Amniocentesis

What is amniocentesis?
Amniocentesis is a test mainly used to learn about the chromosomes of a developing baby. Also called an “amnio,” this test is usually scheduled between 15 and 20 weeks of pregnancy. The test is done by removing a small sample of amniotic fluid (the fluid around the developing baby) and testing the sample for chromosome conditions.

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

Amniocentesis can find chromosome conditions before birth. Amniocentesis can also help find spina bifida, a condition of the spinal column. In some pregnancies, amniocentesis may also include genetic testing to look at a developing baby’s DNA code. This type of genetic testing is only done when the baby is at risk for a specific genetic condition. However, amniocentesis does not test for 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 measures the size of the baby. It also provides 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 (belly) and into the bag of amniotic fluid that surrounds your developing baby. You will feel some discomfort when the needle is put in. Ultrasound is used to guide the needle during the entire procedure. The doctor avoids touching the baby with needle.

The doctor takes out about 2 tablespoons of fluid, and 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 processed in the lab. The chromosomes from the sample can be studied after about 10 to 14 days. Final results are usually 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 miscarriage, or loss of the pregnancy. Amniocentesis, although considered safe, adds a small additional risk for a problem that could lead to a miscarriage. The added risk for miscarriage after amniocentesis is 1 in 500 or less.

After the test, you will be given 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 at the needle site are common and not a cause for concern. If you experience painful cramping, bleeding, fluid leaking from the vagina, or fever you should call your doctor, nurse practitioner or nurse midwife right away. These symptoms are uncommon.

Are the results accurate?
Amniocentesis detects pregnancies with extra or missing chromosomes with high accuracy. This includes Down syndrome, which is caused by an extra chromosome #21 in all of the cells. Testing can also find less common chromosome conditions with extra or missing pieces of chromosomes. However, there are some rare chromosome conditions that cannot be found with amniocentesis.

Most amniocentesis results are normal
Most women who have amniocentesis learn that their baby’s chromosomes are normal. We phone you with this information. 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.

Normal amniocentesis results are not a guarantee that the baby has no health problems or birth defects. There are many birth defects that cannot be found by amniocentesis. Some of these include intellectual disability (not due to Down syndrome), as well as heart defects, cleft palate, and unexpected genetic disorders.

What if the amniocentesis result is not normal?
Once in a while, an amniocentesis result reveals that the developing baby has a chromosome condition. Chromosome conditions are not curable. If this happens, you and your partner will be offered genetic counseling right away. 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 learn about the specific chromosome condition. You will also have time 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?
Of course, getting reassurance about chromosome conditions can ease some of your worries. But learning about a chromosome condition during pregnancy may also be useful. You may use the information to decide whether or not to continue the pregnancy. You may use the information to help you feel more prepared when your baby is born. In some cases, information about the baby’s condition may change your prenatal care or help guide the best place for delivery.

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

- Integrated Screening with or without a Nuchal Translucency (NT) ultrasound
- Cell-free DNA Screening (also called NIPT or non-invasive prenatal testing)
- Quad Marker Screening (for women 14 weeks or later in pregnancy)

Or you may choose an alternative diagnostic procedure to test the baby’s chromosomes such as chorionic villus sampling (CVS).

Your doctor, nurse practitioner, or nurse midwife can review these options with you. If you decide you want amniocentesis, you will meet with a genetic counselor who will review all the pros and cons of the procedure, and answer your questions.

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

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Additional resources
- Learn more about prenatal testing for birth defects on the Genetics website: http://genetics.kp.org
- For pregnancy information, health tools, and classes, visit your provider’s homepage: My Doctor Online
- Access health and drug encyclopedias, interactive programs, health classes, and much more on the Kaiser Permanente website: kp.org
- For health information, programs, and other resources, contact your local Kaiser Permanente Health Education Center or Department. Ask your doctor for the location and contact information.