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 NT Scan (scheduled between 12wks-13wks 3days)

- **The NT Scan (also referred to as First Look or First Screen)** is offered at Emerson Hospital's Maternal Fetal Medicine Department (MFM) by Brigham & Women's perinatologists. It is a non-invasive test that assesses whether you are at increased risk of having a baby with Down syndrome (Trisomy 21), Trisomy 18 & Trisomy 13. It is a combination of an early ultrasound to measure the nuchal translucency (NT) thickness (neck fold) and the NIPT (Non Invasive Prenatal Testing) bloodwork (see description below).
- 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 code used to bill for the NT Scan portion of the testing is: 76813: nuchal translucency and is performed at Emerson Hospital.

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

NIPT (Non Invasive Prenatal Testing) also known as Cell free DNA testing (cfDNA) and 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.

NIPT 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. NIPT testing does not replace the NT scan or comprehensive 2nd trimester ultrasound (fetal structural survey).

NIPT testing may or may not be covered by your health insurance. Emerson Hospital MFM commonly uses Myriad Women's Health Laboratories for NIPT testing (CPT code 81420) but other specialty labs may utilized depending on your specific insurance. Prior authorizations are obtained by Myriad (or other specialty lab used) 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.

You may call to cancel your appointment at any time if necessary or if you simply change your mind.

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.

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 contact **KRISTIN**
at **978-371-0302 x204** to schedule your testing appointments