PATIENT EDUCATION

The American College of Obstetricians and Gynecologists
WOMEN'S HEALTH CARE PHYSICIANS

Pregnancy • EP094

Genetic Disorders

Almost all children in the United States are born healthy. Out of 100 newborns, about three will have a major birth defect. A birth defect is a physical problem or intellectual disability that is present at birth, although some birth defects may not be noticed until the child is older.

Many birth defects are caused by problems with either ***chromosomes*** *or* ***genes****. Genetic counseling and prenatal* ***screening tests*** *can help assess your risk of having a child with certain genetic disorders.* ***Diagnostic tests*** *that can find a limited number of disorders in the* ***fetus*** *also are available.*

This pamphlet explains

- *types of genetic disorders*
- *who is at risk*
- *available tests*

Genes and Chromosomes

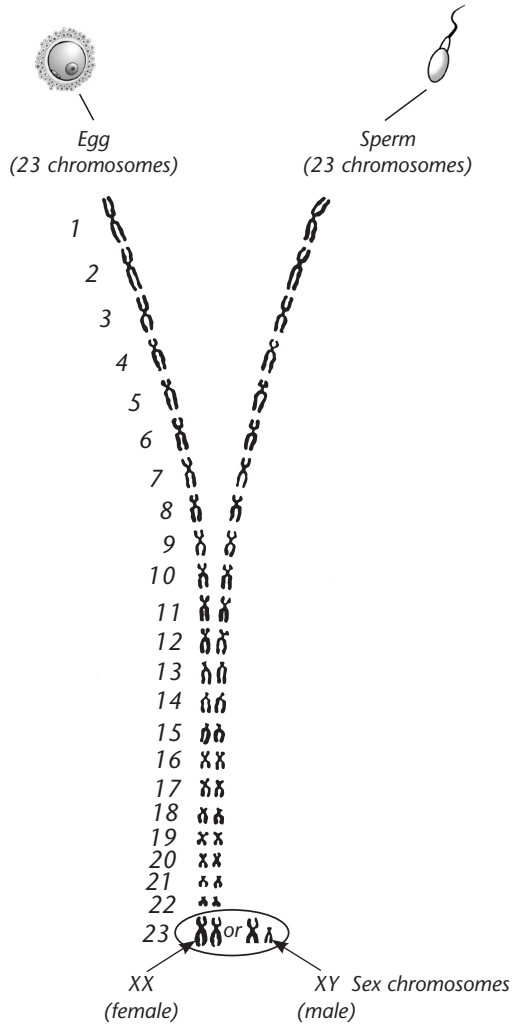
A gene is a small piece of hereditary material called ***DNA*** that controls some aspect of a person's physical makeup or a process in the body. Genes come in pairs and are located on chromosomes. Chromosomes also come in pairs. Most ***cells*** have 23 pairs of chromosomes for a total of 46 chromosomes. ***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 mother and one half come from the father.

A baby's sex depends on the sex chromosomes it gets. 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

Genetic disorders may be caused by problems with either chromosomes or genes. An inherited disorder is caused by a gene that is passed from parent to child. A chromosome disorder is caused by problems with chromosomes. Multifactorial disorders are caused by a combination of factors.

Chromosomes



Every cell in the body (except for eggs and sperm) has 46 chromosomes. One half of a person's genes come from the mother and one half come from the father. Chromosomes carry genes, which also come in pairs. The 23rd pair of chromosomes are the sex chromosomes.

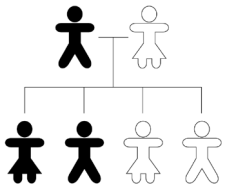
Chromosome Disorders

Having missing or extra chromosomes is a condition called ***aneuploidy***. The risk of having a child with an aneuploidy increases as a woman ages. Most children with chromosome disorders have physical defects and some have intellectual disabilities.

