Genetic Amniocentesis Information

Preferred Women's Healthcare

Amniocentesis is performed during pregnancy to help determine an inherited, genetic abnormalities that a fetus may have. This procedure involves the careful passage of a thin needle through the abdomen of the mother and into the sac of amniotic fluid surrounding the fetus. A small amount of the fluid is then withdrawn for analysis.

Who Should Consider It?

The principle reasons for genetic amniocentesis are:

a) Maternal age 35 years or older (at time of anticipated delivery)
b) Previous child with a proven chromosomal abnormality

Other more rare reasons, such as testing for unusual enzyme deficiencies, neural tube defects, sickle cell disease and so forth, often require additional counseling with a geneticist and are discussed in the enclosed booklet.

What Is Genetic Amniocentesis?

Genetic Amniocentesis 15:

a) Usually performed on an outpatient basis;
b) Able to detect fetal sex and almost all chromosomal abnormalities;
c) Able to detect 85-90% of neural tube (brain and spine) defects in a sample of amniotic fluid by testing for the level of alpha-fetoprotein.

What Are Its Limitations?

The limitations of the procedure are that:

a) It cannot detect nonchromosomal congenital abnormalities such as cleft palate, intestinal or cardiac abnormalities, etc., which occur in approximately 3% of pregnancies;
b) Its accuracy exceeds 99.9% but cannot be totally guaranteed. There is a very remote possibility of error (one in ten thousand cases) in the interpretation of fetal cells grown from amniotic fluid;
c) Fluid cannot be obtained on rare occasions due to technical difficulties or decreased amounts of fluid. The attempt must be postponed, if this occurs;
d) A second amniocentesis (7-14 days later) is needed on rare occasions because cells in the initial sample may not have grown enough to permit valid interpretation.

What Are The Risks?

The principle risk of amniocentesis is spontaneous abortion (miscarriage), which occurs in one of every 200-250 amniocenteses performed (0.05%). Additional risks include, but are not limited to, possible fetal or maternal injury, death, infection, or bleeding. Because there is risk involved, it is important to note that this test is only recommended when there is a specific reason to believe that a detectable genetic disease may be present. The occurrence of these complications in our practice is unusual for several reasons. Most importantly, the physicians who perform this procedure are very experienced practitioners. Secondly, ultrasound is utilized to help determine the gestational age of the fetus, the position of the placenta, and the best area for the proper and safe placement of the amniocentesis needle.

ON THE DAY OF YOUR AMNIOCENTESIS

1. Sign in at the reception desk of Preferred Women's Healthcare at your appointed time. Your ultrasound examination and amniocentesis procedure will be performed in the testing area of our office.

2. Eat a light breakfast before coming for the test; the baby is usually less active after you have eaten. **One hour before your appointment, drink three glasses (about eight ounces each) of liquids to help fill your bladder.** Do not go to the bathroom because a full bladder helps make the procedure easy.

3. Before your appointment, please study the information sent to you. We will review this material briefly with you. Any questions you may have will be answered after our discussion. **Please fill in the "Genetic History Questionnaire."**

4. You will need to **read the informed consent forms for the amniocentesis procedure and the "consent for Chromosomal Analysis."** Please do not sign these forms before reporting to Preferred Women's Healthcare.

5. An ultrasound examination (also called sonography) will be done to determine the size and position of the baby and placenta, as well as to locate the safest place for the insertion of the amniocentesis needle.

6. After the doctor determines the best area to perform the amniocentesis using the ultrasound equipment, your skin will be cleansed with a special soap. A local anesthetic is usually used prior to the amniocentesis. Then a thin needle is inserted through your abdomen into your uterus. A small amount of fluid is withdrawn (less than an ounce) and will be replaced by your system within several hours.

7. The doctor uses ultrasound to guide the needle into position. As the fluid is withdrawn, you might feel a pressure in your abdomen. Most women who have had an amniocentesis state that it is like getting a shot and is not painful. You might feel some mild cramping or mild pressure after the procedure.

8. The tiny hole made by the needle should seal naturally soon after the needle is withdrawn.

9. After the amniocentesis procedure is completed, we will look again at the baby to make sure that all appears well. After your abdomen is cleansed, you may go to the bathroom. If Rh immune globulin (Rhogam) is to be given to you because you are Rh-, the father is Rh+, and your antibedy blood test is negative, that injection is administered prior to your departure. (We will bill you separately for the **laboratory fee and Rh Immune Globulin** Injection.)

10. On rare occasions, adverse side effects occur. Any of the following should be reported to your obstetrician:
    1) Leaking of clear, watery fluid from your vagina
    2) Bleeding from your vagina
    3) The onset of contractions that appear to be occurring at regular Intervals and Increasing In Intensity
    4) Fever

Revised 4/99