SEVEN HILLS
Women's Health Centers

Understanding Prenatal Laboratory Testing

To accept or decline testing please ask the medical assistant for Form 180 ALL – Signature Page for Consent for All Prenatal Testing

We will be happy to answer any questions or concerns you may have.

Congratulations on your Pregnancy!

The information in this booklet is intended solely as a general education aid. It is not a substitute for medical advice from a qualified health care provider. Please discuss any questions you have about information in this booklet with your SHWHC healthcare provider.

Routine laboratory tests are performed during each pregnancy to assess you and your baby's health.

Blood & RH type – Your blood type is determined by 2 factors:

1. Blood group – O, A, B, or AB
2. Rhesus (Rh) status is either positive (+) or negative (-). When a woman is Rh (-) and the baby's father is Rh (+), Rh Immunoglobulin injections are given to prevent antibodies from forming that could harm this or future babies.

Complete Blood Count test measures several components and features of your blood, including:

- Red blood cells, which carry oxygen
- White blood cells, which fight infection
- Hemoglobin, the oxygen-carrying protein in red blood cells
- Hematocrit, the proportion of red blood cells to the fluid component, or plasma, in your blood
- Platelets, which help with blood clotting

Rubella test is used to detect antibodies in the blood that develop in response to a rubella infection or immunization. Pregnant women, who are not immune to rubella, are at risk for developing the infection and having a baby with birth defects.

Infections:

Hepatitis B and hepatitis C viruses infect the liver. Pregnant women who are infected with hepatitis B or hepatitis C virus can pass the virus to their babies. All pregnant women are tested for hepatitis B virus infection. If you have risk factors, you also may be tested for the hepatitis C virus.

Syphilis, chlamydia, and gonorrhea can cause complications for you and your baby. If you have either of these STIs, you will be treated during pregnancy and tested again to see if the treatment has worked.

You may also be tested for chickenpox, toxoplasmosis, and tuberculosis.

HIV attacks cells of the body's immune system and causes ***acquired immunodeficiency syndrome (AIDS)***. If you are pregnant and infected with HIV, you can be given medication and take other steps that can greatly reduce the risk of passing it to your baby.

Urine culture tests the urine for bacteria, which can be a sign of urinary tract infections.

Urine toxicology confirmation tests the urine for various substances.

Glucose Screening Test screening for gestational diabetes (24-28 wks. gestation)

Group B Strep vaginal culture (35-36 wks. gestation)

Optional Testing…. Genetic Carrier Screening

Cystic Fibrosis Carrier Testing

This information was prepared to inform you about Cystic Fibrosis (CF) and CF carrier testing.

CF is a genetic disease that is usually diagnosed in childhood. It causes problems with breathing and digestion. It does not affect intelligence or appearance. Most children with CF need to take medication daily for digestive problems, as well as needing respiratory therapy every day. Lung infections are common and may require frequent antibiotics and/or hospitalizations. Not

all individuals with CF have the same problems. For reasons that are not well understood, some individuals are more severely affected than others. It is often not possible to tell from a prenatal test how severe a child's symptoms will be. In general, people living with CF have a shortened lifespan; some die in childhood, while others may live into their 40's or longer. By adulthood, most people with CF will have some health problems, but many are able to attend school, have careers, and have fulfilling lives. Although there is no cure for CF, there are treatments for symptoms and there is ongoing research on more effective treatments.

The purpose of CF carrier testing is to see if a couple is at an increased risk for having a child with CF. Carrier testing is performed on the mother, and if positive then the father would be tested. If the test shows that a couple is at high risk, additional testing can be done on the developing baby to see whether or not it will have CF. CF cannot be treated before birth. The purpose of testing is to prepare couples for taking care of a child with special health care needs or to help in making decisions about continuing with the pregnancy.

In order for a baby to have CF, it must inherit an abnormal copy of the CF gene from both parents. If a person has only one abnormal copy of the CF gene, that person is a carrier. A carrier does not have CF. When both partners in a couple are carriers, any child that they have has a 1 in 4 (25%) chance to inherit the abnormal copy of the gene from each parent. A child with two abnormal copies of the CF gene will develop CF.

You could be a carrier of CF even if no one in your family has CF and even if you already have children without CF; about 1 out of 30 Caucasians carries the CF gene. If your family background is not Caucasian, your chance of being a carrier is less than 1 in 30. For example, Hispanics have a risk of 1 in 46, African Americans 1 in 65, and Asian Americans 1 in 90. If you have a relative with CF or who is known to be a carrier of CF, your chance of being a carrier is greater based on your family history than your ethnic background. If testing shows that you are a carrier, the result is definite and will not change. However, if you are a carrier and have a new partner for a future pregnancy, testing should be considered for that new partner.

