



Amniocentesis

What is amniocentesis?

Amniocentesis is a test that is mainly used to learn about the chromosomes of a developing baby. Also called an “amnio,” this test is usually performed between 15 and 20 weeks of pregnancy. The test is done by removing a sample of the amniotic fluid that surrounds the developing baby. The fluid is then analyzed for chromosome abnormalities.

Chromosomes are the packages of genetic information that we inherit from our parents and pass on to our children. Many chromosome abnormalities cause intellectual disability and birth defects. Although it is possible for a woman of any age to have a baby with a chromosome abnormality, such as Down syndrome, the chance increases as she gets older.

In addition to finding chromosome abnormalities, amniocentesis can also help find certain other birth defects such as spina bifida, an abnormality of the spinal column. Amniocentesis, however, cannot detect all birth defects or all causes of intellectual disability.

How is it done?

You do not need to do anything special to prepare for the test. It begins with an ultrasound exam. Ultrasound uses sound waves directed at your developing baby to make an image on a video screen. The ultrasound will measure the size of the baby. It will also provide a look at the placenta and the amniotic fluid in which the baby is floating.

A trained doctor performs the amniocentesis. The doctor puts a thin needle through your abdominal wall and into the bag of amniotic fluid that surrounds your developing baby. Ultrasound is used to guide the needle during the entire procedure. You may feel some discomfort when the needle is put in, but your baby will not feel any pain.

The doctor removes about 2 tablespoons of fluid. This step takes about one minute. The fluid is sent to the laboratory in a labeled test tube. Cells from your baby are in the fluid and are placed into a cell culture to grow in the lab. After about 10 to 14 days, the chromosomes from the baby’s cells are studied and counted. Final results are available about 2 weeks after the test.

An amniocentesis appointment lasts less than an hour, but the procedure itself takes only a couple of minutes. Most of the appointment is spent looking at the baby with ultrasound and preparing for the amniocentesis.

Is amniocentesis safe?

In every pregnancy, there is a risk for a problem such as a miscarriage, or loss of the pregnancy. Amniocentesis, although considered safe, adds a small additional risk for a problem that may lead to a miscarriage. The risk for this kind of problem after amniocentesis is less than 1 in 300.

After the test, you will be given an instruction sheet with information about what to expect. We recommend that you do not do any heavy lifting, exercise, or have sexual intercourse for 24 hours after your amniocentesis. Mild cramping and tenderness where the needle was inserted are common and not a cause for concern. If you experience painful cramping, leakage of fluid, bleeding, or fever you should call your doctor, nurse practitioner or nurse midwife right away.

Are the results accurate?

Amniocentesis results are considered more than 99.8 percent accurate. In other words, pregnancies with extra or missing chromosomes (or parts of chromosomes) are detected with accuracy by amniocentesis. This includes Down syndrome, which is caused by an extra chromosome #21 in all of the cells.

There are many kinds of birth defects, however, that cannot be found by amniocentesis. Some of these include intellectual disability not related to Down syndrome, as well as heart defects, cleft palate, and other conditions.

Most amniocentesis results are normal

Most women who have amniocentesis learn that their baby's chromosomes are normal. We will phone you with the test results. Amniocentesis also shows the sex of the baby. When you get the results, you may ask the sex of the baby, or you may choose to wait until the baby is born to find this out. It is your choice.

Keep in mind that normal amniocentesis results are not a guarantee that the baby has no health problems or birth defects.

What if the amniocentesis results are not normal?

Once in a while, an amniocentesis result reveals that the developing baby has a chromosome abnormality. Chromosome abnormalities are not curable. If this happens, you and your partner will be offered genetic counseling immediately. This may include discussion with one or more of the following people:

- your doctor, nurse practitioner or nurse midwife
- your genetic counselor
- a geneticist
- a perinatologist
- another pediatric specialist

You will have the opportunity to discuss all available treatments and options for continuing or ending the pregnancy. Whether to continue or end a pregnancy is entirely your decision. Genetic counselors offer ongoing support to families regardless of the options they choose.

Should I have amniocentesis?

Before deciding if you want amniocentesis, you may want to explore other options, such as prenatal screening tests like:

- Serum Integrated Screening
- Sequential Integrated Screening
- Quad Marker Screening (for women 14 weeks or later in pregnancy)

You may also choose an alternative diagnostic procedure, such as Chorionic Villus Sampling (CVS), to test the baby's chromosomes. Your doctor, nurse practitioner, or nurse midwife can review these options with you.

If you decide you want amniocentesis, you will have the chance to meet with a genetic counselor who will review all the pros and cons of the procedure, and answer your questions.

Knowing about a chromosome abnormality during pregnancy can be useful. You may use the information to decide whether or not to continue the pregnancy. Even if you would not choose to end your pregnancy, knowing about a birth defect before delivery may help you feel more prepared when your baby is born. In some cases, information about the baby's condition may be beneficial to the medical professionals involved in your prenatal care.

Ultimately, the decision to have an amniocentesis is entirely yours.



Additional resources

- Connect to the Genetics Web site at <http://genetics.kp.org> to learn more about prenatal testing for birth defects.
- Visit your doctor's home page at kp.org/mydoctor to use online health tools, view your Preventive Services reminders, check most lab results, and much more.
- Contact your Kaiser Permanente Health Education Center or Department for health information, programs, and other resources.
- Your health and your pregnancy can be seriously affected by violence or abuse. If you are hit, hurt or threatened by a partner or spouse, there is help. Call the National Domestic Violence Hotline at 1-800-799-7233 or connect to ndvh.org.

This information is not intended to diagnose health problems or to take the place of medical advice or care you receive from your physician or other health care professional. If you have persistent health problems, or if you have additional questions, please consult with your doctor.