

## Amniocentesis

A couple's decision to have prenatal diagnosis (CVS or amniocentesis) should be based on medical indications as well as a personal evaluation of the risks and benefits of the procedure. Genetic counseling can provide information about the risk of having a child with a particular disorder and can help couples decide on whether or not to have prenatal testing.

Since the 1970s, amniocentesis has been the most widely available method of prenatal diagnosis. Amniocentesis is most often performed between the 15th and 20th week of pregnancy. An ultrasound should be performed prior to the amniocentesis to examine the fetal anatomy, to confirm the gestational age, to determine the fetal number and location, and to locate the placenta.

- **Procedure**

Prior to the amniocentesis the mother's stomach is cleansed with an antiseptic solution. Under ultrasound guidance, a very thin needle is inserted through the abdominal wall and uterus into the amniotic sac (the fluid filled space surrounding the fetus) in order to obtain a few tablespoons of amniotic fluid. The developing fetus will replace this amount of fluid in approximately 6-12 hours. Most patients report feeling a pinch and some pressure and compare the level of discomfort to that of having blood drawn from the arm.

- **Results**

The amniotic fluid contains fetal cells shed from fetal skin, lungs and urinary tract, as well as chemicals that have been produced by the fetus. The cells can be used to diagnose chromosome disorders (such as Down syndrome) and the chemicals can be used to diagnose open neural tube defects (such as spina bifida). In many cases where individuals are at risk for a rare genetic disorder based on family history or ethnic background, amniocentesis can be used to determine if the fetus is affected. When specialized genetic testing is being considered, it is recommended to contact a genetic counselor in advance in order to coordinate genetic testing. However, it is important to keep in mind that a normal amniocentesis result does not rule out all birth defects or causes for mental retardation.

- **Complications**

The risk for miscarriage associated with the amniocentesis is approximately 1/300 to 1/500 (or about 0.2%- 0.3%). Complications occur in less than 1% of women after amniocentesis. These can include abdominal cramps, leakage of amniotic fluid, or spotting from the vagina. While the majority of these complications resolve, a woman who experiences any of these symptoms following amniocentesis should contact her obstetrician immediately. Any signs of infection, such as fever or chills, should also be reported promptly. Mild abdominal cramps or slight abdominal tenderness at the site of the needle puncture are normal.