

FAQs Amniocentesis

Frequently Asked Questions

What is amniocentesis?

Amniocentesis is a procedure that gathers fluid and cells from your uterus during pregnancy. The sample is then tested to find out whether your pregnancy has certain genetic disorders. This diagnostic test also can look for problems with infection or with lung development.

You can choose to have amniocentesis instead of or in addition to other prenatal genetic tests (see Prenatal Genetic Diagnostic Tests). You may hear amniocentesis called an "amnio" for short.

What does amniocentesis test for?

Amniocentesis can tell you if your pregnancy has missing, damaged, or extra chromosomes. These are signs of a genetic disorder. Amniocentesis also can tell you whether your pregnancy has a specific disorder caused by a genetic mutation. Examples of these disorders include

- cystic fibrosis (CF)
- Down syndrome (trisomy 21)
- sickle cell disease
- Tay-Sachs disease

When is amniocentesis usually done?

Amniocentesis usually is done between 15 and 20 weeks of pregnancy, but it also can be done up until you give birth. It is your choice whether you want to have this test.

How is amniocentesis done?

Amniocentesis usually is done in an office or medical center. The procedure includes the following steps:

- 1. A very thin needle is inserted through the abdomen (belly) and into the uterus. An ultrasound exam is used to guide the procedure.
- A small sample of amniotic fluid is withdrawn from the sac surrounding the fetus.This fluid contains cells that have been shed by the fetus.
- **3.** The sample is sent to a lab for testing.

You and your obstetrician—gynecologist (ob-gyn) can discuss the disorders that you want to test for. Depending on the way the cells are analyzed and the information that you want, results can take from 1 day to several weeks.

What do amniocentesis results mean?

If your test result is negative, it means the pregnancy does not have the disorder that was tested for. But keep in mind that a negative result does not rule out the possibility that the pregnancy has a genetic disorder that was not tested for.

If your test result is positive, it means the pregnancy has the disorder that was tested for. You and your ob-gyn or a genetic counselor should discuss your results and what they mean. They can provide guidance about your choices and options. You also may want to

- talk with a specialist in the disorder who can help you understand life expectancy,
 whether treatment is available, and the care that your child will need
- talk with support groups, counselors, and social workers
- seek additional testing, such as a specialized ultrasound exam

What are the risks of amniocentesis?

Amniocentesis is a safe and highly accurate procedure. While minimal, the risks with amniocentesis include

- a very small chance of miscarriage (about 1 in 900 procedures)
- leakage of amniotic fluid and slight bleeding (which should stop on their own)
- transmission of blood-borne infections such as hepatitis B, hepatitis C, or human immunodeficiency virus (HIV) from the woman to the fetus

Talk with your ob-gyn about the risks of amniocentesis, especially if you have one of the infections listed above.

What are alternatives to amniocentesis?

Chorionic villus sampling (CVS) is the primary alternative to amniocentesis. In this procedure, a sample of tissue is taken from the placenta. The main advantage of CVS is that it can be done earlier, between 10 and 13 weeks of pregnancy. But it also comes with a slightly higher risk of miscarriage (about 1 in 455 procedures).

If you might consider having an abortion, it is safer to do so before week 13 of pregnancy. In this case, CVS may be preferred over amniocentesis because it gives you more time to decide what to do if a test result is positive.

Talk with your ob-gyn about which test is right for you. See Prenatal Genetic Diagnostic Tests for more information.

Who should consider amniocentesis?

All women may have diagnostic testing such as amniocentesis. But amniocentesis or other diagnostic tests may be recommended if you have an increased risk of having a baby with a birth defect. You and your ob-gyn can discuss all of the testing options and your individual risk factors.

Your risk of having a baby with a birth defect may be higher if you are 35 or older, or if you have

had a screening test that shows an increased risk of a birth defect

a genetic disorder

a child with a genetic disorder

a family history of a genetic disorder

Some genetic disorders also are more common in certain ethnic groups.

What else should I consider when deciding whether to have amniocentesis?

It can be helpful to think about what you would do if the result comes back positive.

Some people want to know if their child will be born with a genetic disorder. This gives them time to learn about the disorder and plan for medical care 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 people may decide to end the pregnancy in certain situations.

Other people 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.

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.

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 (CF): An inherited disorder that causes problems with breathing and

digestion.

Diagnostic Test: A test that looks for a disease or cause of a disease.

Down Syndrome (Trisomy 21): A genetic disorder that causes abnormal features of the face and body, medical problems such as heart defects, and mental disability. Most cases of Down syndrome are caused by an extra chromosome 21 (trisomy 21).

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

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.

Hepatitis B: An infection caused by a virus that can be spread through blood, semen, or other body fluid infected with the virus.

Hepatitis C: An infection caused by a virus that can be spread by sharing needles used to inject drugs.

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

Human Immunodeficiency Virus (HIV): A virus that attacks certain cells of the body's immune system. If left untreated, HIV can cause acquired immunodeficiency syndrome (AIDS).

Miscarriage: Loss of a pregnancy that is in the uterus.

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

Obstetrician—**Gynecologist (Ob-Gyn):** A doctor with special training and education in women's health.

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

Screening Test: A test that looks 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. The disorder causes chronic anemia and episodes of pain.

Tay-Sachs Disease: An inherited disorder that causes mental disability, blindness, seizures, and death, usually by age 5.

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

Uterus: A muscular organ in the female pelvis. During pregnancy, this organ holds and nourishes the fetus. Also called the womb.

If you have further questions, contact your ob-gyn.

Don't have an ob-gyn? Learn how to find a doctor near you.

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