



## **AMNIOCENTESIS**

Your physician may offer or recommend prenatal testing using a procedure called amniocentesis. Amniocentesis is typically performed between 15-20 weeks of pregnancy, and involves a small amount of amniotic fluid being withdrawn from the uterus, which will then be tested for chromosome abnormalities. In a small percentage of cases, amniocentesis may cause pregnancy complications, and in rare cases, miscarriage.

The decision to have prenatal genetic diagnosis is an important one; only you can decide if amniocentesis is right for you. You and your partner will have the opportunity to ask questions during your genetics consultation to better understand the risks and benefits of pursuing amniocentesis. The information provided below will acquaint you with our procedures and policies.

### **WHAT TO DO PRIOR TO YOUR VISIT**

You may have a number of concerns regarding amniocentesis and its risks. We suggest you write down your questions. We will be sure to address them during your genetics consultation.

### **FAMILY HISTORY**

It would be helpful if you and your partner review your family histories. It is important to identify any relatives who have inherited conditions, birth defects, chromosomal conditions such as Down syndrome, or who have had any stillbirths or more than two miscarriages. In some cases, medical records on the affected individual may be required in order to provide accurate information about recurrence risks. If you have a specific concern about a genetic condition in your family history and would like to discuss options for prenatal testing for the condition, it is recommended that you schedule a separate genetics consultation prior to the day of your amniocentesis procedure.

### **Rh BLOOD TYPE**

If you decide to have amniocentesis, it is important that your blood group and Rh type be documented prior to your appointment. A RhoGam injection must be administered to all Rh negative women to prevent the possibility of Rh sensitization. Please ask your physician to FAX documentation of your blood type. A blood donor card is also sufficient. If both you and your partner are Rh negative, you will not need to receive RhoGam; however, documentation of his Rh type is required. If documentation of your blood type is not received, or if you are Rh negative and will be receiving RhoGam, please allow for extra time on the day of your appointment.

There are no other specific instructions to prepare for your visit. A light breakfast is recommended. You will *not* need a full bladder for the test.

### **WHAT TO DO ON THE DAY OF YOUR APPOINTMENT**

**It is imperative that you arrive at your appointed time. Please plan to spend a total of 2-3 hours completing all portions of your appointment.**

You will first have a genetics consultation at Dr. Pergament's office, located at 680 North Lake Shore Drive, Suite 1230. This appointment is to discuss your family histories, the risks for chromosome abnormalities, and to address your questions and concerns. Although every effort is made to stay on schedule, there are times when a delay is unavoidable. Your patience and understanding is appreciated if any delay occurs.

Following your consultation at Dr. Pergament's office, you will be directed to the Northwestern Center for Genetic Medicine for the procedure of amniocentesis. Your amniocentesis will be performed by Dr. Lee Shulman or Dr. Jeffrey Dungan.

## **THE AMNIOCENTESIS PROCEDURE**

Amniocentesis involves a small amount of amniotic fluid being withdrawn from the uterus. This sample will be cultured and analyzed for chromosome number and structure, and in the case of prenatal testing for a hereditary genetic condition, the presence of the gene mutation previously identified in the family.

Amniocentesis is performed under ultrasound guidance following a thorough ultrasound evaluation. First, you will be asked to lie down on an examination table for an ultrasound to determine your gestational age and the location of the placenta and fetus. The physician will then identify a pocket of amniotic fluid that is furthest from the fetus, and a slender, sterile needle will be inserted into the amniotic sac. A small sample (usually less than two tablespoons) of amniotic fluid will be removed. This fluid contains fetal skin cells which will be analyzed by our laboratory.

The entire process will take approximately 30 minutes; the amniocentesis itself takes less than one minute. Your partner may accompany you for all portions of your appointment, including your genetics consultation, ultrasound evaluation, and amniocentesis procedure.

## **WHAT TO DO FOLLOWING YOUR AMNIOCENTESIS**

A recovery period following the procedure is not necessary. You should be able to resume normal activities, but we suggest you refrain from heavy lifting, strenuous exercise (such as jogging, aerobics, and tennis), and sexual activity for 24-48 hours following the amniocentesis. You may experience abdominal tenderness, mild uterine cramping similar to menstrual cramps, or some slight fluid leakage. If you are experiencing any discomfort or other symptoms, please contact your obstetrician's office or your obstetrician's emergency line if it is after hours. Please inform us of any symptoms or complications as well.

## **WHEN AND HOW RESULTS ARE PROVIDED**

FISH results are usually available on the same day of the procedure, and provide a result for chromosomes 13, 18, 21, X, and Y. You will be called with this result as soon as it is available. The full chromosome report and amniotic fluid alpha fetoprotein results are usually available approximately 2 weeks after the procedure. You will be called as soon as the final studies are complete. A written report will also be sent to both you and your physician.

## **QUESTIONS?**

If you have any questions concerning the above information, please do not hesitate to contact us at 312-981-4400.