ACOG
The American College of Obstetricians and Gynecologists

FAQs

Prenatal Genetic Screening Tests

Frequently Asked Questions

What is prenatal genetic testing?

Prenatal genetic testing gives parents-to-be information about whether their fetus has certain genetic disorders.

What are genetic disorders?

Genetic disorders are caused by changes in a person's genes or chromosomes. Aneuploidy is a condition in which there are missing or extra chromosomes. In a trisomy, there is an extra chromosome. In a monosomy, a chromosome is missing. Inherited disorders are caused by changes in genes called mutations. Inherited disorders include sickle cell disease, cystic fibrosis, Tay-Sachs disease, and many others. In most cases, both parents must carry the same gene to have an affected child.

What are the two main types of prenatal genetic tests?

There are two types of prenatal tests for genetic disorders:

1. Prenatal screening tests: These tests can tell you the chances that your fetus has an aneuploidy and a few other disorders. This FAQ focuses on these tests.
2. Prenatal diagnostic tests: These tests can tell you whether your fetus actually has certain disorders. These tests are done on cells from the fetus or placenta obtained through amniocentesis or chorionic villis sampling (CVS). FAQ164 Prenatal Genetic Diagnostic Tests focuses on these tests.

Both screening and diagnostic testing are offered to all pregnant women.

What are the different types of prenatal genetic screening tests?

Screening tests can tell you your risk of having a baby with certain disorders. They include carrier screening and prenatal genetic screening tests:

- Carrier screening is done on parents (or those just thinking about becoming parents) using a blood sample or tissue sample swabbed from inside the cheek. These tests are used to find out whether a person carries a gene for certain inherited disorders. Carrier screening can be done before or during pregnancy.
- Prenatal genetic screening tests of the pregnant woman's blood and findings from ultrasound exams can screen the fetus for aneuploidy; defects of the brain and spine called neural tube defects (NTDs); and some defects of the abdomen, heart, and facial features. This FAQ focuses on these tests. They include first-trimester screening, second-trimester screening, combined first- and second-trimester screening, and cell-free DNA testing.

What is first-trimester screening?

First-trimester screening includes a test of the pregnant woman's blood and an ultrasound exam. Both tests usually are done together between 10 weeks and 13 weeks of pregnancy:

- The blood test measures the level of two substances.
- The ultrasound exam, called a nuchal translucency screening, measures the thickness of a space at the back of the fetus's neck. An abnormal measurement means there is an increased risk that the fetus has Down syndrome (trisomy 21) or another type of aneuploidy. It also is linked to physical defects of the heart, abdominal wall, and skeleton.

What is second-trimester screening?

Second-trimester screening includes the following tests:

- The “quad” or “quadruple” blood test measures the levels of four different substances in your blood. The quad test screens for Down syndrome, Edwards syndrome (trisomy 18) , and NTDs. It is done between 15 weeks and 22 weeks of pregnancy.
- An ultrasound exam done between 18 weeks and 22 weeks of pregnancy checks for major physical defects in the brain and spine, facial features, abdomen, heart, and limbs.

What is combined first- and second-trimester screening?

The results from first-and second-trimester tests can be combined in various ways. Combined test results are more accurate than a single test result. If you choose combined screening, keep in mind that final results often are not available until the second trimester.

What is cell-free DNA testing?

Cell-free DNA is the small amount of DNA that is released from the placenta into a pregnant woman’s bloodstream. The cell-free DNA in a sample of a woman’s blood can be screened for Down syndrome, Patau syndrome (trisomy 13) , Edwards syndrome, and problems with the number of sex chromosomes . This test can be done starting at 10 weeks of pregnancy. It takes about 1 week to get the results. A positive cell-free DNA test result should be followed by a diagnostic test with amniocentesis or CVS.

What do the different results of prenatal screening tests mean?

Results of blood screening tests for aneuploidy are reported as the level of risk that the disorder might be present:

- A positive screening test result for aneuploidy means that your fetus is at higher risk of having the disorder compared with the general population. It does not mean that your fetus definitely has the disorder.
- A negative result means that your fetus is at lower risk of having the disorder compared with the general population. It does not rule out the possibility that your fetus has the disorder.

Diagnostic testing with CVS or amniocentesis that gives a more definite result is an option for all pregnant women. Your obstetrician or other health care professional, such as a genetic counselor, will discuss what your screening test results mean and help you decide the next steps.

How accurate are prenatal genetic screening tests?

