



Diagnostic Tests for Birth Defects

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What are prenatal diagnostic tests?

Prenatal **diagnostic tests** are tests that can detect certain disorders before a baby is born.

How are prenatal diagnostic tests different from screening tests?

Prenatal **screening tests** show only whether you are at high risk or low risk of having a baby with a particular disorder. They cannot tell you whether the baby actually has the disorder.

What is a birth defect?

A **birth defect** is a physical problem that is present at birth, although some birth defects may not be noticed until the child is older.

What is a genetic disorder?

Many birth defects are caused by changes in a person's **chromosomes** or **genes**. These types of disorders are called genetic disorders. For about 70% of birth defects, the cause is not known.

What are some types of genetic disorders?

Types of genetic disorders include the following:

- **Aneuploidy**—Aneuploidy is a condition of having missing or extra chromosomes. The most common aneuploidy is called a **trisomy**, in which there is an extra chromosome. Examples include **trisomy 21 (Down syndrome)**, **trisomy 13 (Patau syndrome)**, and **trisomy 18 (Edwards syndrome)**.

- Structural chromosome disorders—Pieces of chromosomes are missing, repeated, or broken off and moved to another chromosome.
- Inherited disorders—These disorders are caused by changes in genes called **mutations**. Mutations can be passed down from parents to their children, or they can appear for the first time in a child. Some inherited disorders are more common in certain races and ethnic groups, such as **sickle cell disease** (African American), **cystic fibrosis** (non-Hispanic white), and **Tay-Sachs disease** (Ashkenazi Jewish and French Canadian).
- Multifactorial disorders—Multifactorial disorders are caused by a number of different factors working together.

What types of genetic disorders can diagnostic tests detect?

Diagnostic tests are available for some, but not all, inherited defects and many chromosomal disorders.

For whom are prenatal diagnostic tests recommended?

Diagnostic tests may be done if a screening test result shows an increased risk of a birth defect or if a couple has risk factors that increase their risk of having a baby with certain birth defects. However, both screening and diagnostic testing may be offered to all pregnant women. You do not have to be a certain age or have a family history of a disorder to have diagnostic tests.

What are the main benefits and disadvantages of having diagnostic testing?

The main benefit of having diagnostic testing instead of screening is that it can detect all trisomies 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 tests can pose some risks to the pregnancy.

How are diagnostic tests for genetic disorders performed?

Diagnostic tests are done on **cells** from the **fetus** obtained through **amniocentesis** or **chorionic villus sampling (CVS)**. The cells can be analyzed using different techniques.

When and how is amniocentesis usually done?

Amniocentesis usually is done between 15 weeks and 20 weeks of pregnancy. To perform the test, a very thin needle is used to withdraw a small amount of **amniotic fluid**. There is a small chance of miscarriage (about 1 miscarriage in every 300–500 procedures) with amniocentesis. This risk may be even lower when amniocentesis is performed by an experienced health care provider.

When and how is chorionic villus sampling usually done?

CVS is performed earlier than amniocentesis, between 10 weeks and 13 weeks of pregnancy. In CVS, a sample of tissue is taken from the **placenta**. When CVS is performed by an experienced health care provider and in a center that performs many of these procedures, the risk of miscarriage with CVS is about the same as the risk with amniocentesis.

What is preimplantation genetic diagnosis?

Preimplantation genetic diagnosis may be offered to couples who are using **in vitro fertilization** 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 have the disorder are transferred.

How are fetal cells tested for genetic disorders?

Several techniques can be used to test fetal cells:

- **Karyotype**—Missing, extra, or damaged chromosomes can be detected by taking a picture of the chromosomes and arranging them in order from smallest to largest. Karyotyping takes about 2–3 weeks to perform.
- **Fluorescence in situ hybridization**—This technique can be used to detect common aneuploidies. Results are available more quickly (usually within 1–2 days) than with traditional karyotyping. Positive test results are confirmed with a karyotype.
- **Microarray** analysis—This technique is used to look for different kinds of abnormalities throughout the entire set of chromosomes. This test can tell you a lot of information, but whether everything that is found is a cause for concern is uncertain. It is important to understand what types of information may be detected and how it can and cannot be used before having this type of analysis.

What happens if diagnostic tests show that there is a problem?

If diagnostic tests show that there is a problem, a **genetic counselor** or other health care provider can help explain the results and provide guidance in making choices and considering options. It may be possible to have additional testing to find out more detail about the defect. You may be referred to a specialist in pediatrics or a **neonatologist** to discuss how the disorder will affect your baby. Before you make any decisions, you should know the life expectancy of the disorder, the

care that your child will need, and whether the disorder can be treated. You also may want to seek out support groups, counselors, and social workers to discuss your concerns and get additional information.

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.

Amniotic Fluid: Water in the sac surrounding the fetus in the mother's uterus.

Aneuploidy: Having an abnormal number of chromosomes.

Birth Defect: A physical problem that is present at birth.

Cells: The smallest units of a structure in the body; 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 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.

Embryo: The developing organism from the time it implants in the uterus up to 8 completed weeks of pregnancy.

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

In Vitro Fertilization: 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 variations that have gone undetected by conventional genetic tests.

Mutations: Permanent changes in genes that can be passed on from parent to child.

Neonatologist: A doctor who specializes in the diagnosis and treatment of disorders that affect newborn infants.

Placenta: Tissue that provides nourishment 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 performed 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.

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.

Tay-Sachs Disease: An inherited birth defect that causes intellectual disability, blindness, seizures, and death, usually by age 5 years. It most commonly affects people of Eastern European Jewish (Ashkenazi Jews), 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 babies with trisomy 18 die before birth or within the first month 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. Many children with Down syndrome live to adulthood.

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

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