There are limitations of CF testing. There are some mutations in the CF gene that our current testing cannot find. **This means that your test result could be normal and you could still be a carrier**. However, these unknown CF mutations are rare, and the likelihood that you are a carrier even though you had a normal test result is very small. If your test shows that you are a carrier, the next step is to test the baby's father. If he has a normal test, the chance that your baby will have CF is very small. No further testing would be recommended in this situation. If both parents test positive, genetic counseling and further tests would be recommended to help decide if you want the baby tested for CF before birth. This could be done with amniocentesis, chorionic villus sampling or noninvasive prenatal testing.

Cystic Fibrosis Carrier Testing Summary

1. I understand that the decision to be tested for CF carrier status is voluntary
2. I understand that this test does not detect all carriers.
3. I understand that if I test positive for CF carrier status, testing the baby's father will be necessary to determine the baby's chance of having CF.
4. I understand that if one parent tests positive for CF and the other do not, it is still possible that the baby will have CF, but that the chance of this is very small.
5. I understand that if both the baby's father and me test positive for CF carrier status, additional testing can be done to determine whether or not the baby will have CF.
6. I understand that if the baby has inherited an abnormal CF gene from each parent, there is no treatment available to prevent the baby from developing CF.

Spinal Muscular Atrophy (SMA) Carrier Testing

SMA (spinal muscular atrophy) is a severe, often fatal, genetic disorder in which muscles involved in many essential functions, such as breathing, eating, and movement, become progressively weaker and ultimately waste away (atrophy) and die.

Almost one in every 40 people carries a mutated SMN1 gene and every year, about one in every 6,000-10,000 babies is born with SMA.

There are three types of SMA. The most common form, type I, which affects about 70% of patient, is the most severe. Children with type 1 SMA usually die from respiratory failure before the age of 2. In fact, SMA is the leading genetic cause of death in early childhood.

Children with type II SMA may be able to sit unaided, but cannot stand or walk unaided. These children typically live past age four. Although they may face many challenges, children with type III SMA are able to walk unaided and have a normal lifespan.

A child can only have the disease if both parents carry the mutated SMN1 gene. When both partners are carriers, there is a 25% (one in four) chance with each pregnancy of having a child with SMA. (Of course, this means that there is a 75% chance that each pregnancy will not result in a child with SMA.)

Fortunately for individuals planning a family, a simple blood test performed can determine with a very high degree of certainty whether you or your partner carries the mutation responsible for SMA.

It is imperative that couple have access to genetic counseling and patient education materials. SMA carrier testing should be both voluntary and confidential.

Fragile X Carrier Syndrome Carrier Screening

Note: For Patients with a Family History of Mental Disabilities or Autism Spectrum Disorders

Fragile X syndrome is the most common cause of inherited intellectual disabilities and is the most common single-cause of autism. Fragile X can be passed on in a family by individuals who have no apparent signs of this genetic condition. In some families a number of family members appear to be affected, whereas in other families a newly diagnosed individual may be the first family member to exhibit symptoms. Approximately 1 of 250 females carries the Fragile X pre mutation and are at risk of having children with Fragile X syndrome.

American College of Gynecology recommendations:

- Screening for Fragile X in women with a family history of Fragile X-related disorders
- Consider screening for women with ovarian insufficiency or failure or elevated follicle-stimulating hormone (FSH) level before the age of 40
- That screening should be offered to any women who requests Fragile X carrier screening, regardless of family history, after genetic counseling
- Offering prenatal testing by amniocentesis or CVS to known carriers of the permutation or full mutation

Fortunately for individuals planning a family, a simple blood test performed can determine with a very high degree of certainty whether you or your partner are carries of the mutation responsible for Fragile X.

It is imperative that couple have access to genetic counseling and patient education materials. Fragile X carrier testing should be both voluntary and confidential.

TAY-SACHS Carrier Testing

Note: For Patients of Ashkenazi Jewish, French Canadian, and Cajun Ethnic Groups

Tay-Sachs disease (TSD) is a fatal genetic disorder, most commonly occurring in children that result in progressive destruction of the nervous system. Tay-Sachs is caused by the absence of a vital enzyme called hexosaminidase-A (Hex-A). Without Hex-A, a fatty substance, or lipid, called GM2 ganglioside accumulates abnormally in cells, especially in the nerve cells of the brain. This ongoing accumulation causes progressive damage to the cells.

