

An experienced physician may perform the amniocentesis using continuous ultrasound guidance to carefully insert a slender needle through the mother's abdominal wall into the uterus at a safe distance from the fetus. A small amount of amniotic fluid containing cells naturally washed from the fetus is quickly withdrawn.

Please do not empty your bladder during the hour before the ultrasound exam. You need a full bladder for this exam. Drink only regular amounts of fluid (without caffeine). Please wear clothing over your abdomen which is easy to remove for the exam.

IS AMNIOCENTESIS SAFE?

Amniocentesis has been proven to be a very safe procedure for both mother and baby. We cannot say amniocentesis is without risk. You should know miscarriages occur spontaneously before and after amniocentesis, independent of the procedure. Medical studies have shown there does not appear to be a statistically increased risk of miscarriage for women having amniocentesis compared to the risk for women who have not had amniocentesis. The possible risk of a spontaneous abortion, is therefore, stated as being equal to or less than 0.5% (1 in 200 chances). The observed number of spontaneous abortions occurring after amniocentesis by an experienced physician using continuous ultrasound has been shown to be less than 1 in 300.

HOW ARE THE TESTS RESULTS REPORTED?

The prenatal chromosome and open spine and skull tests will be completed in less than 10 days. Some special genetic tests take one to three more weeks.

Please inform your physician and counselor how to reach you so your tests can be quickly reported.

Most parents are reassured by these tests because most tests will be normal. If the test shows your fetus has a disorder, the nature of the disorder will be discussed with you. Prenatal diagnosis allows couples to make informed decisions and helps with the management of their pregnancy. Supportive genetic counseling is available.

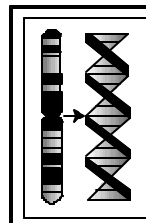
Please discuss any questions you may have with your physician and counselor.

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Amniocentesis

Parents are naturally concerned about the health of their unborn babies (fetuses). Although most women give birth to healthy babies, certain women have a higher-than-average chance of giving birth to a baby with a birth defect. Doctors and genetic counselors rely on personal and family medical histories, physical exams, and various procedures and tests to help identify women who may have a baby with birth defects. Some of these women can benefit from amniocentesis.

Amniocentesis is a medical procedure in which a small amount of amniotic fluid is withdrawn from the amniotic sac surrounding the fetus in the uterus.

WHO SHOULD BE OFFERED AMNIOCENTESIS?

You and your doctor can decide whether you will benefit from the information provided by amniocentesis. Each woman's case is different. Amniocentesis may be recommended for:

- ? Women who are pregnant or planning to become pregnant and will be 35 or older at delivery.
- ? Couples who have had baby or a relative with a chromosome abnormality like Down syndrome, an open spine or skull defect, mental retardation, or other birth defects.
- ? Couples who are known or possible carriers of an inherited disorder like Hemophilia, Muscular Dystrophy, Cystic Fibrosis, Tay Sachs, Thalassemia.
- ? Couples with an increased risk of having a baby with an open spine/open skull or Down syndrome based on pregnancy screening or ultrasound.
- ? Couples who are concerned about having an abnormal baby.

WHAT IS A CHROMOSOME ABNORMALITY?

Chromosomes are units of genetic information. Babies with a chromosome abnormality usually have mild to severe mental retardation and birth defects. The most common chromosome abnormality is Down syndrome. A chromosome test is performed on cells shed from the fetus into the amniotic fluid to determine if the fetus has a chromosome abnormality. The test can also reveal the chromosome sex of the fetus. You may choose whether or not you want to know the sex of your baby.

Chromosome abnormalities are caused at conception when an abnormal sperm or egg from one parent fuses with a normal sperm or egg from the other parent. The abnormal sperm or egg contains extra or missing chromosome material. These abnormal sperm or eggs appear to be present in everyone, but the risk of an abnormal conception increases significantly with the parents' ages.

WHICH TESTS CAN BE PERFORMED ON THE AMNIOCENTESIS FLUID SPECIMEN?

Several different tests may be performed using the amniotic fluid specimen. Specific tests are performed depending on the indications.

- ? **Chromosome analysis** - In the laboratory, the cells from the amniotic fluid are grown in a special nutritional fluid. Chromosomes obtained from the cells of the fetus (unborn baby) are then analyzed using a high power microscope. This prenatal test accurately detects more than 99.9% of chromosome abnormalities. Prenatal chromosome analyses performed by our laboratory are very accurate.

- ? **Open spine and open skull defects** - Approximately 99% of these defects may be detected using ultrasound examination **and** by testing for the fetal proteins, alpha-fetoprotein (AFP) and acetylcholinesterase (AChE) in the amniotic fluid. Skull and spine defects which are too small or covered by skin (closed) cannot be detected by these tests. Ultrasound alone does not detect all of the anomalies.

- ? **Inherited genetic disorders** - Some couples may be carriers of genetic disorders for which prenatal diagnosis can be performed. Carrier testing and prenatal diagnosis are available for many disorders like Tay-Sachs in the Jewish population, Sickle Cell in African-Americans and Latinos, Cystic Fibrosis in Caucasians.

Normal results from these tests do not eliminate the possibility your baby may have birth defects and/or mental retardation not detectable by these tests.

WHEN SHOULD AMNIOCENTESIS BE PERFORMED?

You and your physician may determine the optimal timing for your testing.

HOW IS AMNIOCENTESIS PERFORMED?

If you choose to have amniocentesis, a physician will examine the fetus by ultrasound. A small instrument is gently moved across your abdomen to produce images of the fetus on a TV screen. This exam is safe for the fetus. The ultrasound exam confirms the dating of your pregnancy, detects certain birth defects and identifies most twins. This exam cannot detect all birth defects.