The American College of
Obstetricians and Gynecologists

FREQUENTLY ASKED QUESTIONS
FAQ094
PREGNANCY

Genetic Disorders

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 ***trisomy 21 (Down syndrome)***. Other trisomies include ***trisomy 13 (Patau syndrome)*** and ***trisomy 18 (Edwards syndrome)***. ***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 defective genes that can be passed down by parents to their children. Defective genes can occur on any of the chromosomes. A genetic disorder can be ***autosomal dominant***, ***autosomal recessive***, or ***sex linked***.

What is an autosomal dominant disorder?

An autosomal dominant disorder is caused by just one defective 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% 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***.

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% chance that the child will get the abnormal gene from each parent and will have the disorder. There is a 50% 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% 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 defective gene on the X chromosome.

What are multifactorial disorders?

Multifactorial disorders are caused by a combination of factors. Some factors are genetic, while some are nongenetic. A few of these disorders can be detected during pregnancy.

Do certain people have an increased risk of having a child with a birth defect compared with others?

Most babies with birth defects are born to couples without risk factors. However, the risk of birth defects is higher when certain factors are present. Screening for birth defects begins by assessing your risk factors, such as whether you have a genetic disorder, whether you have a child with a genetic disorder, or whether there is a family history of a genetic disorder. Some genetic disorders are more common in certain ethnic groups.

What is genetic counseling?

In some situations, you may be referred to a ***genetic counselor***. A genetic counselor has special training in genetics. In addition to studying your family health history, he or she may refer you for physical exams and tests. Using this information, 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 tests are available to address concerns about genetic disorders?

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.

When are screening tests offered during pregnancy and what kinds of disorders do they assess?

Screening tests often are part of routine prenatal care and are done at different times during the first and second ***trimesters*** of 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 and other trisomies, as well as ***neural tube defects***. Prenatal screening tests are discussed in detail in FAQ165 Screening Tests for Birth Defects.

What are carrier tests?

Carrier tests are a type of screening test that can show if a person carries a gene for an inherited disorder.

For whom is carrier testing recommended?

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. Cystic fibrosis carrier screening is offered to all women of reproductive age because it is one of the most common genetic disorders.

When is carrier testing done?

Carrier tests can be done before (preconception) or during pregnancy. Preconception carrier testing is discussed in detail in FAQ179 Preconception Carrier Screening.

When are diagnostic tests offered during pregnancy and what kinds of disorders do they detect?

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***, or, rarely, fetal blood sampling. The chromosomes and genes in the cells then can be analyzed using different techniques to diagnose certain inherited defects and many chromosomal defects. Diagnostic tests are discussed in detail in FAQ164 Diagnostic Tests for Birth Defects.

Are there risks associated with diagnostic tests?

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

How do I know which tests to have?

Your health care provider or a genetic counselor can discuss all of the testing options with you and help you decide based on your individual 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, but others want to know in advance. Knowing beforehand gives you time to prepare for having a child with a particular disorder and to organize the medical care that your child may need. You also may have the option of not continuing the pregnancy.

Glossary

Amniocentesis: A procedure in which a needle is used to withdraw and test a small amount of amniotic fluid and cells from the sac surrounding the fetus.

Aneuploidy: Having an abnormal number of chromosomes.

Autosomal Dominant Disorder: A genetic disorder caused by one defective gene; the defective gene is located on one of the chromosomes that is not a sex chromosome.

Autosomal Recessive Disorder: A genetic disorder 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 particular disorder but could pass the gene on to his or her children.

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

Chorionic Villus Sampling: 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 and contain the genes that determine a person's physical makeup.

Cystic Fibrosis: An inherited disorder that causes problems in digestion and breathing.

Diagnostic Tests: Tests that look for a disease or cause of a disease in people who are believed to have or who have an increased risk of a disease.

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

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

Fertilization: Joining of the egg and sperm.

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

Gene: A segment of DNA that contains instructions for the development of a physical trait or control of a process in the body. They are the basic units of heredity and can be passed down from parent to offspring.

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

Hemophilia: A disorder caused by a defective gene on the X chromosome. Affected individuals lack a substance in the blood that helps it clot and are at risk of severe bleeding from even minor injuries.

Huntington Disease: An autosomal dominant disorder that causes loss of control of body movements and mental function. Symptoms typically start between the ages of 35 years and 50 years.

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

Neural Tube Defects: Birth defects that result from incomplete development of the brain, spinal cord, or their coverings.

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

Sex-Linked Disorder: A genetic disorder caused by a defective gene or genes that are located on the sex chromosomes.

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

Trimesters: The three 3-month periods into which pregnancy is divided.

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

Trisomy 13 (Patau Syndrome): A genetic disorder that causes serious heart defects and other problems with development. Most infants with trisomy 13 die within the first year of life.

Trisomy 18 (Edwards Syndrome): A genetic disorder that causes serious mental and developmental problems. Most infants with trisomy 18 die within the first year of life.

Trisomy 21 (Down Syndrome): A genetic disorder in which abnormal features of the face and body, medical problems such as heart defects, and intellectual disability occur.

Turner Syndrome: A condition affecting females in which there is a missing or damaged X chromosome. It causes a webbed neck, short height, and heart problems.

Ultrasound Exam: A test in which sound waves are used 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 the institution or type of practice, may be appropriate.