

## AMNIOCENTESIS

### WHAT IS AMNIOCENTESIS?

Amniocentesis is a procedure that samples a small amount of the amniotic fluid surrounding the developing baby (fetus). Analysis of this fluid allows certain genetic conditions and birth defects to be detected. Amniocentesis is usually done after the 15th week of pregnancy.

### WHO IS OFFERED AN AMNIOCENTESIS?

Two of the most common reasons why women are offered amniocentesis are due to maternal age and abnormal screening results. All women 35 or older at the time of delivery are offered prenatal diagnosis by either CVS or amniocentesis due to the increased chance for the fetus to have a chromosome condition such as Down syndrome. Chromosomes are the packages in our cells that contain our genetic instructions. Most people have 23 pairs of chromosomes or 46 in all. Extra or missing chromosome material can cause birth defects or developmental delays. For example, an extra copy of the 21<sup>st</sup> chromosome causes the learning problems and common facial features associated with Down syndrome.

Women who have a positive first or second trimester screening test are also offered amniocentesis. There are several blood tests that screen pregnancies to determine if the chance for having a baby with a chromosome condition or neural tube defect (an opening in the spine or skull) may be increased. There may be findings on ultrasound in either the first or second trimester of pregnancy that may also increase the chance for one of these conditions. When a screen shows an increased chance, or is screen positive, for one of these conditions amniocentesis is often recommended to confirm or rule out the diagnosis.

Amniocentesis may also be offered if a couple has had a previous child with a genetic condition, a family history of neural tube defect, or if the family history suggests an increased risk for a genetic condition for which prenatal testing by amniocentesis is available.

### HOW IS AMNIOCENTESIS DONE?

Ultrasound is used to locate the fetus and the placenta so the physician can choose the safest spot for insertion of a needle into the uterus (womb) and fluid bag (amniotic sac). The abdomen is then cleansed using an antiseptic. The physician inserts a thin needle through the abdomen (not the navel) into the womb and 2-3 tablespoons of amniotic fluid are withdrawn. The procedure takes only a couple of moments. After the sample is taken, the physician uses ultrasound to check on the fetal heartbeat.

The sample obtained from amniocentesis is then analyzed in a laboratory. The number and size of the chromosomes are studied to determine if a genetic condition is present. It typically takes 10-14 days to get the results from this test. If specific DNA or biochemical testing is being offered based on the family history, the amount of time to obtain the result will depend on the test ordered.

Amniotic fluid contains a protein produced by the fetus called alpha-fetoprotein (AFP). Small amounts of AFP normally pass into the amniotic fluid. Too much AFP in the fluid can be a sign of a neural tube defect (an opening of the spine or skull). In pregnancies known to be at an increased risk for neural tube defects, another chemical produced by the fetus called acetylcholinesterase is measured.

Some women say amniocentesis doesn't hurt at all, while others feel cramping when the needle enters the uterus. It is recommended that a woman avoid heavy lifting or aerobic activity for 24 hours after the procedure.

## AMNIOCENTESIS

### **ARE THE RESULTS OF AMNIOCENTESIS ACCURATE?**

In over 99.7% of cases, the results allow for a clear answer about the fetus's chromosomes. In rare cases, there may be a small chromosome difference that is not detected.

The amniotic fluid alpha-fetoprotein (AFP) is 95% accurate in detecting an open neural tube defect. This test is not as accurate in detecting closed neural tube defects. Acetylcholinesterase testing is 99% accurate in detecting open neural tube defects.

A normal amniocentesis test result can be reassuring about conditions we know the pregnancy is at risk for, but cannot ensure the birth of a healthy baby. Approximately 3 - 5 out of every 100 babies are born with a birth defect, and only some birth defects can be detected by amniocentesis.

### **IS AMNIOCENTESIS SAFE?**

Amniocentesis has been used for over 40 years, with the majority of women having good outcomes. However, amniocentesis does pose a slight risk of discomfort, cramping, bleeding or leaking of amniotic fluid and infection. Following amniocentesis, fewer than 1 in 300 (0.3%) women will have significant procedure-related complications that lead to a loss of the pregnancy.

### **IS AMNIOCENTESIS THE RIGHT TEST FOR ME?**

The decision to have prenatal diagnosis is a personal one. A woman should think about the benefit of knowing the chromosome information versus the risk of the procedure. It may be helpful to discuss the risks and benefits with your husband/partner, family, physician, and/or genetic counselor. If you have questions about amniocentesis or genetic counseling, an appointment for genetic counseling can be made through your physician.

**Department of ObGyn**

**USC Genetic Counseling**

Two Medical Park Rd, Suite 103

P (803) 545-5775

F (803) 434-4596