

Amniocentesis

You are being asked to consider prenatal diagnosis in your current pregnancy. Women who are greater than twelve weeks pregnant will be considering amniocentesis. Amniocentesis is a method of prenatal diagnosis which has been performed on several hundred thousand women over a twenty year period. This brochure was developed to provide you with some basic information on this procedure.

Women who are less than 13 weeks pregnant may wish to consider pursuing a chorionic villus sampling (CVS) procedure instead of amniocentesis.

Women who are between 13 and 15 weeks pregnant may wish to consider late CVS or early amniocentesis. A separate brochure is available which describes CVS.

Who should consider amniocentesis?

Amniocentesis should generally be considered by women age 35 or older at the time of delivery, individuals who have had a child with a chromosome abnormality, individuals who have a chromosome translocation, couples at risk for a prenatally diagnosable genetic disease (e.g., hemophilia or sickle-cell disease), and individuals who have had a child or another family member with a neural tube defect (spina bifida or anencephaly).

When is amniocentesis performed?

Standard amniocentesis is performed between 15-18 weeks after a woman's last menstrual period. It can, however, be performed at later stages if necessary. A newer procedure called early amniocentesis is performed between 13-15 weeks after a woman's last menstrual period.

How is amniocentesis performed?

An ultrasound evaluation is performed to assess fetal development, to locate the placenta, to determine fetal age, and to select a pocket of amniotic fluid. A site is marked on the woman's abdomen and with ultrasound guidance, a thin needle is inserted through the abdomen and into the uterus. A small amount of amniotic fluid is removed and sent to be analyzed.

Does the procedure hurt?

Most women describe the procedure as mildly uncomfortable. Some women do experience cramping after the test. Most women are able to return to normal activities shortly after the test.

What is the risk of miscarriage after amniocentesis?

For standard amniocentesis, the most common complications are vaginal spotting or bleeding, severe cramping, or miscarriage. Infection is not very common and occurs in less than 1 in 1000 cases. The risk for complications with the procedure is 1/2% above the natural complication rate for this period in pregnancy. In early amniocentesis, the risks are still under study, but appear to be approximately the same.

What is analyzed from amniocentesis?

Cells shed from the amnion (the sac surrounding the fetus), fetal skin, and gastrointestinal tract are normally floating in the amniotic fluid. These cells can be grown (cultured) in the laboratory. Chromosome analysis is performed on these cultured cells. AFP (alpha-fetoprotein) is also present in the amniotic fluid and is measured.

What can be detected through amniocentesis?

Chromosome abnormalities (such as Down syndrome) can be detected through amniocentesis. The majority of neural tube defects are detected by elevated levels of AFP in the amniotic fluid. Some specific genetic diseases can be diagnosed by DNA or enzyme analysis if indicated. No method of prenatal testing can guarantee a baby will be born without birth defects or genetic disease. Prenatal testing can only identify the diagnosable problems for which a couple is known to be at risk. Unfortunately, there are many conditions for which prenatal diagnosis is not yet available.

How accurate are the results from amniocentesis?

Chromosome results are greater than 99% accurate. In standard amniocentesis, amniotic fluid AFP is greater than 90% accurate in detecting neural tube defects. The detection of neural tube defects by early amniocentesis has yet to have its accuracy verified. Occasionally, results need to be clarified through blood tests on the parents, ultrasound, a repeat amniocentesis, or fetal blood sampling.

How long do the results take?

Chromosome and amniotic fluid AFP results take approximately 2 weeks for completion. Special studies for other genetic diseases may take longer.

How many appointments will I have?

All women will be scheduled for a genetic counseling session to discuss the procedure in detail, determine whether additional genetic testing is appropriate, and ensure that all your concerns have been addressed. Those selecting amniocentesis are scheduled for the procedure between 15-18 weeks. These two appointments may be scheduled on the same day.

We hope this brochure answers some of your questions about amniocentesis. All the information will be discussed in greater detail during the genetic counseling session. If you have any special considerations or wish to schedule an appointment, please call. We look forward to talking with you soon.