

Amniocentesis

Amniocentesis is a test used for prenatal diagnosis of inherited diseases, Rh incompatibility, neural tube defects, and lung maturity. Normally performed during the second trimester of a pregnancy, this invasive procedure allows the detection of health problems in the fetus as early as fifteen weeks gestation. Although amniocentesis does carry some significant risks, the medical community commonly accepts it as a safe and useful procedure.

Rudimentary forms of trans-abdominal amniocentesis may have been practiced as early as 1877, but the birth of the modern procedure was detailed in *Nature* in 1956 by Fritz Fuchs and Povl Riis. The technology was originally used to determine the sex of the fetus but later advanced to allow the detection of hemophilia in 1960 and muscular dystrophy in 1964. Since then the procedure has been improved and expanded to test for a wide variety of inherited diseases and chromosomal abnormalities.

During amniocentesis, a thin syringe is inserted through the abdomen and into the uterus to extract fluid from the amniotic sac that surrounds the fetus. Two to four teaspoons, or approximately 1 cc of fluid per gestational week, are removed for testing. Ultrasound technology is often used to prevent injury to the fetus or mother. The fetal cells are given a few days to multiply before a karyotype, a picture of the chromosomes to identify chromosomal abnormalities, is performed and test results can be obtained. In cases where amniocentesis is used to determine the lung maturity of a fetus the procedure is performed around or after thirty-six weeks gestation to determine if early delivery is safe, and results are available within hours. Amniocentesis performed before fifteen weeks gestation is called early amniocentesis.

Although early amniocentesis still has high complication rates, both the lung maturity test and the standard amniocentesis procedure have a low likelihood of incurring risks, often as low as a 1% chance. Some possible negative side effects of performing the test include: induced miscarriage from a ruptured amniotic sac; bleeding or infection; heavy cramping; fetal injury; and Rh sensitization, or when an Rh negative mother develops antibodies to the fetus's blood due to Rh protein entering her bloodstream. Because of the potential risks, amniocentesis is only recommended for pregnant women over the age of thirty-five, women who have had previous pregnancies testing positive for chromosomal problems or neural tube defects, and women who have harmful genetic diseases in their family history.

Women who undergo this procedure will be informed of any genetic or chromosomal problems the fetus may have, and in the case of a positive diagnosis, they can use the provided information to either prepare for a child with such a disorder or seek an abortion. While amniocentesis can detect certain diseases, it is often unable to ascertain the severity of the disorders, making the woman's decision more difficult. In addition, the results of amniocentesis can come considerably later in a pregnancy than earlier diagnostic tests, such as chorionic villus sampling (CVS), which can also complicate a woman's decision.

Amniocentesis is a safe, commonplace procedure for prenatal diagnosis of genetic and chromosomal disorders. The technology has advanced significantly since its inception, and with its increased use and availability, the technique is likely to improve further still.

Sources

1. Fuchs, Fritz and Povl Riis. "Antenatal Sex Determination." *Nature* 177 (1956): 330.

2. Olney, Richard S., Cynthia A. Moore, Muin J. Koury, J. David Erickson, Larry D. Edmonds, Lorenzo D. Botto, Hani K. Atrash. "Chorionic Villus Sampling and Amniocentesis: Recommendations for Prenatal Counseling." Centers for Disease Control and Prevention. Morbidity and Mortality Weekly Report 44 (RR-9) (1995): 1-12. <http://www.cdc.gov/mmwr/preview/mmwrhtml/00038393.htm> (Accessed January 2, 2008).
3. Robertson, Audra. "Amniocentesis." MedlinePlus Medical Encyclopedia. 2006. <http://www.nlm.nih.gov/medlineplus/ency/article/003921.htm> (Accessed January 2, 2008).