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## Genetic Testing Options for Birth Defects In the First and Second Trimester

Many genetic testing options are available today for pregnant women to look for certain fetal health problems. There are screening and diagnostic tests available at certain gestational stages in your pregnancy. Down syndrome (trisomy 21), trisomy 13 & trisomy 18 are the most common chromosome problems that cause birth defects. First and second trimester **screening tests** only indicate your **risk** of carrying a baby with one of these conditions. Diagnostic testing will be recommended if you have a positive screening test.

### First Trimester Screening:

- **Nuchal Translucency** done between 11 6/7 weeks – 13 6/7 weeks, which includes a sonogram combined with a maternal blood test to determine fetal risk of Down syndrome as well as trisomy 18. These are chromosomal abnormalities caused by extra copies of chromosomes. When an extra chromosome is present, the fetus may have an increased amount of fluid at the base of their neck, referred to as the nuchal fold. The extra fluid can make the neck larger. The sonogram will measure the size of the nuchal fold. This measurement, combined with your age and the results of the blood test will provide a statistic of the likelihood of a chromosomal disorder.
- **NIPT** (non-invasive prenatal testing) is a maternal blood sample for fetal DNA testing, and can be done as early as 10 weeks or at any time before delivery. This testing screens for Down syndrome, trisomy 13 & trisomy 18 and may detect other abnormalities including triploidy and microdeletion. NIPT also detects the baby's gender. **This is available to women who have tested positive for their first trimester screening, have a family history of genetic disorders, or for women who are 35 and older.** If you meet any of these criteria, the nuchal translucency and NIPT testing may be done together at the same time for your convenience. You may be referred to a specialist for this test.

### Second Trimester Screening:

- **AFP Maternal Serum Screen 4 (MSS4)** is a blood test done between 15 – 19 weeks. This test screens for Down syndrome, trisomy 18 (not trisomy 13), and this test may also detect neural tube defects, such as spina bifida. This test detects 80% of abnormalities, and may miss 20%. It has a false positive rate of 10%; meaning the test detects an abnormality, but there is not.
- **Comprehensive Sonogram** at 20 weeks may be able to detect structural abnormalities of fetal growth and development, including the brain, heart, spine, kidneys and gastrointestinal tract. This screen detects 50% of chromosomal abnormalities.

## **Diagnostic Testing:**

These are invasive genetic tests and are usually performed at a Maternal Fetal specialist office if you have had any positive screening results. Diagnostic testing will determine if your baby will be born with a chromosomal abnormality and certain other conditions.

- **Chorionic Villa Sampling (CVS)** is an invasive genetic test done in the first trimester which involves tissue sampling from your placenta. CVS can determine certain chromosomal conditions, such as Down syndrome and some other conditions, such as cystic fibrosis. CVS poses a slight risk of miscarriage.
- **Amniocentesis** is performed in the second trimester around 17 weeks and can diagnose chromosomal abnormalities such as Down syndrome and neural tube defects such as spina bifida. This is an invasive test where a needle is passed through your abdomen into the uterus to collect a small amount of amniotic fluid surrounding the baby. Like CVS testing there is a slight risk of miscarriage.

Both CVS and amniocentesis are performed by a Maternal Fetal specialist.

## **Frequently Asked Questions:**

- **What is Down syndrome and trisomy 13 & 18?**

A person is normally born with 23 pairs of chromosomes for a total of 46 chromosomes, one set from the mother and the other set from the father. When there is a missing or extra chromosome this is called aneuploidy, which causes birth defects such as Down syndrome and trisomy 18 and trisomy 13. Babies with Down syndrome have an extra chromosome 21 which causes varying degrees of mental retardation and other medical problems involving the heart, digestive tract and possibly other organs. Trisomy 18 and 13 cause profound mental retardation and more severe birth defects. Few babies with trisomy 13 & 18 survive longer than a few months. The chances of conceiving a baby with these conditions increase with a mother's age.

- **What type of test can be done in the first trimester?**

The first trimester **screening** test is called a **nuchal translucency (NT)**. This is not a diagnostic test, but it will tell you the probability of Down syndrome and trisomy 18. This probability is based on 3 criteria combined: your age, an ultrasound (sonogram) of the baby and blood work. These results can alert you that your baby is at an increased risk for one of these chromosomal disorders, or reassure you of a lower risk.

- **How is the nuchal translucency done and when will I know the results?**

This frequently can be done in our office depending on your insurance. The test is performed between 11 6/7 – 13 6/7 weeks of pregnancy with a sonogram and blood work at the same time. During the sonogram a measurement will be taken of the fluid underneath the skin along the back of the baby's neck, called the nuchal translucency (NT). Other aspects of your baby's anatomy will be checked during this sonogram as well, which may include nasal bones, limbs, heart, and other organs. A maternal blood sample will be sent to a specific lab to be analyzed. The results of these tests combined with your age related risk will provide you and your provider with the risk for Down syndrome and trisomy 18. You should receive these results in about a week to ten days.

- **How accurate is the first trimester screening?**

The nuchal translucency screening test can detect 90-94% of abnormalities when they are present, but also has a 5% false positive rate, meaning that the test will indicate an abnormality

when there is not. NIPT detects 98-100%, and misses fewer than 2% of abnormalities. It has a false positive rate of less than 1%.

- **What if the screening shows an increased risk for Down syndrome or trisomy 13 or 18?**

In the event of abnormal first trimester screening, you would be a candidate for **NIPT** screening test which is a more advanced blood test for chromosomal abnormalities. This test is explained under first trimester screening. You may also be referred for additional diagnostic testing.

**Important note below:**

**\*\*\*If a screening test shows an increased risk for one of these conditions, this does not mean that your baby has one of these conditions. You will be able to schedule an appointment with your provider to discuss your risks and options, as well as any need for genetic counseling and further testing. It's also important to know that a lower risk test result does not guarantee that your baby won't have one of these conditions. Only diagnostic testing, usually not performed in our office, can confirm these abnormalities. We will be able to assist you if you are referred for additional genetic counseling, screening and diagnostic tests. These are generally done by a Maternal Fetal specialist office, if necessary.**

- **How much does the test cost and/or is it covered by my insurance?**

Our office will check your insurance benefits to determine if you are eligible to have this testing done in our offices, or if you need to have the testing done elsewhere. Your insurance company will be billed for the testing, but is not guaranteed to be a covered benefit. It is your responsibility to contact your insurance company and verify your benefits and any out of pocket expense that you may incur. The CPT code for the nuchal translucency is 76813. The CPT code for the NIPT is 81420. The LabCorp NIPT is InformaSeq; Quest NIPT is QNatal Advanced.

- **How do I schedule the testing?**

You can call our office at (410)573-9530 to arrange your appointment or if you have questions regarding your screening options. Once you have informed our office that you are interested in genetic testing we will process a Special Order and a triage nurse will call you to schedule your appointment. It is important to remember that the nuchal translucency is a time sensitive test and is unable to be performed after 13 weeks 6 days gestational age. When scheduling a nuchal translucency test, you will be instructed to drink 24 ounces of water 1 hour before the test and hold your urine until the sonogram portion of the test has been completed.

- **What if I decide not to pursue genetic testing?**

You may decide that you do not desire any genetic testing because you are comfortable with the results, no matter what the outcome. It is important to discuss the risks and benefits of testing with your healthcare provider to evaluate if the benefits from the results could outweigh any risks from the procedure.

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