Cystic Fibrosis Testing

This is the most common genetic disorder affecting about 1 in 3,300 people. The chance for a person to be a carrier for this condition depends on their ethnic background. Approximately 1 in 25 Caucasians, 1 in 46 Hispanic individuals, 1 in 65 African Americans and 1 in 90 Asians are carriers for this condition. Cystic Fibrosis causes the body to produce abnormally thick mucus, leading to life-threatening lung disease and digestive problems. Symptoms can be variable ranging from mild to severe. However, the average lifespan for a person with this condition is into their later 30s. If you are negative, this REDUCES your chance to be a carrier, but does not eliminate it. If you are found to be a carrier, testing the father of this baby will be necessary. If you are both found to be carriers, there will be a 25% chance for your baby to have this condition.

BRUNSWICK HILLS
OBSTETRICS & GYNECOLOGY

GENETIC TESTING IN PREGNANCY

Please read and complete this packet for your next visit.
Our Providers will address any of your concerns at your next appointment.

Patient name (printed): _____ Date: _____

Patient signature: _____

Witness: _____ Date: _____

Congratulations on your pregnancy. We know that making decisions regarding available testing during your pregnancy can be difficult. The information in this packet will hopefully clarify the testing options available to you. Additional information is also available on our website **http://www.womensobgyn.com (under genetic counseling services.)**

Expanded Carrier Testing

This screening test can show if a person carries a gene for an inherited disorder. We will test you to see if you carry any of these genes. You will be tested for conditions that are mostly severe and may cause significant health problems; however, some of the conditions are milder. Please note, that 1 in 4 people who have this panel will be found to be a carrier for at least one condition. If you are found to be a carrier, you will be scheduled with a Genetic Counselor to discuss the results further. Your partner will then be tested to see if he is also a carrier. You both need to carry the gene for the inherited disorder to be present in your baby. It is important for you to understand that these are the most comprehensive panels currently available; however it still does not test for every possible genetic condition.

If you are of Jewish Ancestry, we highly recommend you have this panel which is the most cost-effective way to test for Jewish disorders.

The two Lab choices are:

Inherigen is a comprehensive carrier screen that tests for 163 inherited diseases. Inherigen plus will include testing for Cystic Fibrosis, Spinal Muscular Atrophy, and Fragile X. This test is covered by most insurance plans.

Inheritest is a comprehensive carrier screen that tests for 90 inherited diseases. This test includes Cystic Fibrosis. This test is covered by most insurance companies. Maximum out of pocket for this test is $475.

☐ **I want** to be tested for the Expanded Carrier Test (which includes SMA, Fragile X, and Cystic Fibrosis)
Please circle: Inherigen Panel Inheritest Panel
(Please skip to page 3 – if selecting this option)

☐ **I do not want** to be tested for the Expanded Carrier Test.
(Please read page 2 – if selecting this option)

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OBSTETRICS & GYNECOLOGY

IS YOUR BABY AT RISK?

Is your baby at risk for a chromosome problem, such as Down syndrome?

The risk for a chromosome problem in a pregnancy is related to the age of the mother. As women get older, the risk for a chromosome problem increases.

Chromosome problems occur by chance and have nothing to do with your family history. The most common chromosome problem that babies can be born with is called Down syndrome (trisomy 21). About 1 in 800 babies overall are born with Down syndrome.

Typically, women who are 35 years old or older have the option to have an invasive testing, such as chorionic villus sampling or amniocentesis in their pregnancy to learn for sure if their baby has a chromosome problem. However, both these procedures are invasive and carry risks for miscarriage. In addition to these two procedures, there are other non-invasive options which will be reviewed below. Please remember that only amniocentesis and CVS testing are 100% accurate.

The following tests are usually covered by most insurance companies however; we cannot guarantee coverage by your health plan. Please contact your insurance company to verify coverage. Additional information on the testing options listed, is available on our website at http://www.womensobgyn.com/Genetic_Counseling.html.

First Trimester Screening

This test has to be done between 11 weeks 0 days and 13 weeks 6 days. This is a blood test and ultrasound. The ultrasound is done to measure the amount of skin behind the baby's neck, called nuchal translucency. Sometimes you will be asked to return at 16 weeks for a second blood test, called a **sequential screen**. This testing can detect 90% of pregnancies at risk for fetal Down syndrome and about 97% of pregnancies which are affected by two other chromosome problems called trisomy 13 and 18. Please note that sometimes this screen can suggest a risk, but there is nothing wrong with the pregnancy. All patients whose test comes back increase risk will be referred for genetic counseling and the option of further testing.

☐ I want First Trimester Screening: Please contact your insurance company to find out which lab would be the best option for you for the blood work portion of this test.

☐ **I do not want** to be tested for First Trimester screening.

NTD labs: We feel that this laboratory allows you to have the fastest screening result with the highest accuracy (90% accuracy within 5 days). You may visit http://ntdlabs.com/maternal-marker-testing/first_trimester_screen.php for additional information. Please note that this laboratory is out of network for Blue Cross and Blue Shield. The maximum amount you would be responsible for is $75.00. If you need to meet an in network deductible the cost can be up to $165.00.

Integrated Genetics a LabCorp Specialty testing group: This lab is considered in network with Blue Cross and Blue Shield. This lab does not include Trisomy 13 in the testing. This is a two part test. Part one is an ultrasound and bloodwork, part two is an additional blood work at 16 weeks. You may visit https://www.labcorp.com/wps/wcm/connect/IntGeneticsLib/integratedgenetics/resources/pdfs/brochures/patient-brochure-first-screen for additional information. Deductibles may apply. The fees for these tests are around $300.00.

If you are having Twins we highly suggest that you choose NTD Labs.

☐ **I do not want** Chorionic Villus Sampling

☐ I want Genetic Counseling to receive more information on the above testing: Genetic counseling may be out of network. The maximum cost for genetic counseling is $150.

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