ACOG
The American College of Obstetricians and Gynecologists

FAQs

Genetic Disorders and Pregnancy

Frequently Asked Questions

Overview

What are genetic disorders?

Genetic disorders are disorders caused by changes in genes or chromosomes. Genetic disorders may be a physical problem or intellectual disability that is present at birth. They are a type of birth defect. Some genetic disorders may not be noticed until the child is older.

Genetic counseling and screening tests can help assess your risk of having a baby with certain genetic disorders. Diagnostic tests can detect if a specific genetic disorder is present in the fetus.

What are genes?

A gene is a small piece of hereditary material called DNA. Each gene controls some aspect of how your body looks or functions. Genes come in pairs.

What are chromosomes?

Chromosomes are the structures inside cells that carry genes. Chromosomes also come in pairs. Most cells have 23 pairs of chromosomes for a total of 46 chromosomes. Sperm and egg cells each have 23 chromosomes.

During fertilization, when the egg and sperm join, the two sets of chromosomes come together. In this way, one half of a baby's genes come from the baby's mother and one

half come from the baby's father.

Your baby's sex is determined by sex chromosomes. There are two sex chromosomes: X and Y. Egg cells only contain an X chromosome. Sperm cells can carry an X or a Y. A combination of XX results in a girl and XY results in a boy.

Types of Genetic Disorders

What are chromosome disorders?

Chromosome disorders involve having missing or extra chromosomes. This is called aneuploidy . Most children with chromosome disorders have physical defects. Some have intellectual disabilities.

Trisomy is the most common aneuploidy. In trisomy, there is an extra chromosome. A common trisomy is Down syndrome (trisomy 21) . Other trisomies include Patau syndrome (trisomy 13) and Edwards syndrome (trisomy 18) .

Monosomy is another type of aneuploidy in which there is a missing chromosome. A common monosomy is Turner syndrome , in which a female has a missing or damaged X chromosome.

The risk of having a child with chromosome disorder increases as you age.

What are inherited disorders?

An inherited disorder is caused by a faulty gene that can be passed from parent to child. Faulty genes can occur on any of the chromosomes. There are autosomal dominant disorders , autosomal recessive disorders , and sex-linked disorders .

What is an autosomal dominant disorder?

An autosomal dominant disorder is caused by just one faulty gene from either parent. "Autosomal" means that the defective gene is located on any of the chromosomes that are not the sex chromosomes (X or Y).

If one parent has the gene, each child of the couple has a 50 percent chance of inheriting the disorder. An example of an autosomal dominant disorder is Huntington

disease .

What is an autosomal recessive disorder?

Autosomal recessive disorders only happen when both parents carry the gene. An example of an autosomal recessive disorder is cystic fibrosis (CF) .

What does it mean to be a carrier of a recessive disorder?

A carrier of a recessive disorder is a person who carries one copy of a gene that works incorrectly and one that works normally. A carrier may not have symptoms of the disorder or may have only mild symptoms.

If both parents are carriers of an abnormal gene, there is a 25 percent chance that the child will get the abnormal gene from each parent and will have the disorder. There is a 50 percent chance that the child will be a carrier of the disorder—just like the carrier parents. If only one parent is a carrier, there is a 50 percent chance that the child will be a carrier.

What are sex-linked disorders?

Sex-linked disorders are caused by defective genes on the sex chromosomes. An example of a sex-linked disorder is hemophilia . This disease is caused by a faulty gene on the X chromosome.

What are multifactorial disorders?

Multifactorial disorders are caused by a combination of factors. Some factors are genetic and some are not. A few of these disorders can be detected during pregnancy. Sometimes they can be corrected with surgery.

Risk Factors and Testing

Who is at risk of having a baby with a genetic disorder?

Most babies with genetic disorders are born to couples without risk factors. But the risk of genetic disorders is higher when certain factors are present. This is why screening

begins by assessing your risk factors before or during pregnancy. You are at increased risk if

- you have a genetic disorder
- you have a child with a genetic disorder
- there is a family history of a genetic disorder

Some genetic disorders are more common in certain ethnic groups.

What is genetic counseling?

A genetic counselor has special training in genetics. A genetic counselor will study your family health history and may recommend you have physical exams or tests with your obstetrician–gynecologist (ob-gyn) . Using the test results, the counselor will assess your baby's risk of having a problem, discuss your options, and talk about any concerns you may have.

