Quest Advanced®
Women's Health

Prenatal Testing

Amniocentesis

Patient Navigators from Quest can help

Patient Navigators are support specialists who will work directly with you to answer questions about prenatal screening.

- Review your insurance coverage
- Answer billing questions
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You just want your baby to be healthy.

Prenatal (before birth) diagnosis can provide valuable information and insight. Prenatal diagnosis finds out if a baby has a chromosome abnormality or other specific inherited disorders. This brochure can help you learn about a prenatal diagnostic procedure called amniocentesis. It can also help you decide if this procedure is right for you.

Who should consider prenatal diagnosis?

The American College of Obstetricians and Gynecologists recommends making prenatal diagnosis available to all pregnant women, regardless of their age.

Amniocentesis is most commonly used for prenatal diagnosis following an abnormal prenatal screen for a chromosome abnormality, a neural tube defect, or an abnormal ultrasound. It is also an option when there is a family history of an inherited disorder or when the mother and/or father is a carrier of an inherited disorder.

Do I have to have a prenatal diagnosis?

You should discuss your options with your healthcare provider, but the decision about whether to have prenatal diagnosis is up to you.

What is the difference between prenatal *screening* and prenatal *diagnosis?*

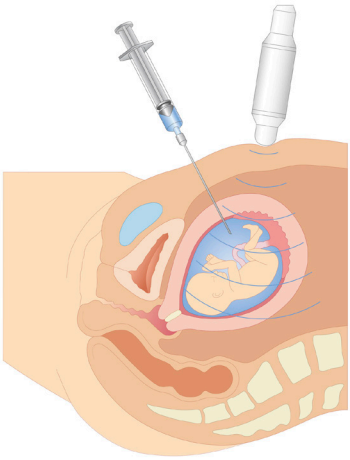
Prenatal *screening* provides information about a baby's risk of having a chromosome abnormality, a neural tube defect, or some other inherited disorder. Prenatal *diagnosis* will tell if the baby most likely has that disorder.

What is amniocentesis?

Amniocentesis is a diagnostic test that is usually performed as an outpatient procedure typically during the 2nd trimester of pregnancy. Most commonly it is used to test for the presence of a chromosome abnormality and/or a neural tube defect. Amniocentesis can also test for a number of other inherited disorders. Cystic fibrosis, sickle cell, and Tay-Sachs disease are examples of these. Diagnostic tests for these inherited disorders are usually only recommended when there is an increased risk of the disorder due to family history, carrier screening results, or other special circumstances.

How is amniocentesis performed?

Under ultrasound guidance, a thin needle is inserted through the abdomen and into the uterus. Your doctor then removes approximately 2 tablespoons of amniotic fluid with the needle. The fluid contains cells and other substances from the baby. The lab can test the fluid and cells to find out if the baby has a chromosome abnormality, a neural tube defect, or some other inherited disorder.



Amniocentesis is usually performed during the 2nd trimester of pregnancy as an outpatient procedure.

How accurate is amniocentesis?

Amniocentesis can detect greater than 99% of chromosome abnormalities and greater than 96% of neural tube defects. The results for other inherited disorders are also very accurate. There is a small chance that cells from the mother may be present in the sample. This may make the results more difficult to interpret.

It is important to emphasize that a negative (normal) result does not guarantee a healthy baby.

What are the risks of amniocentesis?

When performed by an experienced physician, amniocentesis is considered very safe. As with any procedure, there is a risk of complications. Miscarriage, bleeding, or infection may occur.[1] You should discuss the risks and benefits of the procedure with your healthcare provider.

What if the result is abnormal?

If your results are abnormal, you should discuss their meaning and your options with your healthcare provider. Genetic counseling can help you learn more and help you make decisions in the best interest of yourself and your family.

What information is not provided by amniocentesis?

Major structural or genetic birth defects affect approximately 3% of births in the United States.[2] Amniocentesis provides information about neural tube defects, congenital disorders caused by chromosome abnormalities, and any other inherited disorder that was specifically studied.

Amniocentesis does not test for all possible congenital disorders, such as heart conditions or intellectual disabilities.

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For more information, visit **QuestDiagnostics.com** and talk with your healthcare provider.

This brochure is for informational purposes only and is not a substitute for medical advice, diagnosis, or treatment. The diagnosis or treatment of any disease or condition may be based on personal history, family history, symptoms, a physical examination, laboratory test results, and other information considered important by your doctor. Always talk with your doctor about the meaning of your test results and before you stop, start, or change any medication or treatment.

Reference:

1. Friedrich E, Porto M. Aneuploidy screening. In: Di Saia PJ, Chaudhuri G, Giudice LC, Moore TR, Porto M, Smith LH, eds. *Women's Health Review: A Clinical Update in Obstetrics-Gynecology.* Elsevier Inc; 2012:57-70.
2. Update on Overall Prevalence of Major Birth Defects, 1978-2005. Accessed February 5, 2023. https://www.cdc.gov/mmwr/preview/mmwrhtml/mm5701a2.htm

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