GENETIC TESTING OPTIONS

Most babies are born free of birth defects. Of those who are affected, the more common types are Trisomy defects, such as Down syndrome, and Open Neural Tube defects such as Spina Bifida. Testing is optional. Some people want genetic testing done as early as possible, to reassure them that their baby is normal, or to provide a diagnosis early enough in the pregnancy so that all options remain open. Others, who would not change their pregnancy plans in the event of a birth defect, seek to know whether the baby is normal or affected, and if there is a birth defect, to use the remainder of the pregnancy to educate themselves and prepare for a family member with special needs. It is also helpful for your doctor to be prepared for all eventualities. Still others prefer not to undergo any genetic testing. Our goal is to educate you about the options so that you can make a fully informed decision, as well as to support your decision.

SCREENING TESTS

A screening test is NOT the same as a diagnostic test. The screening process is basically a customized statistical risk assessment, to determine your personal risk. A positive screening test is NOT a diagnosis of a birth defect. It provides information that guides decisions about diagnostic testing.

First Look Test (typically scheduled between 12wks-13wks 3days)

- **The First Look Test** is offered at Emerson Hospital MFM by Brigham & Women's perinatologists and at Brigham & Women's Hospital. It is a non-invasive test that assesses whether you are at increased risk of having a baby with Down syndrome or Trisomy 18. It is a combination of an early ultrasound to measure the nuchal translucency (NT) thickness (neck fold) and maternal serum screening (blood work).

This screening test is often covered for pregnant women who desire to know their risk of having an affected pregnancy. Please schedule your appointment and then check with your insurance carrier to see if this testing is a covered benefit for your insurance plan. The CPT codes used to bill for this testing are: 76813: nuchal translucency (NT scan done at Emerson Hospital) and the following maternal serum screening bloodwork which is sent to Esoterix Integrated Genetics a Division of LabCorp: 84702: HcG (human chorionic gonadotropin) 84163: Papp-A (pregnancy plasma protein A) You can always call to cancel your appointment at any time if necessary or if you simply change your mind.

Non Invasive Prenatal Testing (NIPT) - Cell Free DNA (cfDNA)

Cell free DNA testing (cfDNA) is a blood test that screens for the likelihood of aneuploidy of chromosomes 13, 18, 21, X & Y with a high degree of accuracy. Aneuploidy is an abnormal number of chromosomes. Aneuploidy is associated with Down syndrome (also known as trisomy 21 – 3 copies of chromosome 21) and other birth defects.

cfDNA does not screen for neural tube defects, heart defects or abdominal wall defects which are screened for with ultrasound or for other chromosomal & genetic disorders.

This testing is scheduled in conjunction with the NT scan (First Look, see above) at which point the testing will be ordered and drawn for you the day of your appointment. cfDNA testing does not replace the NT scan or comprehensive 2nd trimester ultrasound (fetal structural survey).

cfDNA may or may not be covered by your health insurance. Emerson Hospital MFM commonly uses Myriad Women's Health Laboratories for cfDNA testing (CPT code 81420) but other specialty labs may utilized depending on your specific pregnancy and insurance. Prior authorizations are obtained by Myriad on your behalf (if your insurance requires it) before processing your specimen. The genetic counselor or perinatologist will also discuss possible testing cost at the time of your appointment.

AFP4 Quad Screen (Alpha-Feto-Protein) 16-20 weeks

A blood test to determine if a mother is at increased risk of having a child with certain birth defects including: Open neural tube defects (i.e. Spina Bifida, anencephaly), Trisomy 21 (Down syndrome), and Trisomy 18 (Edwards syndrome). **This blood test is offered to everyone at their 16 week prenatal appointment at Concord OB/GYN.**

Level 2 Ultrasound (2nd trimester)

Some women with certain personal or family medical histories that might affect the baby or management of their pregnancy will be scheduled for a Level 2 comprehensive fetal structural survey. This ultrasound is a targeted ultrasound performed by a Brigham & Women's perinatologist at Emerson MFM in which the baby's anatomy is measured and evaluated, including brain, limbs, abdomen, heart, stomach, kidneys, and placenta. The baby's sex can often be determined at this time. This ultrasound can detect potential problems, but cannot detect every possible problem.

*Typically scheduled at Emerson Hospital MFM, but can also be scheduled at Brigham & Women's Hospital.

