Amniocentesis

Amniocentesis is a common prenatal test in which a small sample of the amniotic fluid surrounding the fetus is removed and examined. First used in 1882 to remove excess amniotic fluid, it has long been performed in late pregnancy to assess anemia in babies with Rh incompatibility and find out if the fetal lungs are mature enough for the baby to be delivered. Today, amniocentesis is often performed in the second trimester of pregnancy (usually 15-18 weeks after a woman's last menstrual period) to test for certain birth defects.

Amniocentesis is the most common prenatal test used to diagnose chromosomal and genetic birth defects. Another test, called chorionic villus sampling (CVS), can diagnose most, but not all, of the same birth defects as amniocentesis. CVS is performed earlier in pregnancy than amniocentesis (usually weeks 10-12), but poses a higher risk of miscarriage and complications.

Amniocentesis is not routinely suggested for all pregnant women because it carries a small risk of miscarriage. Amniocentesis is advised if an increased risk of chromosomal or genetic birth defects, or certain malformations, exists. Amniocentesis may be offered because of:

- **Maternal age:** The risk of bearing children with certain chromosomal birth defects increases as a woman ages. If a woman is 35 or older at the time of delivery, most physicians advise prenatal testing for chromosomal disorders. The most common of these disorders is Down syndrome, a combination of mental and physical abnormalities caused by the presence of an extra chromosome. Down syndrome occurs in approximately one in 1,250 children born to women in their 20s. The chances increase to one in 400 by age 35, and one in 100 at age 40.
- **A previous child or pregnancy with a birth defect:** If a couple already has had a child (or pregnancy) diagnosed with a chromosomal abnormality, any of a wide range of genetic birth defects, or a neural tube defect (see below), they may be advised to conduct prenatal testing during subsequent pregnancies.
- **Suggestive screening test results:** It is increasingly common to screen pregnant women by testing their blood for alpha fetoprotein (AFP) and certain other substances. A high level of AFP suggests a fetus with a neural tube defect (malformation of the spinal cord or brain, such as spina bifida or anencephaly). A low level of AFP and variations in the other substances suggest a chromosome abnormality. Amniocentesis can help detect neural tube defects by measuring AFP in amniotic fluid, and can diagnose most chromosomal abnormalities.
- **Other family history:** Couples without a previously affected child also may be advised to conduct prenatal testing if their family medical histories indicate their children may be at increased risk of inheriting a genetic disorder. Prenatal diagnosis is possible for virtually all chromosomal disorders, but not all genetic ones.

Amniocentesis usually is performed in the second trimester. Some medical centers offer early amniocentesis, done between 11-14 weeks after the last menstrual period. However, early amniocentesis is considered experimental and recent studies suggest that it is riskier than second-trimester amniocentesis (see below).

Besides determining whether a fetus's lungs are mature enough for delivery in cases in which early delivery may be necessary, several additional purposes suggest amniocentesis in the third trimester. Amniocentesis can diagnose uterine infections and may be recommended if a pregnant

woman's membranes have ruptured prematurely. The test also can determine the severity of fetal anemia in babies with Rh incompatibility and help the doctor determine whether the fetus requires lifesaving blood transfusions.

Amniocentesis is performed by inserting a thin, hollow needle into the uterus and removing some of the amniotic fluid that surrounds the baby. During the procedure, the pregnant woman lies flat on her back on a table. Her belly is cleansed with an iodine solution and the physician, using ultrasound to guide him/her, inserts a thin needle through the abdomen and uterus into the amniotic sac. S/he then withdraws about one to two tablespoons of fluid and removes the needle. After the sample is taken, the physician uses ultrasound to make sure that the fetal heartbeat is normal. The entire procedure takes a few minutes. Some women say that amniocentesis does not hurt at all; others feel cramping when the needle enters the uterus or pressure during the short time the fluid is being withdrawn.

Living cells from the fetus float in the amniotic fluid. After a sample of amniotic fluid is removed, these cells are grown in a laboratory for one to two weeks, and then tested for chromosomal abnormalities or various genetic birth defects. Test results usually are available within three weeks. Because AFP can be measured directly, without waiting for cells to grow, results of this test may take just a few days.

Millions of women have had prenatal diagnosis by amniocentesis. In 1976, after careful study, the National Institutes of Health reported that it found mid-trimester amniocentesis for prenatal diagnosis to be safe. However, amniocentesis does pose a slight risk of miscarriage. According to the Centers for Disease Control and Prevention, the rate of miscarriage is between one in 400 and one in 200 procedures. The procedure also carries an extremely low risk of uterine infection (less than one in 1,000), which can cause miscarriage. Studies suggest that the risk of miscarriage following first-trimester amniocentesis may be three times higher than the risk after second-trimester amniocentesis.

More than 95%of the high-risk women who have prenatal diagnosis receive reassuring news that their unborn babies do not have the disorders for which they are tested. However, no one prenatal test can guarantee the birth of a healthy baby, since only some birth defects can be ruled out before birth. Three to four out of every 100 babies have a birth defect. Amniocentesis has an accuracy rate of between 99.4 and 100% in diagnosing chromosomal abnormalities.

Your tasks for this activity are:

- Review the vocabulary handout to familiarize yourself with any unknown vocabulary words used in this description of amniocentesis.
- Decide how to communicate this information to your class in an effective way. You may include all information or only the relevant highlights from what you learned in this handout.
- In your presentation, communicate the advantages and disadvantages of this technology. You will want to discuss this as a group. Mention your suggestions for further improvements to the technology.
- Your presentation must include at least one visual element (more than just reading / lecturing to the class), such as a drawing, graphic, picture, table, graph, chart, skit, song, poem, demo.

Source: Amniocentesis, Medical References, Quick References: Fact Sheets. Updated August 2005. March of Dimes Foundation. Accessed February 17, 2009. http://www.marchofdimes.com/professionals/14332_1164.asp