



The American College of
Obstetricians and Gynecologists

FAQ

FREQUENTLY ASKED QUESTIONS

FAQ094

PREGNANCY

Genetic Disorders

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What are genes?

Genes control your physical makeup. Genes come in pairs. Half of a **fetus's** genes come from the mother. The other half comes from the father. Some traits, such as blood type, are determined by a single gene pair. Other traits—including skin color, hair color, and height—are the result of many pairs of genes working together.

What are chromosomes?

Chromosomes are the structures inside cells that contain genes. A man's sperm and a woman's egg have 23 chromosomes each. All other cells in the body have 46. When an egg is fertilized by a sperm, 23 chromosomes from the mother and 23 chromosomes from the father join to form the 46 chromosomes of the cell that will become the fetus.

What are sex chromosomes?

The sex chromosomes are types of chromosomes that determine a person's sex. There are two types of sex chromosomes: X and Y. Each person has one pair of sex chromosomes—one from the father's sperm and one from the mother's egg. A normal sperm has either an X or a Y chromosome. A normal egg always has an X chromosome. The sex chromosome in the sperm determines the sex of the child. If a sperm with a Y chromosome joins with an egg, the fetus is male (XY). If the sperm has an X chromosome, the fetus is female (XX).

What are genetic disorders?

Genetic disorders are diseases or defects caused by problems with either genes or chromosomes. An inherited disorder is caused by a gene that is passed from parent to child. These disorders can be dominant, recessive, or X-linked. Chromosomal disorders are diseases caused by missing, damaged, or extra chromosomes.

What are dominant disorders?

A dominant disorder is an inherited disorder caused by a single gene. The gene can be from either parent. If one parent has the gene, each child of the couple has a 1-in-2 chance of inheriting the disorder.

What are recessive disorders?

A recessive disorder is an inherited disorder caused by two defective genes. Both parents must carry the defective gene before the problem can occur in their child. If both parents are carriers of the same recessive disorder, each of their children has a 1-in-4 chance of having the disorder.

If you have a recessive gene for a certain disorder, you are a carrier for that disorder. Although you may show no signs of the disorder yourself, you can still pass it on to your children.

What are X-linked disorders?

Disorders that are caused by genes on the X chromosome are called X-linked or sex-linked disorders. In most X-linked disorders, the abnormal gene is recessive.

A male baby inherits one X chromosome from his mother. If the X chromosome he inherits has the disorder, he will get the disorder because he does not have another X chromosome with a normal gene to cancel out the abnormal one. Color blindness is a common X-linked trait.

What are chromosomal disorders?

Chromosomal disorders are disorders caused by missing, damaged, or extra chromosomes. These problems often are caused by an error that occurred when the egg or sperm was forming. Most children with chromosomal disorders have physical defects, and some have mental defects.

The risk of having a child with a chromosomal disorder increases as a woman ages. For instance, a 35-year-old woman has a 1 in 192 (less than 1%) chance of having a baby with a chromosomal disorder. The chance increases to 1 in 66 (about 1.5%) in a woman aged 40 years.

What are multifactorial disorders?

Disorders thought to come from a mix of factors are called multifactorial disorders. A few of these disorders can be detected during pregnancy.

What is carrier testing?

A carrier test detects whether a person is a carrier of a certain genetic defect. Carrier testing of both parents can be done before, during, or after pregnancy.

For a carrier test, a sample of blood or saliva is studied in a lab to detect a defective gene for a certain inherited disorder. All women may be offered or informed about carrier testing for cystic fibrosis. Other tests can be done if your family history, ethnic origin, or other factors raises your risk of being a carrier. There are no carrier tests for most inherited birth defects.

If you are a carrier of a defective gene, your health care provider or genetic counselor will help you understand the chances that the defect could be passed on to your baby. If the carrier testing is done before you are pregnant, you can use the results to decide if you want to get pregnant.

What are screening tests?

Screening tests are performed during pregnancy to assess the risk that a baby has certain birth defects, including Down syndrome, trisomy 13, trisomy 18, and neural tube defects. The following screening tests may be offered:

- Maternal serum screening—These tests measure the level of three or four substances in the mother's blood. They are performed during the second trimester of pregnancy and assess whether the baby is at increased risk of Down syndrome, trisomy 18, abdominal wall defect, and neural tube defects.
- First trimester screening—This screening test combines the results of a special **ultrasound** test called **nuchal translucency screening** and blood (serum) tests (PAPP-A and hCG). It assesses whether the baby is at increased risk of Down syndrome, trisomy 18, and some other chromosomal disorders.

What are diagnostic tests?

Diagnostic tests detect whether certain birth defects are present. They usually are performed if a screening test or other factors raise concerns. The following diagnostic tests may be offered:

- Detailed ultrasound exam—This type of ultrasound exam can help explain abnormal screening test results and provide more detailed information.
- Amniocentesis—In this procedure, a needle is used to withdraw a small amount of amniotic fluid and cells from the sac surrounding the fetus. The amniotic fluid and cells can be tested to detect certain chromosomal problems in the fetus.
- Chorionic villus sampling (CVS)—A small sample of cells is taken from the placenta and tested. CVS detects some of the same chromosomal problems that amniocentesis does.
- Fetal blood sampling—Also known as cordocentesis, this procedure tests for chromosomal defects and other problems. For this test, blood is taken from a vein in the umbilical cord. Fetal blood sampling usually is used when the results of amniocentesis, chorionic villus sampling, or ultrasound are unclear.

Glossary

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

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

Genes: DNA "blueprints" that code for specific traits, such as hair and eye color.

Nuchal Translucency Screening: A special ultrasound test of the fetus to screen for the risk of Down syndrome and other birth defects.

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

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

FAQ094: Designed as an aid to patients, this document sets forth current information and opinions related to women's health. The information does not dictate an exclusive course of treatment or procedure to be followed and should not be construed as excluding other acceptable methods of practice. Variations, taking into account the needs of the individual patient, resources, and limitations unique to institution or type of practice, may be appropriate.

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