GENETICS LABORATORIES GENETICS LABORATORIES

You are being asked to consider prenatal diagnosis in your current pregnancy. Amniocentesis is typically offered to women who are greater than 15 weeks pregnant. Amniocentesis is a method of prenatal diagnosis that has been performed for over 30 years. 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. 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 with a child or other close family member with a neural tube defect (spina bifida or anencephaly). Amniocentesis may also be considered when certain abnormalities are seen on ultrasound or when maternal serum screening has identified an increased risk for certain chromosome or fetal anomalies.

When Is Amniocentesis Performed?

An amniocentesis is typically performed between 15-20 weeks after a woman's last menstrual period. It can, however, be performed at later stages if necessary.

How Is Amniocentesis Performed?

An ultrasound evaluation is performed to assess fetal development, locate the placenta, determine fetal age, and select a pocket of amniotic fluid. A site is identified 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 the laboratory for analysis.

Does The Procedure Hurt?

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

What Are The Risks After Amniocentesis?

For standard amniocentesis, complications can include vaginal spotting or bleeding, severe cramping, or miscarriage. Maternal infection is rare. Traditionally the risk of amniocentesis was quoted to be approximately 1 in 200 but based on new data the risk is currently believed to be lower.

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 can be detected by elevated levels of AFP in the amniotic fluid. Some specific genetic diseases can be diagnosed by DNA or enzyme analysis if indicated. New testing called prenatal chromosomal microarray analysis (CMA) allows for detection of additional chromosome abnormalities not detectable through standard chromosome analysis. See the prenatal CMA brochure for more details about this testing.

No method of prenatal testing can guarantee a baby will be born without birth defects or genetic disease. It is also important to remember that 2-3% of all children are born with a birth defect regardless of whether or not a woman had prenatal testing during her pregnancy. 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. Amniotic fluid AFP is greater than 99% accurate in detecting open neural tube defects. 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 are available within two weeks. Special studies for other genetic diseases may take longer.

How Do I Learn More?

We hope this brochure answers some of your questions about amniocentesis. All the information can be discussed in greater detail during a genetic counseling session, thus it is helpful to schedule an appointment for genetic counseling prior to the amniocentesis procedure. The genetic counselor/healthcare provider will not only discuss the procedure in greater detail, but will also determine whether additional testing is appropriate and ensure that all of your concerns are addressed.

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