Diagnostic testing during pregnancy *Amniocentesis*

Patient Information Series – What you should know, what you should ask.

What I need to know about Amniocentesis?

The accurate test currently accepted for diagnosing chromosomal and genetic abnormalities in pregnancy is an invasive test, such as amniocentesis, which is most commonly done between $15^{0/7}$ to $17^{6/7}$ weeks' of gestation. Unlike the biochemical laboratory screening tests performed in the first and/or second trimester of pregnancy, whose results are only statistical, the results obtained from tests performed on the amniotic fluid sample are diagnostic. Since amniocentesis is an invasive test, the extra risk of pregnancy loss following an amniocentesis, conducted by a suitably trained operator, is expected to be less than 0.5%.

What should I expect during the test?

Amniocentesis is conducted using a delicate needle inserted through the abdominal wall into the uterus. Before needle insertion, the abdominal area is meticulously disinfected to minimize infection risk. With the guidance of ultrasound imaging, the precise location is determined, and a thin needle is introduced to extract small amount of amniotic fluid.

Occasionally, based on medical assessment, a blood sample might be taken from the pregnant person or both partners. If blood extraction is required, you will be informed about it during genetic counseling. For pregnant people, with a negative blood type (Rh-), Rho(D) immune globulin injection is administered after the procedure. The entire process usually lasts just a few minutes.

Is there any preparation needed before the test?

No specific preparation or fasting is needed before the test. If you are currently taking medications like aspirin or other blood thinners, it is advisable to discuss their continuation with your physician before undergoing the test.

What are the post-test instructions?

Following the procedure, 24-72hr of moderate activity can be considered. After the test, you might experience abdominal tenderness, or pressure, along with mild discomfort at the injection site. Some pregnant people may also notice slight vaginal bleeding or a minor amniotic fluid



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leakage. It is crucial to seek medical attention if you experience symptoms such as chills, high fever, excessive bleeding, amniotic fluid loss, or the onset of contractions after the test.

What does the laboratory examine in the amniotic fluid test?

The amniotic fluid sample, containing cells rich in DNA of your baby, undergoes laboratory processing and, in some cases, cell culture to explore the developing fetus' genetic makeup and analyze fetal protein level.

What Could Be the Potential Test results?

The test results can fall into several categories:

Normal result: This means that no detectable genetic differences associated with known genetic syndromes were identified in the fetal examination. However, it is important to note that even a normal result does not guarantee the health of the fetus.

Abnormal result: This indicates that the test revealed evidence of an abnormal genetic aberrations that linked to a recognized genetic syndrome. If necessary, you and your family will be invited to the clinic for a follow-up genetic counseling session. During this session, the test result will be explained in detail, and the possibility of further tests during pregnancy or within the family will be discussed.

Test results with uncertain significance: Test results may include findings with ambiguous implications. These findings pertain to changes, as of the test date, are yet to be determined as causing medical issues or an elevated risk thereof. The presence of such outcomes in the fetus can occasionally lead to uncertainty and emotional strain. Therefore, your medical team will further inform you of how to proceed in these rare cases.

Last updated February 2024

