PATIENT EDUCATION

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

Prenatal Genetic Diagnostic Tests

*Prenatal genetic testing gives parents-to-be information about whether their **fetus** has certain **genetic disorders** before birth. Prenatal **diagnostic tests** can tell you whether your fetus actually has certain disorders. They are different from prenatal **screening tests**, which tell you the chances that your baby will be affected by certain disorders. Both screening and diagnostic tests are offered to all pregnant women.*

This pamphlet focuses on prenatal diagnostic testing and explains

- *prenatal testing options*
- *types of diagnostic tests*
- *test results and what they mean*
- *deciding whether to have diagnostic testing*

Overview of Genetic Disorders

Genetic disorders are caused by changes in a person's ***genes*** or ***chromosomes*** (see box "Genes and Chromosomes"). ***Aneuploidy*** is a condition in which there are missing or extra chromosomes. Inherited disorders are caused by changes in genes called ***mutations***. Inherited disorders include ***sickle cell disease***, ***cystic fibrosis***, ***Tay–Sachs disease***, and many others. In most cases, both parents must carry the same gene to have an affected child.

Prenatal Testing Options

There are two general types of prenatal tests for genetic disorders: 1) screening tests and 2) diagnostic tests. Screening tests can tell you your risk of having a baby with certain disorders. They include carrier testing and prenatal genetic screening tests:

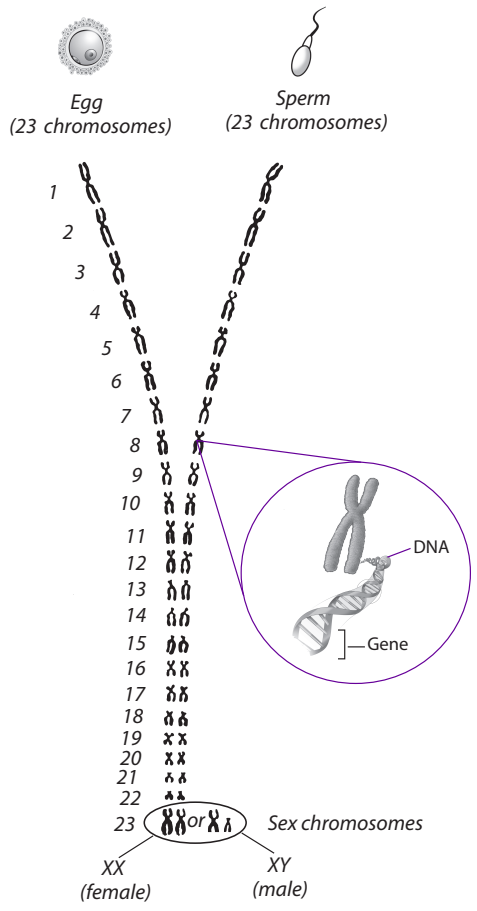
- ***Carrier screening*** is done on one or both parents (or those just thinking about becoming parents) using a sample of blood or tissue swabbed from inside the cheek. Also called "gene testing," these tests are used to find out whether a person carries a gene for certain inherited disorders. Carrier screening can be done before or during pregnancy.
- Prenatal genetic screening tests of the pregnant woman's blood and findings from ***ultrasound exams*** can screen the fetus for aneuploidy; defects of the brain and spine called ***neural tube defects***; and some defects of the abdomen, heart, and facial features.

Prenatal diagnostic tests can tell you, with as much certainty as possible, whether the fetus actually has an aneuploidy or specific inherited disorders for which you request testing. These tests are done on ***cells*** from the fetus or ***placenta*** obtained through ***amniocentesis*** or ***chorionic villus sampling (CVS)***. This pamphlet focuses on these types of tests.

Genes and Chromosomes

A gene is a small piece of hereditary material called ***DNA***. 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.

A baby's sex depends on the ***sex chromosomes*** it gets. Egg cells contain only an X chromosome. Sperm cells can carry an X chromosome or a Y chromosome. A combination of XX results in a girl and XY results in a boy.



Types of Prenatal Diagnostic Tests

The cells needed for prenatal diagnostic testing can be obtained in different ways. The choice of which procedure to use depends on when you are having the test, how quickly the results are needed, and the recommendations of your health care professional.

Amniocentesis

Amniocentesis usually is done between 15 weeks and 20 weeks of pregnancy, but it also can be done up until you have the baby. To perform the test, a very thin needle is used to withdraw a small amount of ***amniotic fluid***. Ultrasound is used to guide the procedure. Depending on the way the cells are analyzed and the information that you want, results can take from 1 day to several weeks. There is a very small chance of pregnancy loss with amniocentesis. Leakage of amniotic fluid and slight bleeding can occur after amniocentesis. In most cases, both stop on their own.