DNA-based carrier testing looks for specific mutations or changes in the gene that codes for Hex-A. Since 1985, when the Hex-A gene was isolated, more than 50 different mutations in this gene have been identified. Nevertheless, some mutations are not yet known. The current tests detect about 95 percent of carriers of Ashkenazi Jewish background and about 60 percent of carriers in the general population.

If both parents are carriers, they may want to consult with a genetic counselor for help in deciding whether to conceive or whether to have a fetus tested for Tay-Sachs. Extensive carrier testing of Ashkenazi Jews has significantly reduced the number of

Tay-Sachs children in this population group. Today most cases of Tay-Sachs disease occur in populations thought not to be at high risk.

Sickle Cell Disease (SCD)

Sickle Cell Disease (SCD) Sickle cell disease (also called SCD) is a condition in which the red blood cells in your body are shaped like a sickle (like the letter C). Red blood cells carry oxygen to the rest of your body. In a healthy person, red blood cells are round and flexible. They flow easily in the blood. A person with SCD has red blood cells that are stiff and can block blood flow. This can cause pain, infections and, sometimes, organ damage and strokes.

In the United States, SCD is most common among blacks and Hispanics. SCD affects about 1 in 500 black births and about 1 in 36,000 Hispanic births in this country. SCD is also common among people with family from Africa, the Caribbean, Greece, India, Italy, Malta, Sardinia, Saudi Arabia, Turkey or South or Central America. If your baby is born with SCD, the baby may be generally healthy or the baby may need special care throughout his life.

What causes SCD?

SCD is inherited. This means it's passed from parent to child through genes. A gene is a part of your body's cells that stores instructions for the way your body grows and works.

Genes come in pairs—you get one of each pair from each parent.

Sometimes the instructions in genes change, this is called a gene change or a mutation, the parents can pass gene changes to their children. Sometimes a gene change can cause a gene to not work correctly. Sometimes it can cause birth defects or other health conditions. A birth defect is a health condition that is present in a baby at birth.

Your baby has to inherit a gene change for sickle cell from both parents to have SCD. If the baby inherits the gene change from just one parent, the baby has sickle cell trait. This means that the baby has the gene change for SCD, but the baby doesn't have SCD. When this happens, the baby is a carrier. A carrier has the gene change but doesn't have the condition. Sickle cell trait cannot become SCD. A few people with sickle cell trait show signs of SCD.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

It is imperative that a couple have access to genetic counseling and patient education materials. SCD carrier testing should be both voluntary and confidential.

Optional Testing…. For Detection of Fetal Abnormalities

Maternal Serum Screening for Birth Defects

Several non-invasive tests are now available to give pregnant women information about the risks of having a baby with certain birth defects. The purpose of this document is to explain the options available to you. You will be requested to indicate whether you request or decline these tests.

The conditions evaluated by these tests include Down syndrome, Trisomy 13, Trisomy 18, and spina bifida. Down syndrome occurs when the baby has an extra copy of chromosome 21. Babies with Down syndrome have mental retardation and usually have physical abnormalities, like heart defects. Trisomy 18 is caused by an extra chromosome 18 and trisomy 13 by an extra chromosome 13. Both of these disorders cause profound mental retardation and multiple birth defects. While both Trisomy 18 and Trisomy 13 are more severe than Down syndrome, they are less common. The risk of having a baby with these problems increases

with a mother's age, but can occur at any age. Spina bifida or a neural tube defect is a problem caused by abnormal development of the neural tube (part of the embryo that forms the spine and brain). These defects can range from an open defect leading to possible paralysis and/or mental retardation to anencephaly or incomplete development of the skull and brain leading to death after birth. About one in every 600 area babies are born with a neural tube defect.

(1) The Early Screen (11-13 weeks)

This is a blood test combined with an 11-13 week ultrasound exam that tells whether you have an increased chance of having a baby with Down syndrome, Trisomy 13, or Trisomy 18. The test involves taking blood from your arm or through a finger stick to measure the levels of two proteins (free Beta and PAPP-A) in your blood. An ultrasound is also performed to measure your baby's nuchal translucency, a fluid filled space at the back of the baby's neck and may examine the nasal bone. This test will identify over 91% of Down syndrome, 97% of Trisomy 18/13, and 40% of heart defects. A negative screen test means that the patient is not at high risk for having a baby with Down syndrome or Trisomy 18/13. A screen negative result does not completely rule out the possibility of a pregnancy with these genetic problems. About 5% (1 in 20) women who take the test are positive and have a greater risk than 1 in 200. Most of these women have normal pregnancies, but these women are offered further diagnostic testing including amniocentesis (detection rates greater than 99%).

