COLUMBIA | OBSTETRICS AND GYNECOLOGY

Your options for genetic screening and carrier testing

We recommend screening for Down syndrome, Trisomy 18, Trisomy 13 and sex chromosomes using Non-Invasive Prenatal Testing (NIPT). The laboratory we use is LabCorp.

Some insurance companies may not cover this test if you are not over the age of 35 or do not have a medical indication. Please call your insurance company to ascertain whether this is a covered benefit and what your financial responsibilities may be.

You can estimate your out-of-pocket cost from LabCorp by visiting their web-site: www.integratedgenetics.com

- Choose the Estimate my Cost Tab
- Choose Pregnancy for the type of test
- Choose MaterniT 21 Plus
- Complete the form using your insurance information

If it is not a covered benefit, you may choose to pay out of pocket for this test. The cost is $299.

- The CPT code is: 81420
- The ICD-10 codes are: Elderly primigravida, first trimester O09.511
    - Elderly primigravida, second trimester O09.512
    - Elderly multigravida, first trimester O09.521
    - Elderly multigravida, second trimester O09.522
    - Encounter for supervision of normal pregnancy, unspecified, first trimester Z34.91
    - Encounter for supervision of normal pregnancy, unspecified, second trimester Z34.92
    - Abnormal biochemical finding on antenatal screening O28.01 Abnormal ultrasound finding O28.3

What is NIPT?

Non-invasive prenatal testing (NIPT) is a blood test done during pregnancy. This test measures small pieces of DNA in a patient's blood. DNA is the genetic information we inherit from our parents. When you are pregnant, a small amount of the DNA in your blood comes from the placenta. NIPT uses this DNA to tell us about your baby's chance to have certain genetic conditions, called chromosome disorders, such as Down syndrome.

How is it done?

NIPT involves a simple blood test. It can be done any time after the 9-10th week of pregnancy.

What can NIPT tell me?

NIPT can determine if your pregnancy has a low chance or a high chance to have a few chromosome disorders, including Down syndrome (trisomy 21), trisomy 18, and trisomy 13. NIPT can identify 99% of pregnancies with these conditions. Down syndrome causes a baby to have changes in development such as intellectual disabilities and in some cases, a heart defect. Trisomy 18 and 13 cause a baby to have birth defects and frequently pass away before one year of age. NIPT also screens for sex

chromosome disorders, which cause milder symptoms. You can find more information about this test at the American College of Obstetricians and Gynecologist website: https://www.acog.org/womens-health/infographics/cell-free-dna-prenatal-screening-test.

What does NIPT NOT tell me?

NIPT will not tell you about problems that are screened for by ultrasound, such as spina bifida, heart defects, or other birth defects. It will also not test for all possible genetic conditions that a baby can have.

How long does it take to get results from NIPT?

Results from NIPT usually take 7-10 days. You will get your result at your next doctor's visit.

What does it mean if my NIPT result is low risk (normal)?

Most pregnant patients get a low risk NIPT result. A low risk result is very reassuring. It means that your baby does not appear to have Down syndrome, trisomy 18, or trisomy 13. In rare cases, a baby who has one of these conditions will be missed by the test. This is because NIPT is not 100% accurate. A normal result does not guarantee that the baby is healthy. Keep in mind that this test does not look for all chromosome or genetic disorders.

What if NIPT does not give ANY result?

In some pregnancies, NIPT is unable to give a result. This does not usually mean there is a problem with the developing baby. NIPT may not give a result in heavier women or if the blood was drawn early in pregnancy. Sometimes when a baby has a chromosome disorder, NIPT is unsuccessful and there is no result. If your NIPT does not give a result, you can decide to repeat the NIPT or choose a different type of genetic test.

What does it mean if my NIPT result is high risk (abnormal)?

A high risk result may mean that the developing baby has a chromosome condition. Even when the test result is abnormal, however, there is a chance that the baby does not actually have the condition. This is because NIPT is not 100% accurate. You will be referred to speak to a genetic counselor and will be offered additional testing to determine more about the health of the pregnancy.

What follow-up testing will I be offered if my result shows a high risk?

You will be offered a diagnostic procedure called chorionic villus sampling (CVS) or amniocentesis. CVS and amniocentesis can determine with certainty if your pregnancy has a chromosome disorder. Your doctor and genetic counselor will tell you more about how the CVS and amniocentesis are performed if your result is high risk.

What if I want more information even if my blood test is normal?

All pregnant women can choose to do further testing, like the CVS or amniocentesis, if they would like to know about chromosome disorders with certainty. If you have specific questions about testing, your family history, or other genetic test results in your pregnancy, your doctor can refer you to a genetic counselor.

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Genetic carrier screening

We offer genetic carrier screening to our patients. The company we use is Myriad and the test name is Foresight Carrier Screen. It is a blood test that analyzes your DNA to see if you are a carrier of mutations of specific diseases.

The Foresight Carrier Screen looks for serious conditions that you could pass on to your child. Some are conditions you may have heard of, such as cystic fibrosis. Some conditions can be treated early, others require lifelong management, and still others have no treatments.

DNA contains two copies of every gene - one inherited from a mother, and one from a father. These genes sometimes pass on inherited conditions which are caused by changes in genes called mutations.

Most people carry at least one mutation in a gene included in the Foresight Carrier Screen. This usually only becomes an issue if both you and your partner have a mutation in one copy of the same gene. When this happens, even though neither of you have any symptoms, there is a 1 in 4 chance for each pregnancy that your child will be affected by the condition associated with the gene.

There are a few conditions where only the mother needs to carry a mutation for her children to be at risk of developing symptoms.

This testing may be a covered benefit from your insurance plan. If it is not a covered benefit, the out of pocket cost for this test is $199.

For a list of all the diseases in this panel or for billing information, visit this website: https://myriadwomenshealth.com/patient-foresight/