The American College of Obstetricians and Gynecologists

FAQ

FREQUENTLY ASKED QUESTIONS
FAQ165
PREGNANCY

Screening Tests for Birth Defects

What is a birth defect?

A birth defect is a problem that is present at birth, although it may not be noticed until the child is older. Birth defects may affect any part of the body, including major organs such as the heart, lungs, or brain. The defect may affect the baby's appearance, a body function, or both.

What causes birth defects?

Some birth defects are caused by problems with ***chromosomes***. Others are caused by a ***gene*** that is passed from parent to child. Some birth defects result from exposure to harmful agents.

What are some examples of chromosome disorders?

Aneuploidy is a condition in which there are missing or extra chromosomes. The most common aneuploidy is called a ***trisomy***, in which there is an extra chromosome. A common trisomy is ***trisomy 21 (Down syndrome)***. Other trisomies include ***trisomy 13 (Patau syndrome)*** and ***trisomy 18 (Edwards syndrome)***.

A ***monosomy*** is a condition in which there is a missing chromosome. A common monosomy is ***Turner syndrome***, in which a female has a missing or damaged X chromosome.

What are inherited disorders?

Inherited disorders are caused by defective genes. These disorders are passed down by parents to their children. Some inherited disorders are more common in certain races and ethnic groups, such as ***sickle cell disease*** (African American), ***cystic fibrosis*** (non-Hispanic white), and ***Tay–Sachs disease*** (Ashkenazi Jewish, Cajun, and French Canadian).

What other things can cause birth defects?

Birth defects also may be caused by exposure to harmful agents, such as medications, chemicals, and infections. Some birth defects may be caused by a combination of factors. For most birth defects, the cause is not known.

How can I find out if I am at increased risk of passing on a genetic disorder?

Your health care provider or a ***genetic counselor*** can help find out if you are at increased risk of passing on a genetic disorder by asking about your personal and family health history.

What factors may increase my risk of passing on a genetic disorder?

Most babies with birth defects are born to couples without risk factors. However, the risk of birth defects is higher when certain factors are present. You are at increased risk if

- you have a genetic disorder
- you already have a child who has a genetic disorder
- there is a family history of a genetic disorder
- you belong to an ethnic group that has a high risk of certain genetic disorders

What types of prenatal tests are available to address concerns about birth defects?

The following prenatal tests are available:

- ***Carrier*** tests—These screening tests can show if a person carries a gene for an inherited disorder. Carrier tests can be done before or during pregnancy. Cystic fibrosis carrier screening is offered to all women of reproductive age because it is one of the most common genetic disorders.
- ***Screening tests***—These tests assess the risk that a baby will have Down syndrome and other chromosome problems, as well as ***neural tube defects***. These tests do not tell whether the ***fetus*** actually has these disorders.
- ***Diagnostic tests***—These tests can provide information about whether the fetus has a genetic condition and are done on ***cells*** obtained through ***amniocentesis***, ***chorionic villus sampling***, or, rarely, fetal blood sampling. The cells can be analyzed using different techniques.

What are the different types of screening tests for birth defects that can be performed during pregnancy?

Screening tests are performed during different ***trimesters*** of pregnancy. The following table lists the different types of screening tests:

Table 1. Prenatal Screening Tests

Screening Test	*Test Type*	*What Does It Screen for?*	*Down Syndrome Detection Rate*
Combined first-trimester screening	Blood test for PAPP-A and hCG, plus an ***ultrasound exam***	Down syndrome Trisomy 13 Trisomy 18	82–87%
Second-trimester single screen for neural tube defects	Blood test for AFP	Neural tube defects	85%
Second-trimester triple screen	Blood test for AFP, hCG, and ***estriol***	Down syndrome Trisomy 18 Neural tube defects	69%
Second-trimester quad screen	Blood test for AFP, hCG, estriol, and ***inhibin-A***	Down syndrome Trisomy 18 Neural tube defects	81%
Integrated screening	Blood test for PAPP-A and an ultrasound exam in the first trimester, followed by quad screen in the second trimester	Down syndrome Trisomy 18 Neural tube defects	94–96%
Integrated screening (blood test only)	Same as integrated screening but no ultrasound exam	Down syndrome Trisomy 18 Neural tube defects	85–88%

continued

Table 1. Prenatal Screening Tests *(continued)*

Screening Test	*Test Type*	*What Does It Screen for?*	*Down Syndrome Detection Rate*
Contingent sequential	First-trimester combined screening result: —Positive: diagnostic test offered —Negative: no further testing —Intermediate: second-trimester screening test offered	Down syndrome Trisomy 18 Neural tube defects	88–94%
Stepwise sequential	First-trimester combined screening result: —Positive: diagnostic test offered —Negative: second-trimester screening test offered	Down syndrome Trisomy 18 Neural tube defects	95%

Abbreviations: ***AFP, alpha-fetoprotein; hCG, human chorionic gonadotropin; PAPP-A, pregnancy-associated plasma protein A***

Do I have a choice between having screening tests or having diagnostic tests?

