

# Amniocentesis

## What is amniocentesis?

Amniocentesis is a procedure done during pregnancy to test a fetus for certain kinds of birth defects. It involves the withdrawal of a small amount of amniotic fluid (the fluid surrounding a developing fetus) from the uterus. This procedure is typically performed after 15 weeks gestation.

## Who should undergo amniocentesis?

People with specific risks for chromosome abnormalities, inherited genetic diseases, open neural tube defects or other conditions for which tests are available should consider amniocentesis.

- Women age 35 or older at the time of delivery have a greater risk than women under age 35 for having a baby with Down syndrome or other chromosome abnormalities.
- If someone in the family, especially a close relative, is known to have a chromosome abnormality or inherited disease, there may be increased risk for another child to be born with the same condition. In this case, the mother's age does not matter.
- If the parents of the unborn baby have been found to be carriers of an inherited disease, the pregnancy is at increased risk for that disease. Many inherited diseases can be diagnosed before giving birth.
- If a close relative has been born with an open neural tube defect, such as spina bifida or anencephaly, there may be an increased risk to other pregnancies in the family.
- When a maternal serum screen result shows that a pregnancy has a high risk for Down syndrome, trisomy 18 or open neural tube defects, amniocentesis for diagnostic testing should be considered.
- If an ultrasound shows an abnormality, amniocentesis may be recommended.

## What tests are usually included in the genetic testing of amniotic fluid?

The test most commonly performed on amniotic fluid is a chromosome analysis, a test for disorders such as Down syndrome. Testing for individual genetic disorders such as cystic fibrosis, Tay-Sachs disease and sickle cell disease can be done on the same sample when indicated. Amniotic fluid can also be tested for the alpha fetoprotein level. This level helps determine the likelihood for open neural tube defects, such as spina bifida and anencephaly. Even identity testing to determine who the father is can be performed from an amniocentesis sample.

## What cannot be tested with amniocentesis?

Every pregnancy has a risk — usually 3 percent to 5 percent — for birth defects that do not have a known cause. A few examples are cleft lip, non-specific mental retardation and most heart defects. Amniocentesis is not able to test for these types of conditions. Furthermore, tests for single-gene disorders are not typically done unless it is known there is a significant risk to the fetus at the time of the amniocentesis procedure.

## What will I need to do before the procedure?

You should read all the material we send you, including this brochure, the consent form and the precautions that we ask you to follow after the test. Please write down any questions you have for us and bring them with you to the clinic.

Your physician will explain the amniocentesis procedure and its risks. You will be then asked to sign the informed consent form. You may decide not to have the procedure up to the time that we perform it.

### **How is amniocentesis performed?**

Before the procedure, the doctor will perform an ultrasound scan (sonogram) which shows a picture of the uterus, the placenta, the amniotic fluid and the fetus on a screen. After reviewing the image, the doctor will insert a very thin needle through the woman's abdomen into the uterus and draw out about 1 ounce of amniotic fluid. This part of the procedure lasts only a few minutes. After the sample is taken, another ultrasound check will be done.

Some women say amniocentesis doesn't hurt at all, while others say they feel pressure or cramping. Often, people find that the waiting period for the test results is the most difficult part of the procedure.

### **How safe is amniocentesis?**

Since it was developed in the late 1960s, genetic amniocentesis has been performed on hundreds of thousands of pregnant women. Though it is now a routine procedure, it does have some risks:

- Miscarriage — while uncommon, miscarriage is the most serious risk of amniocentesis. Some miscarriages would happen anyway, but a few are caused by the procedure. The average risk for miscarriage is about one in 200 pregnancies. Women who have early amniocentesis (before 15 weeks gestation) have a somewhat higher risk of miscarriage, up to one in 100.
- Bleeding, cramping, leaking of fluid from the vagina — these symptoms, which happen to about 1 percent of women who have amniocentesis, do not usually result in a miscarriage. A woman who has these symptoms should call her doctor for advice.
- Infection — great care is taken to prevent infection, and infection following amniocentesis is very rare. However, a woman with fever or flu-like symptoms after amniocentesis should call her doctor for advice.
- Harm to the fetus — since the ultrasound picture gives the doctor exact information about the location of the fetus inside the uterus, the risk that the needle will touch the fetus is extremely low.

### **What can I expect after amniocentesis?**

Some women experience soreness or mild cramping for a day or so after the procedure. Spotting or vaginal bleeding may also occur.

We recommend that you take it easy for the next 24 hours. If you are on your feet a lot, we encourage you to take off for two days or alter your home and/or work schedule to allow you to sit. We can provide documentation of these recommendations for your employer.

If you have questions or symptoms that are of concern to you after your amniocentesis, please contact us. We particularly ask you to watch for severe cramping, increasing bleeding, leakage of clear fluid,

increasing soreness or tenderness in the abdomen and/or fever. If any of these symptoms occur, please call your obstetrician or the Maternal Fetal Care Center at **414-805-6624** and ask to speak to a nurse. If you are calling after hours, please call Labor and Delivery at **414-805-3939**.

**When will my amniocentesis test results be ready?**

Testing for chromosomal abnormalities, such as Down syndrome, usually takes 10 to 14 days. Preliminary results may sometimes be available 24 to 48 hours after the procedure. Other specialized testing such as DNA testing or biochemical analysis may take longer.

We will call you and your physician or midwife with your results and will also send them a copy of your results.

If your results are abnormal, we will discuss them with you and additional testing that may be recommended.