to 20 weeks of pregnancy)	<ul style="list-style-type: none">• Finds more than 99 percent of chromosome abnormalities, including Down syndrome• Finds most neural tube and abdominal wall defects• Chance of serious complications is between 1 in 300 and 1 in 500

Screening Tests

- Do not pose a risk for pregnancy complications.
- Will miss some cases of Down syndrome and other chromosome and birth defects.
- If a test shows an increased risk of birth defects, you will be offered follow-up with a genetic counselor to discuss more testing options.

TEST	DESCRIPTION
<p>California Prenatal Screening Test (10 to 13 weeks 6 days of pregnancy and 15 to 20 weeks)*</p>	<ul style="list-style-type: none"> • Available to all pregnant women • Estimates your risk of having a baby with Down syndrome, trisomy 18, neural tube and abdominal wall defects, and other birth defects • Blood test(s) and sometimes a first trimester ultrasound called nuchal translucency (NT) • If the result is positive, CVS, amniocentesis, or NIPT will be offered
<p>Non-Invasive Prenatal Testing (10 to 24 weeks of pregnancy)</p>	<ul style="list-style-type: none"> • A blood test generally offered to women at higher risk of having a baby with chromosome abnormalities • Shows whether you are at high risk of having a baby with certain chromosome abnormalities, such as Down syndrome, trisomy 18, and trisomy 13

*Refer to the California Prenatal Screening Program booklet for more information.



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