



About Amniocentesis

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Am' ne o sen te' sis: the process of extracting amniotic fluid for analysis to determine the presence of genetic defects during pregnancy.

Introduction

Amniocentesis is primarily used to diagnose whether an unborn baby has one of two birth defects: a chromosome abnormality or a neural tube defect (NTD). Other disorders may be tested for when indicated by a person's family medical history. This procedure is usually performed between 15 and 22 post-menstrual weeks of a pregnancy. Women who are between 10 and 12 postmenstrual weeks may wish to consider a procedure called chorionic villus sampling (CVS), as an alternative to second trimester amniocentesis.

It is important to remember that there is no prenatal test which detects all birth defects. The decision to have prenatal diagnosis is best made by you. We encourage you to carefully read the following, to better understand some basic information regarding amniocentesis.

How is Amniocentesis Done?

A small sample of amniotic fluid (fluid surrounding the unborn baby) is obtained from the mother's uterus under ultrasound guidance. Ultrasound or sonography is the use of sound waves to visualize fetal and placental location, in order to choose the safest area for removal of amniotic fluid. Since ultrasound involves the use of sound waves, it is not a form of x-ray or radiation. A thin needle is inserted through the abdominal wall to withdraw about 2 tablespoons of amniotic fluid for analysis. The procedure takes 2-3 minutes. Menstrual-like cramping may occur during or after the test. Avoiding heavy lifting and strenuous exercise for 2-3 days are usually the main restrictions after amniocentesis.

When are Test Results Available?

Results from the test (standard chromosome analysis and amniotic fluid alpha-fetoprotein quantitation) take 7-10 days and are immediately sent to your doctor upon completion. Additional studies for other diseases may take longer.

What is the Risk of Amniocentesis?

The risk for complications from having an amniocentesis performed is about 1 out of every 200 women, or 0.5%. Complications include vaginal spotting or bleeding, leakage of amniotic fluid, severe cramping, fever, or infection. There is approximately a 1 in 1,000 chance that these complications could progress and result in miscarriage. Parents (one or both) are required to sign a consent form before having amniocentesis. The consent form states that you have been informed of and understand the risks and benefits of the procedure.

Who Should Consider Amniocentesis?

Amniocentesis may be considered for:

- Any pregnant woman who will be 35 years of age or greater at delivery.
- Individuals who have a child with a chromosome problem such as Down syndrome.
- Individuals who themselves have a chromosome translocation/abnormality.
- Couples at risk for a prenatally diagnosable genetic condition such as Tay-Sachs disease, cystic fibrosis, or sickle cell anemia.

- Individuals who have a close relative with a neural tube defect such as spina bifida or anencephaly.
- Women who have had an abnormal maternal serum screening blood test, indicating an increased risk for Down syndrome, trisomy 18, or neural tube defect.
- Circumstances when an ultrasound indicates possible fetal abnormalities.

Which Birth Defects Can be Detected From Amniocentesis?

Chromosome Abnormalities

Amniocentesis is often performed to detect the presence of a chromosome abnormality in an unborn baby. A chromosome is a structure in your cells that contains your inherited material, or genes. Humans usually have 46 total chromosomes in every cell of their body. Having too many or too few chromosomes results in birth defects and/or health problems. There have been several hundred different types of chromosome abnormalities seen in humans, with clinical outcomes ranging from near normal, to life threatening. Down syndrome is a disorder resulting from the presence of an extra #21 chromosome. Children with Down syndrome, for example, are mentally retarded and have specific facial features. They may also be born with heart problems, gastrointestinal tract (stomach) problems, or other birth defects. The chances for a baby to be born with Down syndrome or another chromosome abnormality increases as a woman becomes older.

Most of the time, chromosome abnormalities like Down syndrome are **not** inherited, so in **any** pregnancy, there is a chance to have a baby with Down syndrome or another chromosome problem. This is true regardless of a person's family medical history or how many healthy children they may already have. However, women who will be 35 years of age or greater at delivery, who have an increased risk for Down syndrome or another chromosome abnormality from their first trimester screening or maternal serum screening blood test, who already have a child with a chromosome problem, or who themselves have a chromosome translocation/abnormality are routinely offered prenatal diagnosis for fetal chromosome analysis. In these situations the risk for these problems is high enough to warrant consideration of the test. Many chromosome problems such as Down syndrome cannot be seen on ultrasound.