What types of prenatal genetic tests are available?

Screening tests assess the risk that a baby may be born with a specific genetic disorder. Diagnostic tests can detect if a specific genetic disorder is present in the fetus.

[ACOG Explains: Prenatal Genetic Testing]

What screening tests are offered before pregnancy?

Carrier screening is a type of screening test that can show if a person carries a gene for an inherited disorder. Carrier screening can be done before or during pregnancy. Read Carrier Screening to learn more.

Who can have carrier screening?

Anyone who is thinking about pregnancy or already pregnant can have carrier screening. Your partner may be tested too.

Carrier screening can test for many disorders, including cystic fibrosis (CF), spinal muscular atrophy (SMA) , and hemoglobinopathies . You can have tests for specific

disorders based on your risk factors, or you can have a test for many disorders without regard to risk factors.

What screening tests are offered during pregnancy?

Carrier screening can be done any time during pregnancy. Other screening tests that can be done during pregnancy include the following:

- Blood tests that measure the level of certain substances in your blood combined with an ultrasound exam . These tests assess the risk that a baby will have Down syndrome, other trisomies, or neural tube defects (NTDs) .
- Cell-free DNA screening (also called noninvasive prenatal testing or NIPT). Cell-free DNA is the small amount of DNA that is released from the placenta into your bloodstream. The cell-free DNA in a sample of your blood can be screened for Down syndrome, other trisomies, and problems with the number of sex chromosomes.

Read Prenatal Genetic Screening Tests to learn more about these tests.

Are there risks to the fetus with screening tests?

There are no risks to the fetus with any of these screening tests.

When are diagnostic tests offered during pregnancy?

Diagnostic tests may be recommended if a screening test shows an increased risk of a birth defect. Diagnostic tests should also be offered as a first choice to everyone during pregnancy, even if you do not have risk factors. Diagnostic tests can detect if a specific birth defect or genetic disorder is present.

How are diagnostic tests done?

Diagnostic tests are done on cells from the fetus obtained through amniocentesis , chorionic villus sampling (CVS) , or rarely, fetal blood sampling. The chromosomes and genes in the cells are analyzed using different techniques to diagnose certain inherited defects and many chromosomal defects. Read Prenatal Genetic Diagnostic Tests to learn more.

Are there risks to the pregnancy with diagnostic tests?

Yes, diagnostic tests carry a very small risk of pregnancy loss.

Which tests should I have?

Your ob-gyn or a genetic counselor can discuss all of the testing options with you and help you decide.

Do I have to have these tests?

Whether you want to be tested is a personal choice. Some people would rather not know if they are at risk or whether their child will have a disorder. Others want to know in advance. Knowing beforehand gives you time to prepare for having a child with a disorder and to organize the medical care that your child may need. Some parents may decide to end the pregnancy in certain situations.

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

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.

Autosomal Dominant Disorders: Genetic disorders caused by one defective gene. The defective gene is located on one of the chromosomes that is not a sex chromosome.

Autosomal Recessive Disorders: Genetic disorders caused by two defective genes, one inherited from each parent. The defective genes are located on one of the pairs of chromosomes that are not the sex chromosomes.

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

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.

Egg: The female reproductive cell produced in and released from the ovaries. Also called the ovum.

Fertilization: A multistep process that joins the egg and the sperm.

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

Gene: A segment of DNA that contains 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.

Hemoglobinopathies: Any inherited disorder that affects the number or shape of red blood cells in the body. Examples include sickle cell disease and the different forms of thalassemia.

Hemophilia: A disorder caused by a mutation on the X chromosome. Affected people are usually males who lack a substance in the blood that helps clotting. People with hemophilia are at risk of severe bleeding from even minor injuries.

Huntington Disease: A disorder that causes loss of control of body movements and mental function.

Monosomy: A condition in which there is a missing chromosome

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

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

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-Linked Disorders: Genetic disorders caused by a change in a gene located on the sex chromosomes.

Sperm: A cell produced in the male testicles that can fertilize a female egg.

Spinal Muscular Atrophy (SMA): An inherited disorder that causes wasting of the muscles and severe weakness. SMA is the leading genetic cause of death in infants.

Trisomy: A problem where there is an extra chromosome.

Turner Syndrome: A problem that affects women when there is a missing or damaged X chromosome. This syndrome causes a webbed neck, short height, and heart problems.

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.

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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