BRUNSWICK HILLS
OBSTETRICS & GYNECOLOGY

Patient name (printed): _____Date: _____

Patient signature: _____

Witness:_____ Date: _____

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.**

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. Blue Cross/ Blue Shield HMO cannot use this lab.

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

□ **I do not want** to be tested for the Expanded Carrier Test.

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 mucous, 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.

Spinal Muscular Atrophy testing:

This is the second most common genetic disorder in the U.S. affecting about 1 in 6,000-10,000 individuals of all racial and ethnic groups. Approximately 1 in 41 people carry the gene for this disorder. There are several forms of this disorder causing variable age of onset and severity of symptoms. In general, progressive muscle degeneration occurs affecting crawling, walking, swallowing and head and neck control. In the most severe form, children have difficulty with breathing by age two. A blood test can determine if you are a carrier for this condition. 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. This test is covered by most insurance companies. Maximum out of pocket is usually $75.

Fragile X carrier testing:

This is the most common cause of inherited mental retardation affecting about 1 in 4,000 males and 1 in 8,000 females. It has been estimated that about 1 in 260 women are carriers for this condition. People with this condition have delayed speech and language, a degree of mental retardation, behavioral characteristics such as hyperactivity, autism and poor eye contact.
This test is covered by most insurance companies. Maximum out of pocket is usually $75.

Brunswick Hills OB/GYN follows the recommendations of the American Congress of Obstetricians and Gynecologists. We test every patient for Cystic Fibrosis, Spinal Muscular Atrophy, and Fragile X syndrome.

□ I **do not** want to be tested for Spinal Muscular Atrophy, Fragile X , and Cystic Fibrosis.

Genetic counseling is available if you would like to discuss any of these testing options. Genetic counseling may be considered out of network. Maximum cost for genetic counseling is $150.00.

These testing options are usually covered by most insurance companies however; we cannot guarantee coverage by your health plan. Please contact you insurance company to verify coverage. Deductibles may apply.

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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 you 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.

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.

Non-invasive Prenatal Screening:

Depending on the laboratory used, this test may be called the Harmony, Informaseq test, MaternT21 testing, or the Verify test, and Panorama. This is a new test which has only been available since the beginning of 2012. This is a blood test that can be collected starting at 10 weeks of pregnancy. The test works by finding the fetal DNA in your blood and testing that DNA for Down syndrome, trisomy 13 and trisomy 18. The laboratories can also test and give information about the baby's gender. Please note that this test **does not** replace CVS or amniocentesis. However, this test has been shown to be over 99% accurate for Down syndrome and also accurate for trisomies 13 and 18. **Currently, this test is offered to women who are 35 years old or older.** If this test does suggest a chromosome problem, a CVS test or amniocentesis test would still be recommended to confirm whether or not the blood test is correct.

Genetic counseling is required to receive this test. The genetic counselor will let you know which test is the best option for you. Please note that genetic counseling may be out of network. The maximum amount that you would be responsible for is $150.00.

□ I want Non-invasive Prenatal Screening

Amniocentesis:

This is a highly accurate diagnostic test. It is usually performed between 16 and 22 weeks of pregnancy. The amniocentesis is performed by inserting a small needle through your abdomen into the uterus under ultrasound guidance to collect about 2 tablespoons of amniotic fluid. However, the test does carry a risk for a complication which includes miscarriage of pregnancy. The risk of this procedure is up to 1 in 200 (0.5%). All patients undergoing an amniocentesis test are encouraged to meet with a genetic counselor prior to the procedure.

□ I want Amniocentesis:

Chorionic Villus sampling (CVS):

This is another accurate diagnostic test that gives similar information to amniocentesis. However, this test is done between 10-13 weeks of pregnancy. This test is done by either inserting a needle through the women's abdomen or through the cervix to collect some cells from the placenta. How the test is done is dependent on where the placenta is located. The risk of this procedure for complications including miscarriage is up to 1 in 100 (1%). By obtaining the placental cells, you can get accurate information about the baby's chromosomes. All patients undergoing an amniocentesis test are encouraged to meet with a genetic counselor prior to the procedure.

□ I 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.