

Amniocentesis

Definition

Amniocentesis is a procedure used to diagnose fetal defects in the early second trimester of pregnancy. A sample of the amniotic fluid, which surrounds a fetus in the womb, is collected through a pregnant woman's abdomen using a needle and syringe.

Tests performed on fetal cells found in the sample can reveal the presence of many types of genetic disorders, thus allowing doctors and prospective parents to make important decisions about early treatment and intervention.

Purpose

Amniocentesis has been used routinely to test for Down syndrome, by far the most common, nonhereditary, genetic birth defect, afflicting about one in every 1,000 babies. By 1997, approximately 800 different diagnostic tests were available, most of them for hereditary genetic disorders such as Tay-Sachs disease, sickle cell anemia, hemophilia, muscular dystrophy, and cystic fibrosis .

Amniocentesis, often called amnio, is recommended for women who will be older than 35 on their due-date. It is also recommended for women who have already borne children with birth defects, or when either of the parents has a family history of a birth defect for which a diagnostic test is available.

Another reason for the procedure is to confirm indications of Down syndrome and certain other defects which may have shown up previously during routine maternal blood screening.

The risk of bearing a child with a nonhereditary genetic defect such as Down syndrome is directly related to a woman's age- the older the woman, the greater the risk. Thirty-five is the recommended age to begin amnio testing because that is the age at which the risk of carrying a fetus with such a defect increases the risk of miscarriage.

One of the most common reasons for performing amniocentesis is an abnormal alpha-fetoprotein (AFP) test. Alpha-fetoprotein is a protein produced by the fetus and present in the mother's blood. A simple blood screening, usually conducted around the 15th week of pregnancy, can determine the AFP levels in the mother's blood.

Amniocentesis is generally performed during the 16th week of pregnancy, with results usually available within three weeks. It is possible to perform an amnio as early as the 11th week, but this is not usually recommended because there appears to be an increased risk of miscarriage when done at this time.

The advantage of early amnio and speedy results lies in the extra time for decision making if a problem is detected.

Description

The word amniocentesis literally means "puncture of the amnion," the thin-walled sac of fluid in which a developing fetus is suspended during pregnancy.

During the sampling procedure, the obstetrician inserts a very fine needle through the woman's abdomen into the uterus and amniotic sac and withdraws approximately one ounce of amniotic fluid for testing. The relatively painless procedure is performed on an outpatient basis, sometimes using local anesthesia.

The physician uses ultrasound images to guide needle placement and collect the sample, thereby minimizing the risk of fetal injury and the need for repeated needle insertions.

Once the sample is collected, the woman can return home after a brief observation period. She may be instructed to rest for the first 24 hours and to avoid heavy lifting for two days.

The sample of amniotic fluid is sent to a laboratory where fetal cells contained in the fluid are isolated and grown in order to provide enough genetic material for testing. This takes about seven to 14 days.

An alternative to amnio, now in general use, is chorionic villus sampling(CVS) which can be performed as early as the eighth week of pregnancy.

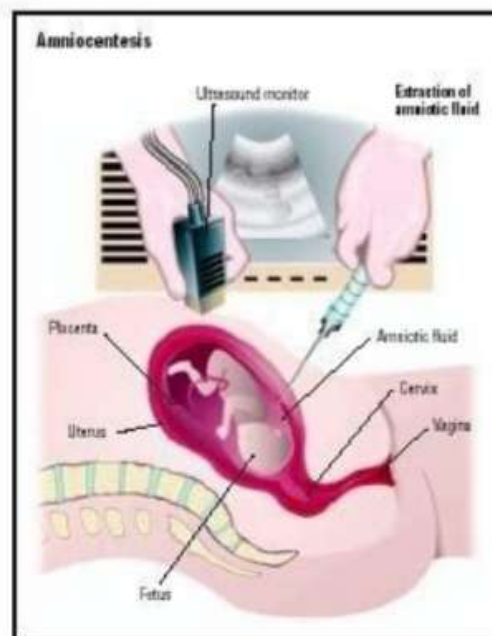
Preparation

It is important for a woman to fully understand the procedure and to feel confident in the obstetrician performing it. The patient should feel free to ask questions and seek emotional support before, during and after the amnio is performed.

Aftercare

Necessary aftercare falls into two categories: physical and emotional.

Physical Aftercare: During and immediately following the sampling procedure, a woman may experience dizziness, nausea, a rapid heartbeat, and cramping. Once past these immediate hurdles, the physician will send the woman home with instructions to rest and to report any complications requiring immediate treatment, including:



To perform amniocentesis, a physician uses an ultrasound monitor to visualize the fetus while inserting a syringe to extract amniotic fluid for analysis.

- Vaginal bleeding. The appearance of blood could signal a problem.
- Premature labor. Unusual abdominal pain and/or cramping may indicate the onset of premature labor. Mild cramping for the first day or two following the procedure is normal.
- Signs of infection. Leaking of amniotic fluid or unusual vaginal discharge, and fever could signal the onset of infection.

Emotional Aftercare:

Once the procedure has been safely completed, the anxiety of waiting for the test results can prove to be the worst part of the process. A woman should seek and receive emotional support from family and friends, as well as from her obstetrician and family doctor.

Procedure:

The whole procedure takes about 45 minutes. First, Pregnant women must have a full bladder before beginning, which can be difficult considering that it sometimes, feels like the baby is already dancing on your bladder. Despite the discomfort, this step is important in order to better visualize the baby during the procedure.

Then, women are asked to lie on her back while an ultrasound is done so that the doctor can see the baby on a screen. Oftentimes, a numbing cream or injection is placed on the belly where the procedure will occur. The doctor will also wash her belly with an antimicrobial liquid to prevent any infections.

Finally, a long, thin needle is inserted into the belly and into the uterus (womb) where a sample of amniotic fluid is collected (about two to three teaspoons).

The amniotic fluid is the liquid that surrounds the baby in the womb; it contains cells from the fetus. This fluid sample is sent for testing, and the results take about two weeks to return. Most women discover that their fetus is normal. The mother usually waits another 20-30 minutes after the procedure before going home, to ensure both mom and babies are okay.

Risks:

- Maternal/fetal hemorrhage.
- Infection.
- Fetal injury.
- Miscarriage.

Result:

Negative results from an amnio analysis indicate that everything about the fetus appears normal and the pregnancy can continue without undue concern. A negative result for Down syndrome means that it is 99% certain that the disease does not exist.

Positive results on an amnio analysis indicate the presence of the fetal defect being tested for, with an accuracy approaching 100%. Prospective parents are then faced with emotionally and ethically difficult choices regarding treatment options, the prospect of dealing with a severely affected newborn, and the option of elective abortion.