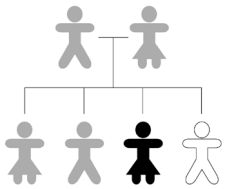
The most common aneuploidy is a ***trisomy***, in which there is an extra chromosome. A common trisomy is ***trisomy 21 (Down syndrome)***. Other trisomies include ***trisomy 13 (Patau syndrome)*** and ***trisomy 18 (Edwards syndrome)***. A ***monosomy*** is a condition in which there is a missing chromosome. A common monosomy is ***Turner syndrome***, in which a female has a missing or damaged X chromosome.

Types of Genetic Disorders

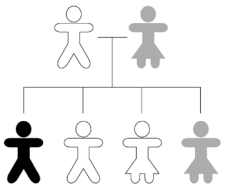
Dominant Disorder. *If one parent has a dominant gene disorder, there is a 50% chance that it will be passed to each child.*



Recessive Disorder. *If both parents carry the recessive gene for a disorder, there is a 25% chance that a child they have will have the disorder, a 50% chance that a child will be a carrier, and a 25% chance that a child will not get the gene at all.*



X-linked Disorder. *If a woman is a carrier of an X-linked disorder and she has a son, there is a 50% chance that he will have the disorder. If she has a daughter, there is a 50% chance the daughter will be a carrier. If the father has an X-linked disorder, all the daughters will be carriers and none of the sons will be affected.*



Affected person *Carrier* *Unaffected person*

Inherited Disorders

Inherited disorders are 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***.

Autosomal Dominant Disorders. Just one defective gene from either parent can cause a dominant gene disorder. The disorder is called "autosomal" when the defective gene is located on any of the 22 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 a dominant disorder is ***Huntington disease***. This nerve disorder causes loss of control of movements and mental function, usually starting at 35–50 years of age.

Autosomal Recessive Disorders. For recessive disorders, both parents must carry the gene for the problem to occur in their child. A ***carrier*** of a recessive disorder has one version of a gene that works incorrectly and one that works normally. Carriers of recessive disorders often are not aware that they have a defective gene. A carrier may not have symptoms of the disorder or may have only mild symptoms.

Some recessive disorders occur more often in certain races and ethnic groups. The following are examples of autosomal recessive disorders:

- ***Sickle cell disease***—In this disorder, red blood cells have a crescent shape that causes them to get stuck in the blood vessels. This cuts off the flow of oxygen to organs, causing episodes of severe pain and organ damage. It occurs most often in African Americans.
- ***Tay–Sachs disease***—This disorder causes blindness, seizures, and death, usually by age 5 years. It occurs most often in people of Eastern European Jewish, French Canadian, and Cajun descent.
- ***Cystic fibrosis***—This disorder causes severe problems with breathing and digestion and can lead to early death. It is most common in non-Hispanic white individuals.

If both parents are carriers, 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.

Sex-Linked Disorders. Disorders that are caused by defective genes on the sex chromosomes are called sex-linked disorders. An example of a sex-linked disorder is ***hemophilia***. The most common type of hemophilia is caused by a ***mutation*** on the Xchromosome.

Multifactorial Disorders

Multifactorial disorders are caused by a combination of factors. Some factors are genetic, while some are non-genetic. A few of these disorders can be detected during pregnancy. Sometimes they can be corrected with surgery. Examples of multifactorial disorders include the following:

- Abdominal wall defect—The muscle and skin that cover the wall of the abdomen are missing and the bowel either is enclosed in a clear sac (omphalocele) or sticks out through a hole in the abdominal wall (gastroschisis).
- Cleft palate—A gap occurs in the roof of the mouth.
- Hydrocephalus—In this condition, fluid builds up in the skull. It can cause an enlarged head, seizures, and brain abnormalities.
- ***Neural tube defects***—These defects occur when the fetal spine does not close normally.

Risk Factors

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. When you have your prepregnancy checkup or start prenatal care, your health care professional may ask you questions about your health and family history. You are at increased risk if

- you have a genetic disorder
- you already have a child who has a genetic disorder
- there is a family history of a genetic disorder
- you belong to an ethnic group that has a high rate of carriers of certain genetic disorders

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.

