



The American College of  
Obstetricians and Gynecologists



FREQUENTLY ASKED QUESTIONS  
FAQ164  
PREGNANCY

## Diagnostic Tests for Birth Defects

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### What are diagnostic tests for birth defects?

Prenatal diagnostic tests are able to detect some birth defects before a baby is born.

### What is a birth defect?

A birth defect is a problem that is present at birth, although it may not be noticed until the child is older. Birth defects may affect any part of the body, including major organs such as the heart, lungs, or brain. The defect may affect the baby's appearance, a body function, or both.

### What causes birth defects?

Chromosomal defects are caused by missing, damaged, or extra **chromosomes**. Common chromosomal disorders are **Down syndrome** and **trisomy 18**.

Inherited disorders are caused by defective **genes**. These disorders are passed down by parents to their children. 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).

Birth defects also may be caused by being exposed to harmful agents, such as medications, chemicals, and infections. Some birth defects may be caused by a combination of all of these factors. For about 70% of babies born with birth defects, the cause is not known.

## What other types of tests are available in addition to diagnostic tests to address concerns about birth defects?

- Screening tests—Screening tests assess the risk that a baby will have Down syndrome and trisomy 18, as well as **neural tube defects**. 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 an **ultrasound exam** and blood tests that measure the level of certain substances in the mother's blood (see [FAQ165 Screening Tests for Birth Defects](#)).
- Carrier tests—Carrier tests are screening tests 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 (see [FAQ179 Preconception Carrier Screening](#)).

## What types of disorders can diagnostic testing detect?

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

## For whom are 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. Diagnostic tests also are available as a first choice for all pregnant women, even for those who do not have risk factors.

## What are the advantages and disadvantages of having diagnostic tests compared with having screening tests?

The main benefit of having diagnostic testing instead of screening is that it can detect all chromosomal disorders in which there is an extra chromosome, as well as many other disorders in which chromosomes are missing or damaged. The main disadvantage is that invasive diagnostic tests can pose some risks to the pregnancy.

## What diagnostic tests are available?

Common tests currently used to diagnose birth defects include the following:

- Targeted ultrasound exam
- **Amniocentesis**
- **Chorionic villus sampling (CVS)**
- Fetal blood sampling

## What is a targeted ultrasound exam?

A targeted ultrasound exam focuses on defects in specific organs or features that may indicate a chromosomal disorder. Although a targeted ultrasound exam is useful in showing a defect or feature in more detail, it cannot diagnose the underlying cause of a defect. Amniocentesis, CVS, or fetal blood sampling still may be needed to find the cause.

## What is amniocentesis?

Amniocentesis is a prenatal diagnostic test performed between week 15 and week 20 of pregnancy. To perform amniocentesis, a thin needle is guided through the woman's abdomen and uterus. A small sample of **amniotic fluid** is withdrawn and sent to a lab. Amniotic fluid contains cells from the **fetus** that can be analyzed using different techniques.

## What complications are associated with amniocentesis?

Complications of amniocentesis may include cramping, vaginal bleeding, infection, and leaking amniotic fluid. There is a small chance of miscarriage (about 1 miscarriage in every 300 to 500 procedures). The risk of these complications may be even lower when amniocentesis is performed at a hospital or center that is experienced in doing these procedures.

## What is chorionic villus sampling (CVS)?

CVS is performed earlier than amniocentesis, after 9 weeks of pregnancy. To perform CVS, a small sample of tissue is taken from the **placenta**. The tissue contains cells with the same genetic makeup as the fetus. Cells are analyzed in the same way as those obtained from amniocentesis.

## What complications are associated with CVS?

Complications of CVS include leakage of amniotic fluid and infection. 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 may be the same as the risk with amniocentesis.

## What is fetal blood sampling?

Fetal blood sampling is a specialized test that may not be available at all centers. Fetal blood sampling may be done at 18 weeks of pregnancy or later. To perform the test, a needle is guided through the abdomen and the wall of the uterus, and blood is taken from the vein in the umbilical cord.

## What complications are associated with fetal blood sampling?

Possible complications of this procedure are infection, bleeding, and leakage of amniotic fluid. There is a small (1–2%) chance of miscarriage after fetal blood sampling.

## What happens if a diagnostic test result shows that there is a problem?

Your health care provider or a **genetic counselor** 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, but sometimes it is not possible to tell how severe a birth defect will be. You may be referred to a specialist in pediatrics or a **neonatologist** to discuss how your pregnancy will be affected and the care that the baby will need. 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.

**Chorionic Villus Sampling (CVS):** A procedure in which a small sample of cells is taken from the placenta and tested.

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

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

**Down Syndrome:** A genetic disorder in which mental retardation, abnormal features of the face and body, and medical problems such as heart defects occur.

**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.

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

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

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

**Placenta:** Tissue that provides nourishment to and takes waste away from the fetus.

**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 mental retardation, blindness, seizures, and death, usually by age 5 years. It occurs mostly in people of eastern European Jewish descent (Ashkenazi Jews) and among French Canadians.

**Trisomy 18:** A genetic disorder that causes serious problems with development. Most infants with trisomy 18 die within the first year of life.

**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.

**FAQ164:** 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.

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