If a screening test shows an increased risk of a birth defect, diagnostic tests may be done to determine if a specific birth defect is present. Diagnostic testing may be done instead of screening if a couple is at increased risk of certain birth defects. Diagnostic testing also is offered as a first choice to all pregnant women, even those who do not have risk factors. Your health care provider will discuss all of the testing options with you and recommend the tests that best fit your needs.

What are the advantages and disadvantages of diagnostic tests compared with screening tests?

The main benefit of having diagnostic testing instead of screening is that it tells you whether or not the baby will be born with a chromosome disorder or a specific inherited disorder. The main disadvantage is that diagnostic tests can pose some risks to the pregnancy.

Do I have to have these tests?

Although screening tests for birth defects are offered to all pregnant women, it is your choice whether to have them done. Knowing whether your baby is at risk of or has a birth defect beforehand allows you to prepare for having a child with a particular disorder and to organize the medical care that your child may need. You also may have the option of not continuing the pregnancy.

Glossary

Alpha-fetoprotein (AFP): A protein produced by a growing fetus; it is present in amniotic fluid and, in smaller amounts, in the mother's blood.

Amniocentesis: A procedure in which a needle is used to withdraw and test a small amount of amniotic fluid and cells from the sac surrounding the fetus.

Aneuploidy: Having an abnormal number of chromosomes.

Carrier: A person who shows no signs of a particular disorder but could pass the gene on to his or her children.

Cells: The smallest units of a structure in the body; the building blocks for all parts of the body.

Chorionic Villus Sampling: A procedure in which a small sample of cells is taken from the placenta and tested.

Chromosomes: Structures that are located inside each cell in the body and contain the genes that determine a person's physical makeup.

Cystic Fibrosis: An inherited disorder that causes problems in digestion and breathing.

Diagnostic Tests: Tests that look for a disease or cause of a disease in people who are believed to have or who have an increased risk of a disease.

Estriol: A substance made by the placenta and the liver of the fetus.

Fetus: The developing organism in the uterus from the ninth week of pregnancy until the end of pregnancy.

Gene: A segment of DNA that contains instructions for the development of a physical trait or control of a process in the body. Genes are the basic units of heredity and can be passed down from parent to offspring.

Genetic Counselor: A health care professional with special training in genetics and counseling who can provide expert advice about genetic disorders and prenatal testing.

Human Chorionic Gonadotropin (hCG): A hormone produced during pregnancy; its detection is the basis for most pregnancy tests.

Inhibin-A: A substance made by the placenta during pregnancy.

Monosomy: A condition in which there is a missing chromosome.

Neural Tube Defects: Birth defects that result from incomplete development of the brain, spinal cord, or their coverings.

Pregnancy-Associated Plasma Protein-A (PAPP-A): A protein made by the fetus and placenta during pregnancy.

Screening Tests: Tests that look for possible signs of disease in people who do not have symptoms.

Sickle Cell Disease: An inherited disorder in which red blood cells have a crescent shape, causing chronic anemia and episodes of pain. It occurs most often in African Americans.

Tay-Sachs Disease: An inherited birth defect that causes mental retardation, blindness, seizures, and death, usually by age 5 years. It occurs mostly in people of Eastern European Jewish (Ashkenazi Jews), Cajun, and French Canadian descent.

Trimesters: The three 3-month periods into which pregnancy is divided.

Trisomy: A condition in which there is an extra chromosome.

Trisomy 13 (Patau Syndrome): A genetic disorder that causes serious heart defects and other problems with development. Most infants with trisomy 13 die within the first year of life.

Trisomy 18 (Edwards Syndrome): A genetic disorder that causes serious mental and developmental problems. Most infants with trisomy 18 die within the first year of life.

Trisomy 21 (Down Syndrome): A genetic disorder in which abnormal features of the face and body, medical problems such as heart defects, and intellectual disability occur.

Turner Syndrome: A condition affecting females in which there is a missing or damaged X chromosome. It causes a webbed neck, short height, and heart problems.

Ultrasound Exam: A test in which sound waves are used to examine internal structures. During pregnancy, it can be used to examine the fetus.

If you have further questions, contact your obstetrician–gynecologist.

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Cystic Fibrosis: Prenatal Screening and Diagnosis

What is cystic fibrosis (CF)?

Cystic fibrosis (CF) is a genetic disorder. It is caused by an abnormal ***gene*** that is passed from parent to child. It is a lifelong illness that affects all of the organs of the body and often causes problems with digestion and breathing. It does not affect a person's looks or mental ability. In some cases, CF poses a serious risk to a person's health and shortens the life span. Despite their physical problems, many people with CF attend school, have careers, and lead full lives.

