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Genetic Testing:
Which Prenatal Tests Are Right For Me?

Maternal Fetal Medicine

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This brochure is an overview of prenatal testing options for chromosome conditions such as Down syndrome. With advances in technology, there are now more options than ever. Before choosing any test, it may be helpful to discuss this information with your doctor and/or genetic counselor. This will help you get the knowledge and support you need to make the best decisions for you and your pregnancy.

Why would I consider prenatal testing?

Most babies are born healthy. However, all pregnancies have a 3-5% chance for a baby with a birth defect or intellectual disability. Prenatal testing can help detect some of these conditions. Your test results may reassure you, help you prepare for a baby with special needs, or alert you if a problem is detected. However, not all health or developmental concerns can be detected by prenatal testing.

What are chromosome conditions?

Chromosomes are bundles of DNA in our cells. We typically have two copies of each chromosome. Sometimes there is an extra or missing chromosome. This extra or missing DNA may cause a variety of concerns, including birth defects and intellectual disability. As an example, Down syndrome happens when there is an extra chromosome 21. There are other chromosome conditions besides Down syndrome. They can be milder or more severe depending on which chromosome is involved. Most chromosome conditions happen by chance, and are not passed through the family.

What kind of tests are available?

During pregnancy you have the option of a screening test and/or a diagnostic test.

What is a prenatal screening test?

Prenatal screening tests determine the chance for a health condition in a baby. However, they will not tell if a condition is absolutely present or absent. They typically involve a blood draw and/or ultrasound. Some women use screening tests to help decide whether or not to have a diagnostic test.

What is a prenatal diagnostic test?

Prenatal diagnostic tests tell for certain that a condition is present or absent in a pregnancy. They involve a procedure, such as CVS or amniocentesis.

Screening tests are overall less invasive, while diagnostic tests provide more accurate information.

SCREENING TESTS

Early Anatomy Ultrasound

This ultrasound is performed between 11-14 weeks, and checks for uncommon, but major, birth defects. It will also look at the nuchal translucency (NT), a pocket of fluid at the back of the baby's neck. The NT measurement is used in some Down syndrome screens, and is also used to estimate risk for other conditions and birth defects.

An early anatomy ultrasound is recommended for all patients, regardless of if they decide to proceed with other screening or diagnostic genetic tests. It is difficult to see certain birth defects in the first trimester; therefore, a detailed ultrasound at 20-22 weeks is also recommended.

First Trimester Only/ Combined Screen

The First Trimester Only/Combined screen requires a single blood draw and an early anatomy ultrasound with NT, both done between 11-14 weeks. The result estimates the chance of Down syndrome and Trisomy 18. This screen will detect up to 90% of pregnancies with Down syndrome. About 5% of patients will have results in the high risk range, and will be offered diagnostic testing. The majority of pregnancies with a high risk result do not have Down syndrome. This is called a false positive screening result.

Cell-Free Fetal DNA (CFFD) Testing

This is a blood test which looks at small pieces of DNA found in a woman's blood stream during pregnancy. CFFD can detect higher or lower amounts of DNA from select chromosomes, including chromosomes 21, 13 and 18. It can be drawn any time after 10 weeks, and will detect at least 99% of pregnancies with Down syndrome. False positive results are rare (<0.1%), but possible. Therefore, it is recommended that any abnormal result be confirmed with diagnostic testing.

CFFD is available to all women; however, its accuracy and insurance coverage varies, depending upon whether your pregnancy is considered "high risk" or "low risk" of having a chromosome condition. High risk patients include women who are age 35 or older, have had a previous pregnancy or child with a chromosome condition, or have had other screening tests (ultrasound or blood) that suggested an increased risk for a chromosome condition.

CFFD is rapidly changing. New conditions are frequently being added. Screening test accuracy will vary, depending on the specific condition.

Genetic Disease Carrier Screening

A small number of babies are born with genetic conditions that are passed through the family (inherited). Many of these conditions happen with no family history of the disease. A blood test called genetic carrier screening can help identify couples at risk of having a baby with some genetic conditions. Most carriers are healthy; however, when both parents are carriers of the same genetic condition, there is an increased risk to have a baby with that condition.

Every ethnic group has genetic conditions more common to it. A patient's ethnicity can be used to guide which carrier screens are offered. As mixed ethnicities are common, there is a trend to offer larger panels of carrier screens to all couples, regardless of ethnicity. Test accuracy depends on ethnicity and condition. No screen has a 100% detection rate; therefore carrier risk can be reduced, but not completely eliminated with a normal result.

DIAGNOSTIC TESTS AND PROCEDURES

Procedures

CVS
Chorionic villus sampling (CVS) can be done between 11-13 weeks. It involves sampling a small amount of placental tissue under ultra- sound guidance.

Amniocentesis
Amniocentesis can be done any time after 16 weeks. It involves removing a small amount of amniotic fluid (water that surrounds the baby) under ultrasound guidance.

Diagnostic tests can be done on samples from either CVS or amniocentesis procedures.

Diagnostic tests

Diagnostic tests can be done on samples from either CVS or amniocentesis procedures.

Karyotype/chromosome studies
This is a traditional test that creates a picture of chromosomes from cells. It will detect extra or missing chromosomes, and large changes in the structure of the chromosomes.

Microarray
This is a newer test that will detect smaller missing or extra pieces of DNA which could be missed by traditional chromosome studies. It will also detect most extra or missing chromosomes.

Frequently asked questions about prenatal diagnostic procedures

How safe are these procedures?
The doctors at Providence Maternal Fetal Medicine are specialists with years of experience with high-risk pregnancies and these procedures. The most recent studies suggest that the risk for complications or miscarriage after CVS or amniocentesis is low.

Will my activities be restricted following my procedure?
You can do most normal activities including walking and driving. However, for 24 hours after your procedure, you should not lift anything heavier than 10 pounds, participate in strenuous activity or exercise, or have sexual intercourse.

Will a normal test guarantee a healthy baby?
No. The diagnostic tests will identify some conditions, such as Down syndrome. However, they cannot detect all possible concerns such as cerebral palsy, autism, heart defects, and many forms of intellectual disability.

How can I learn more about my options?
You can learn more about your options by talking to your doctor or a genetic counselor. Your doctor can refer you to meet with a genetic counselor at Providence Maternal Fetal Medicine. In addition, online videos about prenatal genetic testing options can be viewed at www.swedish.org/genetic testing. Information is also available on the **Circle by Providence** app.

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