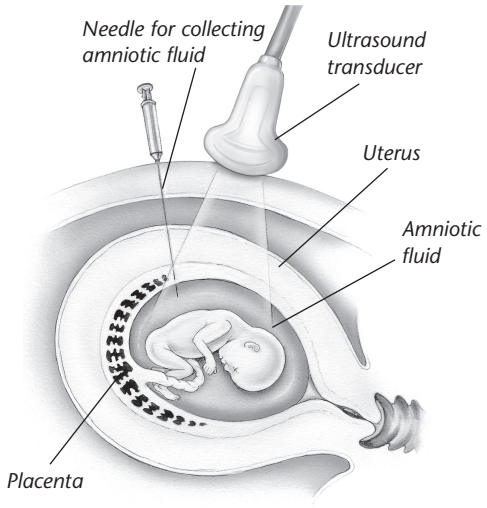
Chorionic Villus Sampling

In CVS, a sample of tissue is taken from the placenta. The main advantage of having CVS over amniocentesis is that CVS is performed earlier than amniocentesis, between 10 weeks and 13 weeks of pregnancy. The chance of miscarriage with CVS is slightly higher than the chance of miscarriage with amniocentesis.

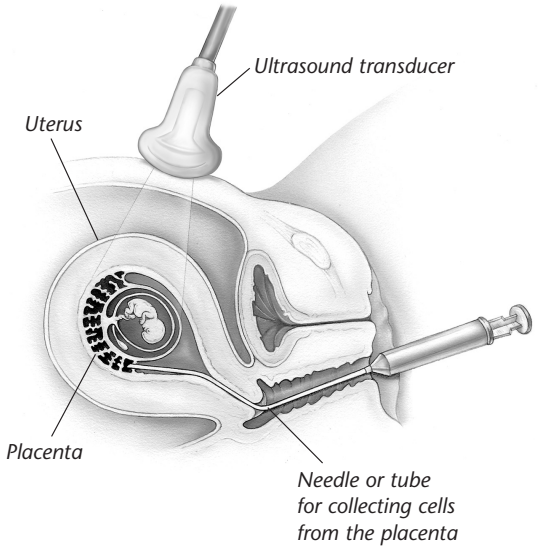
Preimplantation Genetic Diagnosis

Preimplantation genetic diagnosis may be offered to couples who are using ***in vitro fertilization (IVF)*** to become pregnant and who are at increased risk of having a baby with a genetic or chromosomal disorder. Before an ***embryo*** is transferred to a woman's uterus, it is tested for certain genetic disorders and mutations. Only embryos that do not test positive for the disorders are transferred.

Amniocentesis



Chorionic Villus Sampling



How the Cells Are Analyzed

A number of technologies are used in prenatal diagnostic testing. Your ***obstetrician*** or ***genetic counselor*** can assess what information is being sought and help select the tests that are best for your situation.

- ***Karyotype***—Missing, extra, or damaged chromosomes can be detected by taking a picture of the chromosomes and arranging them in order from largest to smallest. This is called a karyotype. Karyotyping results are ready in 1–2 weeks after the cells are sampled.
- ***Fluorescence in situ hybridization (FISH)***—This technique can be used to detect common aneuploidies involving chromosomes 13, 18, and 21 and the X and Y chromosomes. Results are ready more quickly (usually within 1–2 days) than with traditional karyotyping. Positive test results are confirmed with a karyotype.
- Chromosome ***microarray*** analysis—This test can look for different kinds of chromosome problems, including aneuploidy, throughout the entire set of chromosomes. It can find some chromosome problems that karyotyping can miss. Results can be ready in about 7 days.
- DNA testing—Tests for specific gene mutations can be done by request. For example, if you and your partner are carriers of the cystic fibrosis gene, you may want to request prenatal diagnostic testing for this specific mutation.

Diagnostic Test Results

Most of the time, the results of a diagnostic test are negative (normal). A negative result does not rule out the possibility that the baby will have a genetic disorder. It only tells you that the baby does not have the particular disorder that was tested for. Prenatal testing cannot detect all possible disorders or problems in a fetus.

If a diagnostic test result is positive (it shows that the baby has the disorder tested for), your obstetrician or genetic counselor can help explain the results and provide guidance about your choices and options. A specialist in the disorder can help you understand the life expectancy of the disorder, whether treatment is available, and the care that your child will need.

