KAISER PERMANENTE®

REGIONAL GENETICS DEPARTMENT

Amniocentesis

What is amniocentesis?

Amniocentesis (amnio) is a procedure to learn if a developing baby has a chromosome condition. It can also test for other genetic conditions. It is usually done between the 15th to 22nd weeks of pregnancy, but it can be done later in pregnancy, when needed.

A small sample of amniotic fluid (the fluid around the baby) is collected for testing. The fluid has cells with the baby's DNA, so an amnio can diagnose genetic or chromosome conditions before birth.

Who is offered amnio?

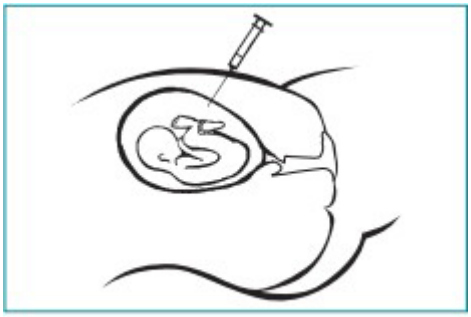
Amnio is offered to anyone who wants to know about chromosome conditions during pregnancy. Most chromosome conditions happen randomly and do not run in families. These conditions often cause intellectual disability and birth defects. It is possible for a person of any age to have a pregnancy with a chromosome condition. The chance for some chromosome conditions, like Down syndrome, increases with age.

Amnio may also be offered if the baby is at-risk for a genetic condition based on the family history, ultrasound findings, or genetic carrier testing in the parents. Amnio can also help find spina bifida, a birth defect of the spine.

How is it done?

Amnio is done by a specially trained doctor. Ultrasound is used to show the size of the baby, the location of the placenta, and the amniotic fluid around the baby. It can also detect some birth defects.

The doctor passes a very thin needle through your abdominal wall (belly) and into the fluid that surrounds the baby. You may feel some discomfort when the needle is put in. It takes about 1 minute to collect the 3 tablespoons of fluid needed for the test. Ultrasound helps the doctor safely collect the fluid without touching the baby with needle.



AMNIOCENTESIS

The fluid is sent to the laboratory in a labeled test tube. Results are usually available about 2 weeks after the amnio. A small number of samples might take longer when microarray is done.

The procedure only takes a few minutes. Most of the visit is spent looking at the baby with ultrasound and preparing for the amnio. An amnio appointment usually lasts less than an hour.

What needs to be done before an amnio appointment?

There is nothing special that needs to be done before an amnio. You can eat and drink as usual. You do not need a full bladder for this ultrasound. If you are taking a blood thinner, tell your genetic counselor.

E-Handout (Rev. Aug 2022) RL 8.5

Is amnio safe?

Every pregnancy has a chance for miscarriage or loss even when no testing is done. Amnio is considered safe, but adds a very small risk for miscarriage. The added chance for miscarriage after amnio is about 1 in 1000.

What happens after amnio?

After the test you are 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 amnio. Mild cramping and tenderness at the needle site are common and not a cause for concern. If you experience painful cramping, blood or fluid coming 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?

Amnio detects pregnancies with extra or missing chromosomes with high accuracy. This includes conditions like Down syndrome, trisomy 18, and trisomy 13. Testing can also find less common chromosome conditions with extra or missing pieces of chromosomes. However, there are very rare chromosome conditions that cannot be found by amnio.

What if the amnio results are normal?

Most people who have amnio learn that their baby's chromosomes are normal. We call you with this information. You can ask about the sex chromosomes, which predicts the baby's sex. You may also choose to wait until the baby is born to find this out. It's your choice.

Normal results do not guarantee that the baby has no health problems or birth defects. It is not possible to test for all conditions or causes of intellectual disability.

What is my chance to have a baby with a chromosome condition?

The chance for many chromosome conditions, including Down syndrome, trisomy 18, and trisomy 13, depends on how old you will be when you deliver.

- At age 20 - **1 in 525** (about 0.2%)
- At age 25 - **1 in 475** (about 0.2%)
- At age 30 - **1 in 385** (about 0.3%)
- At age 35 - **1 in 200** (0.5%)
- At age 38 - **1 in 100** (1.0%)
- At age 40 - **1 in 65** (1.5%)

The chance for a condition caused by an extra or missing piece of chromosome (called a copy number variant) is the same in any pregnancy, no matter how old you are: **About 1 in 200 (0.5%)** Microarray testing can be requested to find these type of chromosome conditions.

What if the amnio results are not normal?

Once in a while, an amnio result shows that the developing baby has a chromosome or genetic condition. If this happens, genetic counseling is offered right away. You may talk with one or more of the following people about the diagnosis:

- Your doctor, nurse practitioner or nurse midwife
- Your genetic counselor
- A geneticist
- A perinatologist
- A pediatric specialist

You will be informed about the specific condition. You will also have time to discuss available treatments and options for continuing or ending the pregnancy. The decision to continue or end a pregnancy is entirely your choice. Genetic counselors offer ongoing support to families regardless of the options they choose.

What you need to know about Amniocentesis (amnio)

- Examines fetal cells from the amniotic fluid (fluid around the baby)
- Detects chromosome conditions and open spine defects during pregnancy
- Can test for other genetic disorders in at-risk pregnancies
- Procedure is usually done between 15 to 22 weeks in pregnancy
- Uses a thin needle to go through the abdomen (belly) into the amniotic sac (bag of water)
- Activity is restricted for 24 hours after the procedure
- Risk for a miscarriage from amnio is about 1 in 1000
- Test is very accurate
- Results take about two weeks

Should I have amnio?

Think about what information you want during pregnancy. Getting normal amnio results can ease some of your pregnancy worries. It may also help to learn about a genetic or chromosome condition during pregnancy. You could use the information to decide whether or not to continue the pregnancy. You could 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 amnio, you may consider a prenatal screening test like:

- Cell-free DNA Screening
  (also called non-invasive prenatal testing/NIPT)

Or consider a different diagnostic procedure to test the baby's chromosomes, like:

- Chorionic villus sampling (CVS)

Your doctor, nurse practitioner, or nurse midwife can review these options with you. If you decide you want amnio, 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 amnio is entirely yours.

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Additional resources

- Learn more about prenatal testing for birth defects on the Genetics website: **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.

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