Genetic Support Foundation

Amniocentesis

Amniocentesis is an optional procedure that can be done in pregnancy to test for certain genetic conditions and birth defects. Amniocentesis is usually performed at approximately 15-22 weeks of pregnancy at a doctor's office that specializes in doing this procedure. Using ultrasound as a guide, a very thin needle is inserted through the mother's abdomen into the amniotic sac, a fluid filled area surrounding the baby. A small amount of fluid that contains some of the baby's cells is withdrawn and sent to the laboratory for testing. The lab can look at the cells to determine if there are certain genetic conditions present in the baby.

Typically, it takes only a couple of minutes to perform an amniocentesis. Many women experience some brief discomfort with amniocentesis, such as a sensation of cramping, pressure, or pinching. Usually women return to their normal activities within 12 days following amniocentesis. Preliminary results for common chromosome conditions (including Down syndrome) can generally be reported within 48-72 hours from when the lab receives the fluid.[1] It can take about two weeks for the complete test results to return from an amniocentesis.

What Does Amniocentesis Test For?

It is important to keep in mind that no prenatal genetic test, not even amniocentesis, can look for all genetic conditions or birth defects. The most common conditions detected by amniocentesis with traditional chromosome analysis are chromosome conditions, such as Down syndrome (trisomy 21), trisomy 18, trisomy 13, and differences in the number of X or Y chromosomes (sex chromosome variations). Amniocentesis can also detect open neural tube defects, such as spina bifida, if requested. The prognosis for these conditions can vary tremendously depending on the condition and even between individuals with the same condition. In some cases, additional genetic testing may be ordered that can assess for other conditions, especially if a known genetic condition runs in a family or an abnormal ultrasound finding is seen.

[1] Although preliminary results are thought to be very accurate, false positives and false negative results can occur with this test. It is recommended that individuals wait for the final results before making any permanent decisions about the pregnancy.

Some doctors are offering prenatal chromosomal microarray (instead of traditional chromosome analysis) with amniocentesis. Prenatal chromosomal microarray will look for the chromosome conditions listed above, but it also looks for others called microdeletions and microduplications. For more detailed information, including the pros and cons of prenatal chromosomal microarray, visit: **http://tinyurl.com/h27hrmy**

How Certain are Amniocentesis Results?

Amniocentesis is a diagnostic test which means results are considered to be definitive for the conditions tested. In other words, this test can give you "yes" or "no" answers. Rarely, lab errors or uncertain results may occur.

What are the Risks of Amniocentesis?

Most women have no complications from having an amniocentesis. However, amniocentesis does increase the risk for some complications including infection, leaking of amniotic fluid, preterm labor, or miscarriage. When an amniocentesis is performed by a doctor who specializes in this procedure and does them on a routine basis, the risk for miscarriage is approximately 1 in 900, or 0.11%.[2] Over 99% of the time, no serious complications occur.

Making a decision that is right for you…

Making a decision about amniocentesis can be difficult because it is not risk-free. However, a diagnostic procedure such as amniocentesis can provide more definitive information about genetic conditions in your baby, and can provide information about more genetic conditions than screening tests, such as blood tests and ultrasounds. Some individuals feel that they would want this information during pregnancy a some do not feel this information is helpful. Decisions about genetic testing should be based on your own beliefs, values, needs, and personality.

You can find more information and videos about prenatal genetic testing options at: www.geneticsupportfoundation.org and www.doh.wa.gov.

[2] Based on American College of Obstetricians and Gynecologists/Society for Maternal-Fetal Medicine Practice Bulletin 162, Prenatal Diagnostic Testing For Genetic Disorders, May 2016.