AMNIOCENTESIS

WHAT IS AMNIOCENTESIS?

It is an invasive obstetric procedure in which a small amount of fluid is removed from the amniotic sac for genetic and/or laboratory analysis.

REASONS FOR THE PROCEDURE

Out of 100 newborns, about two or three have major birth defects. Birth defects can be genetic, occurring in genes or chromosomes, or caused by exposure to harmful agents. Laboratory examination of amniotic fluid helps diagnose certain abnormalities of the unborn child. A typical genetic amniocentesis result gives information about the number of chromosomes (as an example, there is an extra chromosome in Down syndrome and one missing in Turner syndrome) and the sex of the baby. It does not show whether the baby has other inherited or genetic disease unless the lab is told to test for a specific disease because of a family or obstetrical history, which places the baby at risk. However, a normal test result does not guarantee that the baby will be normal. Amniocentesis cannot identify all birth defects, including heart defects, clubfoot or cleft lip and palate.

The best time to do amniocentesis is between the 15th and 18th weeks of pregnancy. There is ample fluid for the testing, and time to treat certain problems before the baby is born, and enough time exists if the parents decide not to continue with the pregnancy. Amniocentesis is often done for one or more of the following reasons:

- Maternal age is over 35 years.
- Patient had abnormal results from blood screening test, such as maternal serum screening (alpha feto-protein alone, double, triple or quad screening test).
- Risk of metabolic disease or neural tube defect (because of previous experience or family history).
- Either one or both parent has a chromosome abnormality.
- Mother has previously had a child with a chromosome abnormality, such as Down syndrome.
- Mother carries a sex-linked abnormality, and the sex of the unborn child must be determined.

HOW ACCURATE ARE THE RESULTS?

No test is absolutely perfect, but the chromosome test for Down syndrome is very reliable. The test will also pick up other changes in the chromosomes. However, very small abnormalities of the chromosomes may be not be seen but these are very rare. If you are having the amniocentesis for other genetic disorders, you should discuss the accuracy of the test with your genetic doctor. It is reassuring to note that more than 95% of amniocentesis tests indicate no abnormalities.

DESCRIPTION OF PROCEDURE

- No specific prior preparation is required. There is no need to fast before the procedure.
- An ultrasound scan of the baby and the amniotic sac is done prior to the procedure. This helps to pinpoint the exact location for the needle entry.
- The abdominal skin area will be cleaned with an antiseptic.
- A local anesthetic is injected into the abdomen at the site of needle entry.
- A hollow and very fine needle is inserted through the abdominal wall into the uterus. The needle will cause temporary pain, but should not hurt more than any injection. Some women report mild to moderate cramping, or a feeling of pressure, during the procedure.
- Amniocentesis is usually performed using continuous ultrasound to allow constant view of the needle's path and avoid injuring the baby.
- A small amount of amniotic fluid is withdrawn through the needle. The needle is removed, and the fluid is sent to the laboratory for analysis.
- The amniotic fluids contain cells from the baby and are grown in a special culture. Next, the chromosomes in these cells are studied for abnormalities. This shows if there is an extra chromosome (as in Down syndrome) or other chromosomal defects.

- Results of the genetic analysis of the amniotic fluid will take 14 to 16 days. However an early result for primary trisomies 13, 18 and 21 can be done if specially requested and the results will be available in approximately 24 to 72 hours.

POSTPROCEDURE CARE

- Arrange to have someone drive you home following the procedure.
- Bathe and shower as usual. You may wash the injection site gently with mild, unscented soap.
- You may experience some mild lower abdominal cramping for a day or two. Take paracetamol for the discomfort or pain if necessary.
- Rest for 4 to 6 hours following the procedure. There are usually no further restrictions on your normal activities.

POSSIBLE COMPLICATIONS

- Miscarriage triggered by procedure (rare) – the risk is about 0.5%.
- Damage to the baby (uncommon), such as slight dimpling of the skin where the baby was poked during the procedure and, very rarely, death.
- Infection.
- Vaginal bleeding
- Leakage of amniotic fluid.

SEE YOUR DOCTOR IMMEDIATELY IF THERE IS:

- Significant loss of fluid from the vagina.
- Nausea and vomiting.
- Pain in the lower abdomen or shoulder.
- Bleeding from the vagina or puncture site.
- Signs of infection, including headache, muscle aches, dizziness or a general ill feeling and fever.

Disclaimer

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