This test does not allow for the detection of spina bifida or neural tube defects; however this can be evaluated in the second trimester by ultrasound or by another blood test (AFP). There is no risk to the pregnancy in the performance of this test.

(2) The Quad Screen (15-20 weeks)

This is a blood test that measures four substances (AFP, HCG, estriol, and inhibin A) produced by a developing fetus and passed into the mother's blood. This test is performed between 15-20 weeks of pregnancy. An abnormal Quad Screen may indicate:

a. (a) the pregnancy is either at an earlier or later stage than believed

b. (b) the mother is carrying more than one baby

c. (c) the baby being carried may have a birth defect

d. (d) the baby being carried may have a chromosomal defect

e. (e) the baby being carried is normal and for some unknown reason the test is falsely abnormal

The most common birth defect detected by the Quad Screen is spina bifida / neural tube defect. The test will detect about 75% of all pregnancies with a neural tube defect. Other birth defects that may be detected include abdominal wall defects, intestinal obstructions, and certain kidney problems.

The most common genetic defect detected by the Quad Screen is Down syndrome (Trisomy 21). The test will detect about 80% of all pregnancies with Down syndrome and 60-80% of Trisomy 18 pregnancies.

Approximately 5-6% of Quad Screen tests have an abnormal value. An abnormal test does not necessarily indicate the presence of an abnormal fetus. Conversely, a normal test does not guarantee a normal baby. There is no risk to the pregnancy in the performance of the Quad Screen.

(3) Cell Free Fetal DNA Test also known as NIPT (10 - 40 weeks)

*Note: Optional for Patients of any Age but Recommended for Patients 35 Years and Older or with Other Risk Factors; for patients whose **BMI is greater than 29** it is recommended to perform this test after 12 weeks gestation.*

Noninvasive prenatal testing that uses cell free fetal DNA from the plasma of pregnant women offers tremendous potential as a screening tool for fetal aneuploidy. Circulating cell free fetal DNA, which comprises approximately 3–13% of the total cell free maternal DNA, is thought to be derived primarily from the placenta, and is cleared from the maternal blood within hours after childbirth (1). Recently, cell free fetal DNA analysis has become clinically available for women at increased risk of fetal aneuploidy.

Aneuploidy is the disorders of chromosome number in which the number of chromosomes is above or below the normal (46). Common forms of aneuploidy are **trisomy** in which there is one extra chromosome and monosomy in which there is one less, than the normal 46.

Indications for Considering the Use of Cell Free Fetal DNA

- Maternal age 35 years or older at delivery
- Fetal ultrasonographic findings indicating an increased risk of aneuploidy
- History of a prior pregnancy with a trisomy
- Positive test result for aneuploidy, including first trimester, sequential, or integrated screen, or a quadruple screen.
- Parental balanced robertsonian translocation with increased risk of fetal trisomy 13 or trisomy 21.

Conclusions:

- A negative cell free fetal DNA test result does not ensure an unaffected pregnancy.
- A patient with a positive test result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results.
- Cell free fetal DNA does not replace the accuracy and diagnostic precision of prenatal diagnosis with CVS or amniocentesis, which remain an option for women.

Summary: Please understand that these tests are not mandatory. It is entirely your decision as to whether you want to have either of these tests done. Neither of the test discussed are used for diagnostic purposes – they provide estimates of risk only.

- If either test is abnormal, further testing will be offered and may include a second blood test, ultrasound, or amniocentesis.
- You are encouraged to contact your insurance company for coverage information.
- Should you have any questions about this test at any time, I urge you to call your physician's office. We will be happy to answer any questions you may have.

ACTION to be taken by the patient:

- **If you are interested in the Early Screen, please contact our office prior to 11 weeks gestation to schedule testing**
- **If you are interested in the Quad Screen, please contact our office prior to 15 weeks gestation to schedule testing**
- **If you are interested in the Cell Free Fetal DNA Test , please contact our office any time after 10-12 weeks gestation to schedule testing**
- **You should check with your insurance company if you are interested in having any optional testing**
- **To accept or decline optional testing please ask the medical assistant for Form 180 ALL Signature Page for Consent for All Prenatal Testing**

We will be happy to answer any questions or concerns you may have.