ACOG
The American College of Obstetricians and Gynecologists

FAQs

# Genetic Disorders

## Frequently Asked Questions

### What are genes?

A gene is a small piece of hereditary material called DNA that controls some aspect of a person's physical makeup or a process in the body. 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.

### What determines my baby's sex?

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.

### What causes genetic disorders?

Genetic disorders may be caused by problems with either chromosomes or genes.

### What causes chromosome disorders?

A chromosome disorder is caused by problems with chromosomes. Most children with chromosome disorders have physical defects and some have intellectual disabilities.

## What is aneuploidy?

Having missing or extra chromosomes is a condition called aneuploidy. The risk of having a child with an aneuploidy increases as a woman ages.

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.

## What is an inherited disorder?

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 is a carrier?

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 of the disorder.

## 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.

## Who is at risk of having a baby with a birth defect?

Most babies with birth defects are born to couples without risk factors. But the risk of birth defects is higher when certain factors are present. This is why screening for birth defects begins by assessing your risk factors, such as whether

- 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 will be born with a specific birth defect or genetic disorder. Diagnostic tests can detect if a specific birth defect or genetic disorder is present in the fetus .

## What screening tests are offered during pregnancy?

Screening tests include blood tests that measure the level of certain substances in the mother's 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) .

There also is cell-free DNA screening. 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, other trisomies, and problems with the number of sex chromosomes.

See 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.

## What is carrier testing?

Carrier testing is a type of screening that can show if a person carries a gene for an inherited disorder.

## Who should have carrier testing?

Carrier testing often is recommended for people with a family history of a genetic disorder or people from certain races or ethnic groups who are at increased risk of having a child with a specific genetic disorder. CF screening and spinal muscular atrophy (SMA) screening are offered to all women of reproductive age because CF and SMA are the most common genetic disorders.

## When is carrier testing done?

Carrier testing can be done before or during pregnancy. See Carrier Screening to learn more about prepregnancy carrier testing.

## 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 testing also is offered as a first choice to all pregnant women, even those who 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

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. See Prenatal Genetic Diagnostic Tests to learn more.

## Are there risks to the pregnancy with diagnostic tests?

Diagnostic tests carry risks, including an increased risk of pregnancy loss.

## How do I know which tests to have?

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

## Do I have to have these tests?

Whether you want to be tested is a personal choice. Some couples 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. You also may have the option of ending 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.

**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.

**Birth Defects:** Physical problems that are present at birth.

**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.

**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.

bleeding from even minor injuries.

**Huntington Disease:** An 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.

**Prenatal Care:** A program of care for a pregnant woman before the birth of her baby.

**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.

FAQ094

Last updated: January 2021

Last reviewed: February 2020



*This information is designed as an educational aid for the public. It offers current information and opinions related to women's health. It is not intended as a statement of the standard of care. It does not explain all of the proper treatments or methods of care. It is not a substitute for the advice of a physician. Read ACOG's complete disclaimer.*