In addition to your obstetrician or other health care professional, support groups, counselors, and social workers can listen to your concerns and help answer questions. It may be possible to have additional testing, such as a specialized ***ultrasound exam***, to find out more detail about the defect. In some cases, though, it is not possible to predict whether the defect will be mild or severe.

Deciding Whether to Have Diagnostic Testing

Screening and diagnostic tests are voluntary. It is your choice whether to have these tests. Your personal beliefs and values are important factors in the decision about prenatal testing.

It can be helpful to think about what you would do if a diagnostic test result comes back positive. Some parents want to know beforehand if their child will be born with a genetic disorder. This gives parents time to learn about the disorder and plan for medical care that the child may need. If the disorder is very serious and the life expectancy is short, ***hospice care*** for the baby can be planned. Some parents may decide to end the pregnancy in certain situations. Other parents do not want to know this information before the child is born. They may decide not to have any testing at all. There is no right or wrong answer.

Choosing Between Screening and Diagnostic Testing

Any woman of any age, regardless of risk factors, can choose to have diagnostic testing instead of or in addition to screening. The main benefit of having diagnostic testing instead of screening is that it can detect all conditions caused by an extra chromosome and many other disorders in which chromosomes are missing or damaged. Diagnostic tests also are available for many inherited disorders. The main disadvantage is that diagnostic testing carries a very small risk of losing the pregnancy.

Your obstetrician or other health care professional may review your risk factors for having a baby with a genetic disorder. Factors that increase the risk of having a child with a genetic disorder include the following:

- One or both parents are older.
- One or both parents have a genetic disorder.

- The couple already has a child with a genetic disorder.
- There is a family history of a genetic disorder.

Even if you have risk factors, it does not mean that your baby will have a disorder. A genetic counselor or other health care professional with expertise in genetics can be useful in some situations. A genetic counselor can study your family health history, recommend specific tests, and interpret test results.

Timing of Prenatal Testing

Keep in mind that certain tests can be done only at certain times during pregnancy. For example, if you first seek prenatal care after 20 weeks of pregnancy, some tests will not be available to you. Tests that are done earlier allow parents more time to make decisions if a test result is positive. If ending the pregnancy is being considered, it is safer to do so within the first 13 weeks of pregnancy.

Finally…

Diagnostic tests are able to detect some genetic disorders before a baby is born. Most of the time, the results of a test confirm that the baby is healthy. If the results are abnormal, more testing may be done to clarify the results. Your health care professional or genetic counselor can help guide you through your options.

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.

Amniotic Fluid: Fluid in the sac that holds the fetus.

Aneuploidy: Having an abnormal number of chromosomes.

Carrier Screening: A test done on a person without signs or symptoms to find out whether he or she carries a gene for a genetic disorder.

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: 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.

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

Embryo: The stage of development that starts at fertilization (joining of an egg and sperm) and lasts up to 8 weeks.

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

Fluorescence In Situ Hybridization (FISH): A screening test for common chromosome problems. The test is done using a tissue sample from an amniocentesis or chorionic villus test.

Genes: Segments of DNA that contain 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.

Genetic Disorders: Disorders caused by a change in genes or chromosomes.

Hospice Care: Care that focuses on comfort for people who have an illness that will lead to death.

In Vitro Fertilization (IVF): A procedure in which an egg is removed from a woman's ovary, fertilized in a laboratory with the man's sperm, and then transferred to the woman's uterus to achieve a pregnancy.

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

Microarray: 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 changes that can be missed by the routine genetic tests.

Mutations: Changes in a gene that can be passed from parent to child.

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

Obstetrician: A doctor who cares for women during pregnancy and their labor.

Placenta: An organ that provides nutrients to and takes waste away from the fetus.

Preimplantation Genetic Diagnosis: A type of genetic testing that can be done during in vitro fertilization. Tests are done on the fertilized egg before it is transferred to the uterus.

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

Sex Chromosomes: The chromosomes that determine a person's sex. In humans, there are two sex chromosomes, X and Y. Females have two X chromosomes and males have an X and a Y chromosome.

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

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

Tay–Sachs Disease: An inherited disorder that causes mental disability, blindness, seizures, and death, usually by age 5. It most commonly affects people of Eastern or Central European Jewish backgrounds, as well as people of French Canadian and Cajun backgrounds.

Ultrasound Exams: Tests in which sound waves are used to examine inner parts of the body. During pregnancy, ultrasound can be used to check the fetus.

This is EP164 in ACOG's Patient Education Pamphlet Series.

ISSN 1074-8601

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