DIAGNOSTIC TESTS

Diagnostic tests are offered when a screening test is positive. Some women who will be age 35 or older at the time of delivery, or who have strong risk factors such as prior children or family members with birth defects, choose to pursue diagnostic testing. Others prefer to wait for screening test results before deciding. If you are interested in diagnostic testing a consultation with a genetic counselor at Maternal Fetal Medicine will be scheduled for you to go over your history and options for testing. These tests carry a statistically small risk to the baby which is discussed in full before the test is performed.

CVS Testing (Chorionic Villi Sampling) 11-13weeks

Guided by ultrasound a small catheter is inserted through the abdomen or vagina. A very small sampling of chorionic villi (tiny parts of the placenta) are taken and analyzed for genetic defects producing a definitive result, and the baby's sex is also determined.

*Typically scheduled at Brigham & Women's Hospital.

Amniocentesis 16-20weeks

Guided by ultrasound a needle is inserted into the abdomen. A sample of amniotic fluid is withdrawn. The cells in the fluid are analyzed for genetic defects producing a definitive result, and the baby's sex is also determined.

*Typically scheduled at Emerson Hospital MFM, but can also be scheduled at Brigham & Women's Hospital.

Please see or contact **KRISTIN**
(Outside Scheduling) at **978-371-0302 x204** to schedule your testing appointments

GENETIC CARRIER SCREENING

Genetic Carrier Screening is done by a simple blood draw and will determine if you carry the genes for certain genetic disorders. Some genetic disorders are more common among people of specific racial and ethnic heritage. These tests will help to identify if you are at risk of having a child affected by the most commonly screened inherited genetic disorders. This information allows you to make informed reproductive choices.

The following genetic carrier screening panel may be offered to you today

- **Cystic Fibrosis (CF)** - A blood test can determine if you are a carrier of CF. Both parents must carry the gene for this disease in order for the baby have a (25%) chance of being born with it. People affected by CF are now starting to live longer into their 30's, but it is still considered a serious disease. It is a life-threatening disorder that causes severe, progressive lung damage and nutritional deficiencies.
- **Spinal Muscular Atrophy (SMA) –** Both parents must be carriers in order for the baby to have a (25%) chance of being born with it. Carrier status can be detected by a blood test. Spinal Muscular Atrophy is a neuromuscular disorder that causes severe muscle weakness and progressive loss of voluntary muscle control. It is a life shortening disorder.
- **Fragile X –** Fragile X is the most common cause of inherited intellectual disability. A blood test can determine if you are carrier of Fragile X. Fragile X syndrome in its early stages can include delayed speech & developmental milestones which progress into moderate intellectual disability. Fragile X is caused by mutations in the fragile X gene located on the X chromosome and typically passed on from carrier mothers and more commonly affect boys. There are usually no life threatening health concerns associated with fragile X syndrome and treatment involves early intervention & management of symptoms.
- **Tay Sachs Disease** – Both parents must be carriers in order for the baby to have a (25%) chance of being born with it. Tay Sachs is caused by an enzyme deficiency that allows a harmful substance to build up in the brain, causing mental and physical deterioration, and it is generally fatal by age 5. This is more commonly found among people of Ashekenazic (Eastern European) Jewish, French Canadian or Louisiana Cajun descent. People of Ashekenazic descent are also at increased risk for a number of other, "related" genetic disorders.

The nurse may also recommend a more broad & comprehensive carrier screening panel based on your family history and ethnicity.

Concord OBGYN uses Invitae Laboratories for personal genetic carrier screening. Carrier screening can be drawn by blood sample in the lab and also by saliva kit that can be mailed to your home! After signing the consent form, you will asked to provide your email address. Invitae will do a benefits investigation with your specific insurance plan on your behalf and send an email to you with detailed information about the cost if processed through your insurance vs their self-pay option. Some plans cover genetic testing fully and other plans may be subject to deductibles and coinsurances. **Please be on the lookout for an email from Invitae within a 7 days of having your bloodwork drawn or shipping your saliva kit back to Invitae to receive your benefits investigation and choose your preference of insurance vs patient pay or to cancel your testing if you so choose.** Prior authorizations are obtained by Invitae on your behalf (if your insurance requires it) before processing your specimen.