Integrated Genetics has been a leader in genetic testing and counseling services for over 25 years.

This brochure is provided by Integrated Genetics as an educational service for physicians and their patients.

For more information on our genetic testing and counseling services, please visit our web sites:

www.mytestingoptions.com
www.integratedgenetics.com

Amniocentesis

A procedure that allows a pregnancy to be tested for certain kinds of birth defects

InSight® is a registered service mark of Esoterix Genetic Laboratories, LLC. ©2012 Laboratory Corporation of America® Holdings. All rights reserved. rep-040-v4-0612

About Integrated Genetics

Integrated Genetics has been a leader in genetic testing and counseling services for over 25 years.

This brochure is provided by Integrated Genetics as an educational service for physicians and their patients.

For more information on our genetic testing and counseling services, please visit our web sites:

www.mytestingoptions.com
www.integratedgenetics.com

Family history of an open neural tube defect

If a close relative has been born with an open neural tube defect, such as spina bifida or anencephaly, there may be an increased risk to other pregnancies in the family.

Abnormal maternal serum screening test

Screening tests performed on a sample of blood from a pregnant woman can identify pregnancies at risk for the common chromosome abnormalities, including Down syndrome and open neural tube defects. When the screening results show that a pregnancy has a high risk for one of these problems, amniocentesis for diagnostic testing is recommended.

Abnormal ultrasound

If an ultrasound shows an abnormality, amniocentesis may be recommended by your doctor.

How safe is amniocentesis?

Since it was developed in the late 1960’s, genetic amniocentesis has been performed on hundreds of thousands of pregnancies. Though it is now a routine procedure, it does have the following risks:

Miscarriage

Miscarriage is uncommon, but is the most serious risk of amniocentesis. Some miscarriages would happen anyway, but a few are caused by the procedure. The risk for miscarriage is 1 in 300 to 1 in 500. Early amniocentesis (before 15 weeks) has a higher risk, up to 1 in 100.

Bleeding, cramping, or leaking of fluid from the vagina

These symptoms, which happen in about 1% of women having amniocentesis, do not usually result in a miscarriage. However, a woman having these symptoms should call her doctor for advice.

Infection

Great care is taken to prevent infection. Therefore, infection following amniocentesis is very rare. However, a woman with fever or any flu-like symptoms after amniocentesis should call her doctor for advice.

Harm to the fetus

Since the ultrasound image gives the doctor exact information about the location of the fetus inside the uterus, the risk that the needle will harm the fetus is extremely low.

Rh problems

If a woman having an amniocentesis has Rh negative blood type, and the baby’s father has Rh positive blood type, the woman should have an injection of Rh immune globulin following the procedure. This helps prevent Rh disease in the baby.

Repeat testing

Very rarely, a doctor cannot obtain enough amniotic fluid, or the testing cannot be performed for some other reason. Neither of these situations indicates that something is necessarily wrong with the pregnancy, but amniocentesis would have to be repeated in order to get results.

Options after testing

Most women who have amniocentesis have normal test results. The few people who turn out to have abnormal results will need to speak with their doctors or with a genetic specialist about what their results mean and what to do going forward.

Deciding whether or not to have amniocentesis usually depends upon your particular risk factors for certain conditions, your concerns, your family situation and your feelings. Your health care provider can help to answer any additional questions you may have. All decisions about whether or not to have genetic testing are up to you.

Integrated Genetics

Client Services
(800) 848-4436

www.mytestingoptions.com www.integratedgenetics.com
Amniocentesis

Amniocentesis is a procedure that allows a pregnancy to be tested for certain kinds of birth defects. Since you are considering having an amniocentesis, this brochure will help answer some of your questions about the procedure and the tests that can be done.

What is amniocentesis?

