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. 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.

Women who are interested in cfDNA are required to meet with the genetic counselor to discuss your history and the scope and limitations of the testing before the test is performed. This counseling 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 testing is recommended for women with the following risk factors:

* 35 years or older at the time of delivery
* Aneuploidy in a previous pregnancy
* Abnormal result from First Look Test (serum screening, NT scan)
* Certain abnormalities found on ultrasound

If you do not have one of the aforementioned risk factors, but would like to discuss the testing with a genetic counselor, an appointment to meet with them can be set up for you at the time of your First Look test – please be sure to mention this to either the nurse at your visit today or when calling Kristin to schedule the appointment.

cfDNA may or may not be covered by your health insurance. Emerson Hospital MFM currently use Myriad Women's Health Laboratories for cfDNA testing. Prior authorizations are obtained by Myriad on your behalf (if your insurance requires it) before processing your specimen. The genetic counselor will also discuss possible testing cost at the time of your genetic counseling 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