# 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, 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 or chromosomal anomaly, seek to know whether the baby is affected 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.

## CHROMOSOMAL SCREENING TESTS

**What is chromosome screening?**

Everyone is offered the chance to determine whether their pregnancy has an atypical chromosome makeup. Chromosomes are large structures that contain DNA. Pieces of DNA within chromosomes are called genes. A human cell typically has 46 chromosomes in 23 pairs. An example of a condition caused by a difference in chromosome number is Down syndrome, which is due to an extra copy of chromosome 21. Some chromosome conditions can impact children's health and learning. Typically, chromosome conditions occur by chance and are not inherited. There is typically NOT a family history of a chromosome condition. A screening test helps to determine your estimated risk for a chromosome condition and is NOT the same as a diagnostic test. A positive screening test is NOT a diagnosis of a birth defect. It provides information that guides decisions about diagnostic testing.

**What chromosome screening tests are available?**

**1st trimester NT Scan & NIPT**

**The NT Scan** is scheduled between 12 weeks - 13 weeks & 3 days of pregnancy with Dr. Tamara Takoudes of Maternal Fetal Medicine here in our Concord office. This is an ultrasound performed at a specific time in pregnancy to measure the nuchal translucency (thickness of the neck fold). This visit is done in conjunction with NIPT (Non Invasive Prenatal Testing) bloodwork (see description below).

**Non Invasive Prenatal Testing (NIPT)**

NIPT (Non Invasive Prenatal Testing) also known as 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. Sex chromosome screening & testing is optional and can identify conditions such as Turner syndrome and Klinefelter syndrome. NIPT detects pieces of the genetic code, called DNA, that come from the placenta and are in the patient's blood. This testing is performed after 10.5 weeks of pregnancy. NIPT <u>does not</u> 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 (see above) at which point the testing will be ordered and drawn for you the day of your appointment. NIPT testing <u>does not</u> replace the NT scan or comprehensive 2nd trimester ultrasound (fetal structural survey).

These screening tests are 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. Concord OBGYN offers Panorama NIPT testing through Natera (CPT code 81420). Prior authorizations are often required by insurance companies and obtained on your behalf by Natera before processing your specimen. Natera will reach out to you directly via text or email if your estimated cost through your insurance exceeds their self-pay price for testing ($249.00). You will have the option to choose to put the claim through your insurance, or to self-pay. Mass Health patients do not receive a bill for NIPT testing.

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 Dr. Tamara Takoudes of Boston MFM in our Concord office on Wednesdays 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 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 Dr. Takoudes 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. Diagnostic tests can detect trisomies and syndromes such as Cri-du-Chat syndrome, Williams's syndrome, and some associated with autism or intellectual disability.

**What diagnostic testing is available?**

## 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 can also definitively be determined.

## 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 can also be definitively determined.

Both CVS & Amniocentesis are performed by Dr. Tamara Takoudes of Boston MFM in our Concord office on Wednesdays.

**What can diagnostic tests detect?**

- A karyotype can detect an extra or missing copy or large piece of any chromosome including chromosomes 13, 18, 21, X and Y.
- A microarray can detect extra or missing pieces of chromosomes too small to find with NIPT or karyotype.

**Which test is right for me?**

Your providers at Concord OBGYN can discuss your options with you to help you make the best decision for you. Some questions to consider when making your decision:

- Would you want to know if the pregnancy has a chromosomal condition?
- Would you prefer a screening test (no risk to the pregnancy with results that reduce risks for some common chromosome conditions) or a diagnostic test (small procedure related risks with results providing a definitive answer about many chromosome conditions)?

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