Testing

Many types of tests are available to help 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.

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. Your health care professional or a genetic counselor can discuss the options with you and help you decide.

Carrier Tests

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. Carrier tests are a type of screening test that can show if a person carries a gene for an inherited disorder. Carrier tests can be done before or during pregnancy. Cystic fibrosis carrier screening is offered to all women of reproductive age because it is one of the most common genetic disorders.

Screening Tests

Screening tests often are part of routine prenatal care and are done at different times during pregnancy. Screening tests cannot tell whether the fetus actually has a disorder, only the risk that a specific defect is present.

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. There are no risks to the unborn baby with having these screening tests.

Diagnostic Tests

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 carry risks, including an increased risk of pregnancy loss.

Diagnostic tests are done on cells 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. A ***karyotype*** is a way to study chromosomes. A set of chromosomes from a cell is magnified, photographed, and arranged in order of size. A karyotype can show whether there are missing, extra, or damaged chromosomes. ***Fluorescence in situ hybridization*** and ***microarray analysis*** also are techniques that can detect common aneuploidies. Tests for specific genetic disorders, such as cystic fibrosis, also can be done.

Finally…

Most babies are born healthy, but a small number are born with a birth defect. Screening tests can help detect the risk of a genetic disorder, while diagnostic tests are available for some genetic disorders. Your health care professional or a genetic counselor will assess your risk of having a child with a genetic disorder and can help you decide which tests are best suited for your individual situation.

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: 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: 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.

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 stage of prenatal development that starts 8 weeks after fertilization and lasts until the end of pregnancy.

Fluorescence in Situ Hybridization: A laboratory technique that is used to screen for common chromosome problems, such as trisomy 21, in cells obtained by amniocentesis or chorionic villus sampling. Results are available fairly quickly because the cells do not need to be grown in a culture prior to testing.

Genes: Segments of DNA that contain instructions for the development of a person's physical traits and control of the processes 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 mutation on the X chromosome. Affected individuals are usually males who 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.

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

Microarray Analysis: 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 variations that have gone undetected by conventional genetic tests.

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

Mutation: A permanent change in a gene that can be passed on from parent to child.

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 signs or symptoms.

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

Sickle Cell Disease: An inherited disorder in which red blood cells have a crescent shape, causing chronic anemia and episodes of pain. It occurs most often in African Americans.

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

Tay–Sachs Disease: An inherited disorder that causes intellectual disability, blindness, seizures, and death, usually by age 5 years. It most commonly affects people of Eastern and Central European Jewish, Cajun, and French Canadian descent, but it can occur in anyone.

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

Trisomy 13 (Patau Syndrome): A chromosomal disorder that causes serious problems with the brain and heart as well as extra fingers and toes, cleft palate and lip, and other defects. Most infants with trisomy 13 die within the first year of life.

Trisomy 18 (Edwards Syndrome): A chromosomal disorder that causes severe intellectual disability and serious physical problems such as a small head, heart defects, and deafness. Most of those affected with trisomy 18 die before birth or within the first month of life.

Trisomy 21 (Down Syndrome): A chromosomal disorder in which abnormal features of the face and body, medical problems such as heart defects, and intellectual disability occur. Many children with Down syndrome live to adulthood.

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 but does not usually cause developmental delays.

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

This Patient Education Pamphlet was developed by the American College of Obstetricians and Gynecologists. Designed as an aid to patients, it sets forth current information and opinions on subjects related to women's health. The average readability level of the series, based on the Fry formula, is grade 6–8. The Suitability Assessment of Materials (SAM) instrument rates the pamphlets as "superior." To ensure the information is current and accurate, the pamphlets are reviewed every 18 months. The information in this pamphlet 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.

ISSN 1074-8601

To order print copies of Patient Education Pamphlets, please call 800-762-2264 or order online at sales.acog.org.

The American College of Obstetricians and Gynecologists
409 12th Street, SW
PO Box 96920
Washington, DC 20090-6920