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WHAT ARE MY DIAGNOSTIC TESTING OPTIONS?

There are two diagnostic testing options available during pregnancy: chorionic villus sampling (CVS) and amniocentesis. These tests typically provide a "yes" or "no" answer about whether a pregnancy has a chromosome abnormality or other genetic condition. These testing options are much more comprehensive than blood/screening tests but are considered invasive and involve some risk.

Chorionic villus sampling (CVS)

Chorionic villus sampling (CVS) is typically performed between 11 and 14 weeks of pregnancy. During CVS, a small amount of placental tissue is collected for testing. CVS is performed transcervically or transabdominally, using ultrasound as a guide. How CVS is performed depends on the location of the placenta. The entire procedure typically takes a couple minutes to complete.

- The transcervical method involves passing a thin tube through the opening of the cervix and into the placenta.
- The transabdominal method involves passing a thin needle through the patient's abdomen and uterus into the placenta.

The genetic information found in the placenta is usually identical to the baby's genetic information. However, in a small number of cases, the two can be different and further testing such as amniocentesis may be recommended to clarify CVS results.

Amniocentesis

Amniocentesis is typically performed after 16 weeks of pregnancy. Using ultrasound as a guide, a thin needle is inserted through the pregnant patient's abdomen into the amniotic sac, a fluid-filled area surrounding the baby. A small amount of the amniotic fluid is collected for testing. The fluid contains some of the baby's cells, which contain the baby's genetic information.

What do these tests look for?

The results of invasive testing are diagnostic, meaning that the results are definitive for the conditions tested. There are various genetic tests that can be done through diagnostic testing. Some of the most common conditions tested include Down syndrome, trisomy 13, trisomy 18, and differences in the sex chromosomes. Genetic testing for specific conditions can also be done when there is a known genetic condition in the family or if an abnormal ultrasound finding is detected. However, it is still not possible to test for all genetic conditions or birth defects.

What are the risks?

CVS and amniocentesis procedures are performed in a doctor's office by a maternal-fetal medicine specialist, and the risk for complications from the procedures is less than 1%. While minimal, the risks can include cramping, infection, leakage of amniotic fluid, and premature labor or miscarriage. Patients may also experience some discomfort during the procedures, including the sensation of cramping, pressure, or pinching. Most patients who undergo CVS or amniocentesis do not have any complications and can return to their normal activities one to two days after the procedure.

Is diagnostic testing right for me?

Deciding whether or not to proceed with genetic testing can be difficult. Decisions around genetic testing and the type of testing, invasive or noninvasive, should be based on each patient's specific risk factors, needs, values, personality, and beliefs. It is important to discuss these and any questions you may have with your doctor or genetic counselor.

Where can I learn more about diagnostic testing?

Talking to a genetic counselor may be beneficial to learn more about genetic testing options in pregnancy and help you decide on a personal screening plan. **To schedule an appointment with a genetic counselor, please call our scheduling team at 713-486-9302.**

For more information, scan the QR code or visit **UTPhysicians.com/genetic-counseling.**