Genetic **Amniocentesis**

Most pregnancies result in a healthy baby, but when there is an increased chance that the baby may be born with a problem, prenatal testing is sometimes offered. Amniocentesis is a procedure in which fluid from around the fetus is withdrawn. This fluid contains cells from the fetus and can be used for genetic testing.

The most common reason for prenatal diagnosis is to look at the fetus' chromosomes. The chromosomes are the structures inside the cells of our bodies that contain the hereditary information (the genes). Sometimes, errors occur when egg or sperm cells are formed. These errors happen by chance. No one can cause or prevent them from happening. If one of these cells is involved in conception, the fetus could be left with too many or too few chromosomes being packaged into its cells. Having extra or missing chromosomes causes problems with development, both before and after birth. The most common of these chromosome problems is Down syndrome (Trisomy 21). The risk to have a child with an extra chromosome gets higher as the mother's age gets higher.

PROCEDURES AND RESULTS

Amniocentesis is usually performed after the 15th week of pregnancy as counted from the first day of the mother's last menstrual period. Ultrasound, which uses sound waves to create a picture of the placenta and fetus, is used to determine the location of the fetus and to guide the doctor during removal of a little of the amniotic fluid that surrounds the fetus. The doctor passes a thin needle into the uterus and removes about four teaspoons of amniotic fluid. The fluid is then sent to the laboratory for testing.

Results from chromosome testing usually take between nine and 12 days. If other tests are performed, the length of time for results will vary. When the result is available, the genetic counselor or nurse will telephone the patient directly with the information. Patients and their referring care providers are then sent a written report. Chromosome results are greater than 99 percent accurate. On rare occasions, the culture may fail to grow in the laboratory. If so, a repeat amniocentesis would be offered in order to obtain a result.

Amniocentesis is a relatively safe procedure. However, as with any medical procedure, certain risks must be considered. The risk for a serious complication, such as miscarriage or infection resulting from the procedure, in the United States is approximately 0.2 percent, or one in 500 chances. Normal side effects of the procedure can include menstrual type cramping, abdominal soreness, bruising or slight leakage of amniotic fluid.

It is also important to understand the limitations of amniocentesis. Every couple in the general population has a two to three percent risk of having a child with a birth defect or mental retardation. Many of these cannot be detected before birth. Additionally, genetic amniocentesis only tests for specific conditions for which the fetus is known to be at risk. Therefore, while most chromosome abnormalities can be ruled out, amniocentesis can't guarantee a normal or healthy baby.

We hope this information has been helpful to you. If you have additional questions, please feel free to call Perinatal center of Iowa at (515) 643-6888 to speak to the genetic counselor.

Chorionic Villus Sampling (CVS) is a procedure, similar to amniocentesis, used to determine whether or not there is a chromosome abnormality. The main advantage of CVS is that it can be performed as early as ten weeks gestation. An ultrasound-guided needle is inserted through the abdomen to collect a small sample of chorionic villus tissue from the placenta. If the placenta is positioned on the backside of the uterus, the sample of tissue is obtained through a catheter that is guided by ultrasound through the cervix into the placenta. CVS is proven to be a safe procedure, with a loss rate of approximately one-half percent (one in 200).

The following table gives the approximate risk of having a child with Down syndrome or any chromosome problem at various maternal ages:

MOTHER'S AGE	RISK DOWN SYNDROME	RISK ANY EXTRA CHROMOSOME
35	1 in 385	1 in 200
36	1 in 294	1 in 164
37	1 in 227	1 in 130
38	1 in 175	1 in 100
39	1 in 137	1 in 81
40	1 in 106	1 in 65
41	1 in 89	1 in 50
42	1 in 64	1 in 40
43-48	1 in 50 to 1 in 14	1 in 32 to 1 in 10

Another reason that a woman may be offered amniocentesis is an abnormal maternal serum screening test. Maternal serum screening tests go by many names and may be performed in the first or second trimesters of pregnancy. The tests use the amounts of certain chemicals in the mother's blood – and sometimes ultrasound measurements – to calculate the risk of Down syndrome and some other chromosome problems in the fetus. Whenever the calculated risk is greater than one in 200, amniocentesis is offered.

Prenatal diagnosis is also available for some genetic diseases that are caused by mistakes within single genes as opposed to whole chromosomes. If you know of a genetic condition in your or your partner's family, discuss it with your care provider as soon as possible.

PERINATAL CENTER *of* IOWA

330 Laurel St., Ste. 2300
Des Moines, Iowa 50314
(515) 643-6888
(877) 415-7447
Fax: (515) 643-6899
www.perinataliowa.com

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