What causes CF?

Cystic fibrosis is a ***recessive disorder***. In a recessive disorder, both parents must carry a copy of the abnormal gene for the problem to occur in their child. A person who has one copy of an abnormal gene for a recessive disorder is a ***carrier*** for that disorder, even though he or she may show no signs of it. If both parents are carriers, each of their children has a 25% chance of having the disorder. Put another way, this couple has a 1-in-4 chance of having a child with CF.

What are the symptoms of CF?

The symptoms of CF can vary in type and severity. Many people with CF produce a thick, sticky mucus in their bodies. This mucus builds up and clogs the lungs, which makes it hard to breathe, and can lead to infection. It also can affect the digestive organs, making it hard for the body to break down and absorb food. Most males with CF are sterile and cannot father children.

Is treatment available for CF?

New drugs and treatments have improved the outlook for people with CF, but it is still a lifelong disease. To treat lung problems, most children with CF need to have physical therapy for about a half hour every day. This therapy helps clear mucus from the lungs. It is easy to do and can be done by parents or other family members.

What are risk factors for CF?

The risk of being a CF carrier is higher in certain races and ethnic groups. It occurs more often in white people than in other racial groups. The risk also is increased in families with a history of CF.

Can I be tested to assess whether I am a CF carrier?

Carrier testing can be done for couples planning a pregnancy or during pregnancy to assess their risk. The test is done on a blood sample. Carrier testing also is available to all pregnant women. If testing shows that a couple is at high risk, more testing can be done during pregnancy to see whether their ***fetus*** has CF.

What does it mean if test results for one partner are negative?

If your test results are negative, the chance that you are a CF carrier is small. There are some rare CF gene defects that the test does not detect. For this reason, you could be told your test result is negative, and you could still be a carrier. The likelihood of this is very small.

What does it mean if test results for one partner are positive?

If the test results show that one partner is a carrier, the next step is to test the other partner. Both parents must be CF carriers for the baby to have CF.

If one parent has a negative test result, the chance that the baby will have CF is small. Because the risk is small, if one partner is a carrier but the other has a negative result, no further testing is recommended.

If the father is not available for a carrier test, a genetic counselor may be able to help you decide whether to have prenatal testing of the fetus to see if it has CF.

What does it mean if test results for both partners are positive?

If two people who are both CF carriers have a baby, there is a 25% chance that the baby will have CF. However, it is more likely that the baby will be a carrier, like the parents, and will have the gene but will not have the disease. It also is possible that although the parents are both carriers, the baby will not be a CF carrier.

If both partners are positive, what follow-up tests are appropriate and what do they assess?

If both partners are CF carriers, further prenatal testing can be done to see if the baby has CF. This testing is not recommended when only one partner is a carrier. Parents may want to know if the baby will have CF so that they can prepare for the care of a child with special health care needs, or they may choose to end the pregnancy.

What prenatal tests can be done to detect CF and other disorders?

Prenatal tests done to detect CF and other disorders are ***chorionic villus sampling (CVS)*** and ***amniocentesis*** (see the FAQ Diagnosing Birth Defects). CVS can be performed after 9 completed weeks of pregnancy. Amniocentesis can be performed between 15 weeks and 20 weeks of pregnancy.

What are my options if diagnostic test results show that the fetus has CF?

Two options are available:

1. Continue the pregnancy and prepare for a child with CF. Couples can use this time to learn as much as possible about the disease, current treatment options, and the experiences of other families who have a child with CF.
2. End the pregnancy. Each state has its own laws on pregnancy termination. You should discuss this decision with your health care provider. You also may want to talk with your partner, counselors, and close friends.

What should partners who are CF carriers be aware of when thinking about future pregnancies?

If a test result shows that you are a CF carrier, the result is definite and will not change. If both partners are carriers, it means that in each pregnancy the baby will have a 25% (1-in-4) chance of having CF. If you want to know whether your baby will have CF, you will need to have amniocentesis or CVS in each pregnancy. Other options include the following:

- Adoption
- Using donor sperm or donor eggs (but the donor should be tested for CF carrier status)
- Using in vitro fertilization with your own sperm and eggs, and then using ***preimplantation genetic diagnosis*** to see if the fertilized egg has CF or is a CF carrier.

Glossary

Amniocentesis: A procedure in which a needle is used to withdraw and test a small amount of amniotic fluid and cells from the sac surrounding the fetus.

Carrier: A person who shows no signs of a particular disorder but could pass the gene on to his or her children.

Chorionic Villus Sampling (CVS): A procedure in which a small sample of cells is taken from the placenta and tested.

Fetus: The developing offspring in the uterus from the ninth week of pregnancy until the end of pregnancy.

Gene: A DNA "blueprint" that codes for specific traits, such as hair and eye color.