Amniocentesis is the withdrawal of a small amount of amniotic fluid (the fluid surrounding a developing fetus) from the uterus. The procedure is done in a doctor’s office or a hospital, usually when a woman is about 16 weeks pregnant, counting from the first day of her last menstrual period. Before the procedure, the doctor performs an ultrasound or sonogram, which shows a picture of the uterus, the placenta, the amniotic fluid and the fetus on a screen. After reviewing the image, the doctor inserts a very thin needle through the woman’s abdomen into the uterus and takes out approximately one ounce of amniotic fluid (see drawing below). This part of the procedure lasts only a few minutes. After the sample is taken, another ultrasound check is done. Some women say that an amniocentesis does not hurt at all, while others say they feel pressure or cramping during the procedure. Often, people find that waiting for the test results is the most difficult part.

What tests can be performed on amniotic fluid?

Different tests can be performed on a sample of amniotic fluid, depending on why a particular pregnancy is at risk.

Testing for chromosome abnormalities

Most people who decide to have amniocentesis are at risk for having a baby with a chromosome abnormality. Chromosomes are tiny packages of genetic material, present in every cell of the body, which contain the information needed for a fertilized egg cell to turn into a human being. The amniotic fluid contains cells that have been shed from the fetus. Normally in each human cell there are 46 chromosomes. A missing or an extra chromosome, or a missing or an extra part of a chromosome, causes many changes in the way an unborn baby develops, and almost always leads to serious physical birth defects, mental retardation or both.

The most common chromosome abnormality is Down syndrome, caused by an extra #21 chromosome. People with Down syndrome have a distinct physical appearance, varying degrees of mental retardation and certain birth defects such as congenital heart disease. The most common chromosome abnormalities account for about two-thirds of all the chromosome abnormalities that can be detected by amniocentesis. The uncommon ones usually involve missing or extra parts of chromosomes.

The standard laboratory testing detects over 99% of all chromosome abnormalities. The results are usually available within 10 days.

Testing for inherited diseases

A woman may have amniocentesis because her pregnancy is known to be at risk for one of the inherited diseases that can be tested for prenatally. Examples include Tay-Sachs disease, cystic fibrosis and sickle cell disease. Test results are usually available in 1-5 weeks, depending on the test performed.

Testing for open neural tube defects

Amniotic fluid can also be tested for open neural tube defects, such as spina bifida and anencephaly, when the spinal cord or brain of the baby have not developed properly. The standard test measures a protein called alpha-fetoprotein (AFP), and detects over 90% of all open neural tube defects. The results are usually available in one week or less.

What cannot be tested with amniocentesis?

Every pregnancy has some risk, around 3-5%, for the types of birth defects that do not have a known cause. A few examples are cleft lip, non-specific mental retardation and most heart defects. Amniocentesis is not able to test for these kinds of problems.

Who should consider amniocentesis?

The American College of Obstetricians and Gynecologists recommends that all women, regardless of age, consider the option of amniocentesis for the diagnosis of chromosome abnormalities. In the past, only older women were offered amniocentesis because the risk for the most common chromosome abnormalities was known to increase with maternal age (see chart below). However, there are now non-invasive screening tests performed on a sample of a woman’s blood that can more accurately identify her specific risk for these common chromosome abnormalities. Therefore, a woman may choose one of the following options: to have a blood test to screen for the common chromosome abnormalities, followed by an amniocentesis if the screening test shows an increased risk; or to have diagnostic testing by amniocentesis first, without screening.

Of course, a woman may choose not to have any testing at all. If a woman is unsure whether or not she should have amniocentesis, she should speak with her doctor, a genetic counselor or other specialist in genetics.

In addition to the above choices that everyone has, people with additional specific risks may choose to have amniocentesis. Specific risks include:

Family history of a chromosome abnormality or inherited disease

If someone in the family, especially a close relative, is known to have a chromosome abnormality or inherited disease, there may be increased risk for another child to be born within the family with the same condition.

The parents are carriers of an inherited disease

If the parents of the unborn baby have been found to be carriers of an inherited disease, the pregnancy is at increased risk for the child to be affected. Many inherited diseases today can be diagnosed prenatally.

Maternal Age Risks for Chromosome Disorders in Pregnancy

(numbers are approximate)