With any type of testing, there is a possibility of false-positive results and false-negative results. A screening test result that shows there is a problem when one does not exist is called a false-positive result. A screening test result that shows there is not a problem when one does exist is called a false-negative result. Your health care professional can give you information about the rates of false-positive and false-negative results for each test.

What should I consider when deciding whether to have prenatal genetic testing?

It is your choice whether to have prenatal testing. Your personal beliefs and values are important factors in the decision about prenatal testing.

It can be helpful to think about how you would use the results of prenatal screening tests in your pregnancy care. Remember that a positive screening test tells you only that you are at higher risk of having a baby with Down syndrome or another aneuploidy. A diagnostic test should be done if you want to know a more certain result. Some parents want to know beforehand that their baby will be born with a genetic disorder. This knowledge gives parents time to learn about the disorder and plan for the medical care that the child may need. Some parents may decide to end the pregnancy in certain situations.

Other parents do not want to know this information before the child is born. In this case, you may decide not to have follow-up diagnostic testing if a screening test result is positive. Or you may decide not to have any testing at all. There is no right or wrong answer.

Glossary

Amniocentesis*:* A procedure in which amniotic fluid and cells are taken from the uterus for testing. The procedure uses a needle to withdraw fluid and cells from the sac that

holds the fetus.

Aneuploidy: Having an abnormal number of chromosomes. Types include trisomy, in which there is an extra chromosome, or monosomy, in which a chromosome is missing. Aneuploidy can affect any chromosome, including the sex chromosomes. Down syndrome (trisomy 21) is a common aneuploidy. Others are Patau syndrome (trisomy 13) and Edwards syndrome (trisomy 18).

Carrier Screening: A test done on a person without signs or symptoms to find out whether he or she carries a gene for a genetic disorder.

Cell-Free DNA: DNA from the placenta that moves freely in a pregnant woman's blood. Analysis of this DNA can be done as a noninvasive prenatal screening test.

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

Chorionic Villus Sampling (CVS): 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. They contain the genes that determine a person's physical makeup.

Cystic Fibrosis (CF): An inherited disorder that causes problems with breathing and digestion.

Diagnostic Tests: Tests that look for a disease or cause of a disease.

DNA: The genetic material that is passed down from parent to child. DNA is packaged in structures called chromosomes.

Down Syndrome (Trisomy 21): A genetic disorder that causes abnormal features of the face and body, medical problems such as heart defects, and mental disability. Most cases of Down syndrome are caused by an extra chromosome 21 (trisomy 21).

Edwards Syndrome (Trisomy 18): A genetic condition that causes serious problems. It causes a small head, heart defects, and deafness.

Fetus: The stage of human development beyond 8 completed weeks after fertilization.

Genes: Segments of DNA that contain instructions for the development of a person's physical traits and control of the processes in the body. The gene is the basic unit of heredity and can be passed from parent to child.

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

Genetic Disorders: Disorders caused by a change in genes or chromosomes.

Inherited Disorders: Disorders caused by a change in a gene that can be passed from parents to children.

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

Mutations: Changes in genes that can be passed from parent to child.

Neural Tube Defects (NTDs): Birth defects that result from a problem in development of the brain, spinal cord, or their coverings.

Nuchal Translucency Screening: A test to screen for certain birth defects, such as Down syndrome, Edwards syndrome, or heart defects. The screening uses ultrasound to measure fluid at the back of the fetus's neck.

Obstetrician: A doctor who cares for women during pregnancy and their labor.

Patau Syndrome (Trisomy 13): A genetic condition that causes serious problems. It involves the heart and brain, cleft lip and palate, and extra fingers and toes.

Placenta: An organ that provides nutrients to and takes waste away from the fetus.

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

Sex Chromosomes: The chromosomes that determine a person's sex. In humans, there are two sex chromosomes, X and Y. Females have two X chromosomes and males have an X and a Y chromosome.

Sickle Cell Disease: An inherited disorder in which red blood cells have a crescent shape, which causes chronic anemia and episodes of pain.

Tay–Sachs Disease: An inherited disorder that causes mental disability, blindness, seizures, and death, usually by age 5.

Trimester: A 3-month time in pregnancy. It can be first, second, or third.

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

Ultrasound Exams: Tests in which sound waves are used to examine inner parts of the body. During pregnancy, ultrasound can be used to check the fetus.

If you have further questions, contact your ob-gyn.

Don't have an ob-gyn? Learn how to find a doctor near you.

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