Neural Tube Defects

Amniocentesis can also be performed to detect an open neural tube defect in an unborn baby. Neural tube defects result when the neural tube, or developing spine, fails to close properly. During pregnancy, the human brain and spine begin as a flat plate of cells which 'rolls' into a tube, called the neural tube. If all or part of the neural tube fails to close, leaving an opening, this is known as a "neural tube defect" or NTD. This opening may be left exposed, or can be covered with bone or skin. There are two main types of NTDs: anencephaly and spina bifida.

Anencephaly occurs when the neural tube fails to close at the base of the skull, causing a serious defect of brain development. Babies with anencephaly are stillborn or usually live for only a few days. *Spina bifida* occurs when the neural tube fails to close along the spine, often damaging the spinal cord or tissue that surrounds the spinal cord. Some babies born with spina bifida have minimal or transient (temporary) problems, while some babies with spina bifida have permanent, often serious, physical problems. These can include paralysis, lack of bowel and bladder control, hydrocephaly (water on the brain), and mental retardation. In most cases one or more surgeries may be necessary.

About 1-2 out of every 1000 babies born will have a neural tube defect (either anencephaly or spina bifida); this risk is *not* dependent on the mother's age. Most of these babies (9 out of 10) are born to families who do *not* have a previous family history of NTDs. Therefore, in any pregnancy, there is a chance to have a baby with a neural tube defect. This is true regardless of a person's family medical history or how many healthy children they may already have. For this reason, it is recommended that all women of reproductive age take a multivitamin containing folic acid, a B vitamin which

reduces the risk for NTDs to occur. For families who *already* have relatives with some type of neural tube defect, the chance for their unborn baby to have a NTD *may be higher* than the general population risk. Genetic counseling is recommended for these families to best assess their risk and options for testing.

Depending on the fetal position and size of a neural tube defect, ultrasound alone can be used to detect some of the larger openings. Not all NTDs are visible on ultrasound.

How are Chromosome Abnormalities or Birth Defects Tested for by Amniocentesis?

Chromosomes are analyzed utilizing cells present in the amniotic fluid which are shed from fetal skin, the amnion (sac surrounding the unborn baby), and fetal gastrointestinal and urinary tracts. All of these cells contain chromosomes. In the laboratory these cells are separated from the fluid, placed in an incubator (called a cell culture) and chromosomes are examined under a microscope.

Alphafetoprotein (AFP) is a protein made by the baby's liver during the second trimester of pregnancy. AFP is normally present in the fluid surrounding the baby and crosses the placenta (afterbirth) into the mother's bloodstream. Adults usually do not produce AFP in measurable quantities, so that AFP comes only from the unborn baby (or babies). When an unborn baby has a neural tube defect, high levels of AFP are usually found in the amniotic fluid as well as in the mother's blood. The AFP is elevated because AFP "leaks out" of the spine opening. A small percentage of neural tube defects go undetected from amniocentesis when the spine opening is very small or covered by a flap of skin (called closed spina bifida). In general, AFP quantitation is performed on all amniotic fluid specimens no matter what the primary reason for performing the amniocentesis procedure. If the AFP is elevated, additional studies are done on the fluid to confirm the presence of a NTD.

Some other genetic diseases may also be diagnosed by DNA or enzyme analysis from an amniocentesis when indicated by a couple's family history.

How Accurate are the Results?

Chromosome results are greater than 99% accurate. Measurement of the amniotic fluid AFP is greater than 90% accurate. When amniocentesis is accompanied by high-resolution ultrasound examination performed by an experienced physician, the detection rate for open neural tube defects may be as high as 95-99%. The detection rate of other genetic diseases is dependent upon the method of testing.

Occasionally, results from amniocentesis need to be clarified through blood tests on the parents, additional ultrasound examinations, repeat amniocentesis, or fetal blood sampling.

It is important to remember that amniocentesis does not detect all birth defects or genetic diseases, since prenatal diagnosis is not yet available for many conditions. Every couple in the population has a 3% chance to have a child with a birth defect or health problem, regardless of their family/pregnancy/environmental histories, or amniocentesis results. The majority of women undergoing amniocentesis have normal test results and deliver a normal, healthy baby.

What if My Baby does have a Health Problem?

If a health problem is found in your unborn baby on amniocentesis, your doctor will discuss what this information means for your pregnancy and your options. Your doctor may also refer you to speak with a genetic counselor or other specialist, for further information regarding your baby's findings.

Should you have any questions about this information, or if you would like to make an appointment for genetic counseling, call the Emory Genetics Laboratory at 1-800-366-1502.