ACOG
The American College of Obstetricians and Gynecologists

FAQs

Prenatal Genetic Diagnostic Tests

Frequently Asked Questions

Overview

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.

Inherited disorders (disorders passed from parent to child) are caused by changes in genes. These changes are 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.

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.

What are the two main types of prenatal genetic tests?

There are two general 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 additional disorders. Read Prenatal Genetic Screening Tests to learn about these tests.

2. Prenatal diagnostic tests : These tests can tell you, with as much certainty as possible, whether your fetus actually has an aneuploidy or specific inherited disorders for which you request testing. These tests are done on cells from the fetus or placenta obtained through amniocentesis or chorionic villus sampling (CVS) . This FAQ focuses on these tests.

Both screening and diagnostic testing are offered to everyone who is pregnant.

Types of Diagnostic Tests

What is amniocentesis?

Amniocentesis is a diagnostic test. It is usually done between 15 and 20 weeks of pregnancy, but it can also be done up until you give birth.

To perform the test, a very thin needle is used to withdraw a small amount of amniotic fluid . Ultrasound is used to guide the procedure. Depending on the way the cells are analyzed and the information that you want, results can take from 2 days to several weeks.

There is a very small chance of pregnancy loss with amniocentesis. Leakage of amniotic fluid and slight bleeding can occur after amniocentesis. In most cases, both stop on their own.

What is chorionic villus sampling (CVS)?

In CVS, a sample of tissue is taken from the placenta. The main advantage of having CVS over amniocentesis is that CVS is done earlier than amniocentesis, between 10 and 13 weeks of pregnancy. The chance of miscarriage with CVS is slightly higher than the chance of miscarriage with amniocentesis.

What is preimplantation genetic diagnosis?

Preimplantation genetic diagnosis may be offered to couples who are using in vitro fertilization (IVF) to get pregnant and who are at increased risk of having a baby with a genetic or chromosomal disorder. Before an embryo is transferred to the uterus , it is

tested for certain genetic disorders and mutations. Only embryos that do not test positive for the disorders are transferred.

How are the cells analyzed in prenatal diagnostic testing?

A number of technologies are used in prenatal diagnostic testing. Your obstetrician–gynecologist (ob-gyn) or genetic counselor can help select the tests that are best for your situation:

- Karyotype —Missing, extra, or damaged chromosomes can be detected by taking a picture of the chromosomes and arranging them in order from largest to smallest. Karyotyping results are ready in 1 to 2 weeks after the cells are sampled.
- Fluorescence in situ hybridization (FISH) —This technique can be used to detect common aneuploidies involving chromosomes 13, 18, and 21 and the X and Y chromosomes. Results are ready more quickly than with traditional karyotyping (usually within 2 days). Positive test results are confirmed with a karyotype.
- Chromosome microarray analysis—This test can look for different kinds of chromosome problems, including aneuploidy, throughout the entire set of chromosomes. It can find some chromosome problems that karyotyping can miss. Results can be ready in about 7 days.
- DNA testing—Tests for specific gene mutations can be done by request. For example, if you and your partner are carriers of the cystic fibrosis gene, you may want to request prenatal diagnostic testing for this specific mutation.

Understanding Test Results

What does a negative test result mean?

Most of the time, the results of a diagnostic test are negative (normal). A negative result does not rule out the possibility that the fetus has a genetic disorder. It only tells you that the fetus does not have the particular disorder that was tested for.

What if the test result is positive?

If a diagnostic test result is positive (it shows that the fetus has the disorder tested for), your ob-gyn or genetic counselor can explain the results and provide guidance about

your choices and options.

A specialist in the disorder can help you understand the life expectancy of the disorder, whether treatment is available, and the care that your child may need. Support groups, counselors, and social workers can also listen to your concerns and answer questions.

It may be possible to have more tests, such as a specialized ultrasound exam , to find out more detail about the defect. But in some cases, it is not possible to predict whether the defect will be mild or severe.

Thinking About Testing Options

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

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

It can be helpful to think about what you would do if a diagnostic test result comes back positive. Some parents want to know beforehand if their child will be born with a genetic disorder. This gives parents time to learn about the disorder and plan for medical care that the child may need.

If the disorder is very serious and the life expectancy is short, hospice care for the baby can be planned. 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. They may decide not to have any testing at all. There is no right or wrong answer.

Keep in mind that certain tests can be done only at certain times during pregnancy. Tests that are done earlier allow parents more time to make decisions if a test result is positive. If ending the pregnancy is being considered, it is safer to do so within the first 13 weeks of pregnancy.

[3 Questions to Ask Yourself Before Getting Prenatal Genetic Testing]

How do I choose between prenatal screening and diagnostic testing?

You can choose to have diagnostic tests instead of or in addition to screening tests. A genetic counselor or other health care professional with expertise in genetics can study your family health history, recommend specific tests, and interpret test results.

The main benefit of having diagnostic tests instead of screening is that it can detect all conditions caused by an extra chromosome and many other disorders in which chromosomes are missing or damaged. Diagnostic tests are also available for many inherited disorders.

The main disadvantage is that diagnostic testing carries a very small risk of losing the pregnancy.

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.

Amniotic Fluid: Fluid in the sac that holds the fetus.

Aneuploidy: Having an abnormal number of chromosomes.

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: 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.

Embryo: The stage of development that starts at fertilization (joining of an egg and sperm) and lasts up to 8 weeks.

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

Fluorescence In Situ Hybridization (FISH): A screening test for common chromosome problems. The test is done using a tissue sample from an amniocentesis or chorionic villus test.

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.

Hospice Care: Care that focuses on comfort for people who have an illness that will lead to death.

In Vitro Fertilization (IVF): A procedure in which an egg is removed from a woman's ovary, fertilized in a laboratory with the man's sperm, and then transferred to the woman's uterus to achieve a pregnancy.

Karyotype: An image of a person's chromosomes, arranged in order of size.

Microarray: A technology that examines all of a person's genes to look for certain genetic disorders or abnormalities. Microarray technology can find very small genetic changes that can be missed by the routine genetic tests.

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

Mutations: Changes in a gene that can be passed on from parent to child.

Obstetrician–Gynecologist (Ob-Gyn): A doctor with special training and education in women's health.

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

Preimplantation Genetic Diagnosis: A type of genetic testing that can be done during in vitro fertilization. Tests are done on the fertilized egg before it is transferred to the uterus.

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

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 intellectual disability, blindness, seizures, and death, usually by age 5.

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

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

Uterus: A muscular organ in the female pelvis. During pregnancy, this organ holds and nourishes the fetus. Also called the womb.

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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