SECOND TRIMESTER GENETIC AMNIOCENTESIS
Patient information

AMNIOCENTESIS

Amniocentesis is a procedure that involves the withdrawal of a small amount of amniotic fluid. Fetal cells in the fluid can be analyzed to determine the chromosomal status of the fetus and for the detection of open neural tube defects. The risk of having a baby with a chromosomal abnormality, such as Down Syndrome, increases with a woman's age. The amniocentesis can detect Down Syndrome, or Trisomy 21, as well as other less common chromosomal abnormalities. A small part of the sample obtained is also tested for open neural tube defects, such as spina bifida and anencephaly. The risk for neural tube defects is approximately 1/1000 and is not related to maternal age.

In special circumstances, amniocentesis can also be used to determine the presence or absence of a specific genetic disorder in the fetus. Please notify our office as soon as possible if special testing is required for your pregnancy.

THE PROCEDURE

Amniocentesis is typically performed between 15 – 16 weeks of pregnancy, counting from the first day of the last menstrual period, or as determined by earlier ultrasound examination. During the procedure, a small amount of amniotic fluid is withdrawn from the sac surround the baby. And ultrasound is performed before and during the amniocentesis. You will lie down with your abdomen exposed for the procedure. The procedure is performed by inserting a slender needle through the abdomen into the uterus. Amniotic fluid will be removed by applying gentle suction to a syringe attached to the needle. The procedure is typically performed in a minute or two. Including the ultrasound examination prior to the procedure, you can expect to spend approximately 20 – 30 minutes in the procedure room. During the appointment we will discuss with you any restrictions following the procedure and any follow up recommendations.

RESULTS

The final results of the chromosome analysis are typically available in approximately two weeks, although you have the option of obtaining preliminary F.I.S.H results in 2 – 3 business days. We will review this option with you during your appointment. In order to obtain the final results, the chromosomes within the baby's cells are studied under a microscope. The number and general shape of the chromosomes are checked. An occasional error has been reported, but the accuracy of amniocentesis for chromosome analysis is greater than 99%.

Rarely, we obtain results which will not allow us to establish with certainty whether or not the fetal chromosomes are normal. These situations are resolved on an individual basis and may require further testing such as examination of the parent's chromosomes. Very rarely, an insufficient amount of amniotic fluid is obtained when the amniocentesis is attempted. In that event, another amniocentesis may be recommended.

For families at risk for a specific genetic disorder, the timing of the test results will vary depending upon the laboratory used.

We will contact you by telephone as soon as the results are available. In the event of an abnormal result, we will contact you maternity physician or midwife first. In this case, your maternity physician or midwife will contact you to discuss the test results. They may suggest referral to a genetics counselor or medical geneticist in order to discuss and review the results.

RISKS

Although amniocentesis in the second trimester is a fairly safe procedure, there is some risk involved. Side effects may include the following:

- Cramping
- Bleeding
- Infection
- Leakage of fluid
- **Miscarriage**

Unfortunately, all pregnancies have a risk for miscarriage – whether a test is performed or not. In the second trimester (when an amniocentesis is typically performed), the normal risk for miscarriage generally believed to be between 2 – 3%. The risk for miscarriage is slightly increased with amniocentesis. In our experience, the risk for amniocentesis-related pregnancy loss is 1 in 250 to 1 in 300.

There is a very small risk of infection in your uterus. We will be taking precautions against this, but if an infection should occur, it would require treatment with antibiotics. To date, there is not known to be an increased risk for birth defects, fetal injury, or suboptimal growth in babies following amniocentesis. We are monitoring the outcomes of all pregnancies in which amniocentesis has been preformed to look for unexpected problems from the testing. You will receive a letter after the delivery of your baby, requesting information regarding your pregnancy outcome.

An ultrasound will be performed before and during the amniocentesis. This technique uses echo patterns from sound waves in order to localize the placenta, fetal sac, and to look for the possibility of more than one fetus. Extensive experience in the use of ultrasound during pregnancy is available. To date there is no indication of ultrasound causing harm to the fetus.

With prenatal diagnosis, we attempt to rule out certain diseases in the baby. We are not able to state that the baby is healthy in every way. Therefore, if the amniocentesis test results are normal, it gives a high assurance that the baby does not have a chromosomal disorder. However, it does not guarantee a healthy baby. Approximately 35% of babies are born with a birth defect or inherited disease, such as deafness, blindness, cleft lip or palate to name a

few. Many of these birth defects are undetectable via prenatal diagnosis or ultrasound.

PREPARATION FOR THE PROCEDURE

A full bladder is required in order to perform the amniocentesis. You should drink three 8 ounce glasses of any beverage (preferably water), beginning one hour before your scheduled appointment.

It is imperative that we know your blood type and Rh type by the time of the appointment. If you have not had your blood type analyzed, please do so prior to your appointment. We will contact your doctor's office to obtain this information prior to your appointment.

If you are Rh negative, amniocentesis will only be performed if you are not sensitized. Your maternity care provider should perform this test as part of your routine prenatal blood work. All Rh negative women will receive RhoGam (Rh immune globulin) after the procedure.

Genetic counseling can be provided on a separate day before the amniocentesis is performed. Please contact our office if you wish to schedule this appointment.

We encourage your partner to accompany you for all or part of the appointment, including the ultrasound and amniocentesis. Children are not allowed in the for the amniocentesis procedure.

POST PROCEDURE INSTRUCTIONS

Some women may experience some normal side effects, such as mild contractions, cramping or spotting after the amniocentesis.

We recommend that you avoid the following activities for three days following the procedure:

- Exercise – We ask that you refrain from jogging, aerobics, tennis and other related activities.
- Sexual Intercourse should be avoided for three days after the procedure or until any spotting ends to reduce the risk of intrauterine infection.
- Lifting anything over 20 pounds, including children.

Please contact Dr. Cooper and your maternity care provider, as soon as possible, if you have any symptoms such as bleeding, fluid leakage, general malaise, muscle aches or a temperature.