Greenbrier Obstetrics and Gynecology, P.C.

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Carrier Screening in Pregnancy for Common Genetic Diseases:

Cystic Fibrosis, Spinal Muscular Atrophy, and Fragile X are a few common serious disorders that can occur even without a family history. These three tests are one time only simple blood tests that screen to determine if you are a carrier of the specific gene. A carrier is a person who has a gene that increases the risk to have children with a specific genetic disease. People do not know they are carriers until they have a blood test or an affected child. A negative test result significantly lowers, but does not completely eliminate the risk of being a carrier. Carrier testing is not able to detect all the genetic abnormalities that cause a particular disease.

These tests can be done either when you are planning a pregnancy or after you have become pregnant. The details of each genetic disease is listed below and are seen in all ethnicities and considered the most common.

Cystic Fibrosis (CF – the most common inherited disease of children and young adults): CF is a disorder of mucus production and produces abnormally thick mucus leading to life threatening lung infections, digestion problems, poor growth and more. Symptoms range from mild to severe. The carrier frequency is 1 in 24 to 1 in 97 and both parents need to be carriers for a child to be affected (25% chance). One in 3,500 children born are affected. *Recommended follow up to a positive result: test partner.*

Spinal Muscular Atrophy (SMA – the most common inherited cause of early childhood death): SMA is a progressive degeneration of lower motor neurons. Muscle weakness is the most common type with respiratory failure by the age of 2 years old. Muscles responsible for crawling, walking, swallowing and head and neck control are most severely affected. The carrier frequency is 1 in 47 to 1 in 42 in the US and both parents need to be carriers for a child to be affected (25% chance). One in 11,000 children born are affected. *Recommended follow up to a positive result: test partner.*

Fragile X Syndrome (the most common inherited cause of developmental delays): Unlike CF and SMA, this is an x-linked genetic disease and only carried in the mom. Unfortunately, 1 in 250 females are carriers and a child has a 50% chance of being affected if this is the case. Additionally, 1 in 4,000 boys are affected with Fragile X and 1 in 8,000 girls are affected. Approximately 1/3 of all children born with Fragile X also have autism and hyperactivity. *Recommended follow up to a positive result: genetic counseling and prenatal diagnosis.*

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CONSENT FOR CYSTIC FIBROSIS, SPINAL MUSCULAR ATROPHY, AND FRAGILE X

My signature below indicates that I have read, or had read to me, the information on **Greenbrier Obstetrics and Gynecology, P.C.: Carrier Screening in Pregnancy for Common Genetic Diseases** form and I understand it. Before signing this consent form, I have had the opportunity to discuss carrier testing further with my doctor, someone my doctor has designated, or with a genetics professional. I have all the information I want, and all of my questions have been answered.

INSURANCE COVERAGE: Coverage of these tests is subject to copays, coinsurance, and deductibles. Certain insurances may require pre-authorization and criteria to be met.

I have decided that:

☐ I want CF, SMA, and Fragile X carrier testing.

☐ I **do not** want CF, SMA, and Fragile X carrier testing.

_____ _____

Patient's Name Printed Date

_____ _____

Patient's Signature Witness

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Available Genetic Testing: Genetic tests drawn in our office are described below. Other testing is available at outside locations if desired.

MaterniT21: This is a simple, safe, and accurate non-invasive prenatal blood test for fetal chromosomal abnormalities, Trisomy 13, Trisomy 18, Trisomy 21, and several sex chromosome abnormalities. It is intended for: average risk pregnancies, as well as pregnancies at increased risk for fetal chromosomal abnormalities due to advanced maternal age (patients currently age 35 and up or age 35 at the time of delivery), fetal ultrasound abnormality suggestive of aneuploidy, personal or family history of chromosomal abnormalities, or abnormal serum screening test. The test also offers an optional analysis for fetal sex. This test can be performed as early as 9 weeks of pregnancy. Detection rate for Trisomy 21 is greater than 99.1%. An additional blood test is done at 16- 21 weeks for open spina bifida.

Tetra Screening: This is a blood test which can identify a patient who may be at an increased risk of having a baby with Down Syndrome, Trisomy 18, or an open neural tube defect. It is done between 15-21 weeks of your pregnancy, though the optimal time is between 16-18 weeks. Detection rate is 81% with a false positive rate of 5%.

MSAFP: This screening test is called the maternal serum alpha-fetoprotein test or MSAFP for short. It is a blood test drawn from the mother during the second trimester. It assists in assessing the risk of a fetus having a neural tube defect, like spina bifida or a chromosomal abnormality, like Down Syndrome.

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CONSENT FOR AVAILABLE GENETIC TESTING

My signature below indicates that I have read, or had read to me, the information on **Greenbrier Obstetrics and Gynecology, P.C.: Available Genetic Testing** form and I understand it. I have also read or had explained to me the specific disease(s) or condition(s) tested for, and the specific test(s) I am having, including the test descriptions, principles and limitations. I have had the opportunity to discuss the purposes and possible risks of this testing with my doctor or someone my doctor has designated. I know that genetic counseling is available to me before and after the testing. I have all the information I want and all of my questions have been answered.

INSURANCE COVERAGE: Coverage of these tests is subject to copays, coinsurance, and deductibles. Certain insurances may require pre-authorization and criteria to be met.

I choose to be tested for:

- ☐ MaterniT21
- ☐ Tetra screening
- ☐ MSAFP

☐ I decline all genetic testing.

_____ _____

Patient's Name Printed Date

_____ _____

Patient's Signature Witness