

## Answering Your Questions About: Amniocentesis

### What is amniocentesis?

Amniocentesis is a prenatal diagnostic test that can diagnose or rule out chromosome abnormalities such as Down syndrome and certain birth defects such as open spina bifida. This information is obtained by studying a small sample of amniotic fluid from the amniotic sac (bag of water) that surrounds the baby. The specific testing done on the fluid obtained will depend on the individual patient. Amniocentesis is usually performed between 15 and 20 weeks of pregnancy.

The following are situations that might warrant amniocentesis:

- Maternal age of 35 or older at due date
- Previous child (or close relative) with an open spine defect
- History of a previous child with a chromosomal disorder or birth defect
- Women with an increased risk of having a child with a birth defect from a screening test or ultrasound
- Personal or family history of a genetic condition for which testing is available
- Either parent is known to have a chromosome rearrangement (such as a balanced translocation)

Any woman has the option of pursuing an amniocentesis. However, many insurance companies will not cover the costs for such testing without a medical indication (like those listed above).

Of the indications listed above, maternal age is the most common reason women are offered amniocentesis. This is because the risk for having a baby with Down syndrome or other chromosome condition increases with maternal age. The following table provides the age-related risk for having a child with Down syndrome and the risk to have a baby with any chromosome problem, including Down syndrome.

Maternal Age	Down syndrome risk	All chromosome abnormalities
25	1/1,205	1/476
30	1/885	1/384
35	1/365	1/178
40	1/109	1/63
45	1/32	1/18

The accuracy of the amniocentesis in determining the presence or absence of Down syndrome and other chromosomal conditions is quite high - above 99 percent.

There are two common areas for concern for which an amniocentesis cannot help. The first is exposure to environmental substances, which may cause birth defects. This includes exposure to x-rays or to drugs. Counseling can be provided to women who have concerns in this area, but amniocentesis is generally not recommended. Ultrasound evaluation can often provide some reassurance.

Amniocentesis also cannot test for multifactorial conditions. These conditions arise because of unique genetic and environmental factors, and may include disorders such as congenital heart defects, clubfoot, congenital hip dislocation, and many cases of cleft lip and palate.

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

An ultrasound is used to locate a “pocket” of amniotic fluid that is away from the baby. The ultrasound also helps the doctor locate the safest site for inserting the needle. A specially-trained doctor inserts a thin needle through the woman’s abdominal wall and into the amniotic sac to withdraw approximately two tablespoons of fluid. The fluid is sent to a laboratory for analysis. Local anesthesia is sometimes used to numb the insertion site, but is not required.

### **What are the risks involved with amniocentesis?**

Like any medical procedure, amniocentesis does have risks. The risks to the mother’s health are relatively minor, but include pain and bruising at the site of needle insertion, vaginal bleeding and the possibility of infection. These are rare and not usually of serious consequence.

The risk of miscarriage with amniocentesis is small, approximately one out of every 200-300 procedures. Amniocentesis performed at 14 weeks gestation or less may pose a slightly higher risk (1 in 100). This risk must be weighed against the potential benefit of the information gained from the amniocentesis. The benefit for you is something only you can decide. The risk is lowest when a physician with extensive experience performs the procedure. The OSU Maternal Fetal Medicine practice has been performing amniocenteses for more than three decades.

### **When will results be available?**

Final results are available in approximately two weeks. As soon as your results are available, you or your doctor will be called. Should you wish to discuss your results further, a follow-up appointment can be scheduled.

Amniocentesis is part of the Prenatal Diagnosis Program at The Ohio State University Medical Center. For additional information, you may want to discuss amniocentesis with your physician.

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