Patient Information Sheet
First Trimester Increased Nuchal Translucency

You have recently learned that your baby has an increased nuchal translucency. We expect that you may have questions about what this could mean for your developing baby. Hopefully, this information will address some of your concerns; however, we encourage you to speak to your genetic counselor or healthcare provider for more information. It is important to remember that anyone can have a baby with a birth defect. Birth defects occur in 2-5% of all births and are rarely caused by something that the parents did or did not do before or during pregnancy. Currently, there are no tests available to detect all health problems.

What is an increased nuchal translucency?

In every developing baby, as part of typical development, a small amount of fluid accumulates at the back of the neck, between the skin and the spine. When this pocket of fluid is observed during an ultrasound evaluation, it is called a nuchal translucency. Nuchal translucency measurements are generally taken between the 11^{th} and 14^{th} weeks of pregnancy. When there is more fluid than usual, it is referred to as an increased nuchal translucency.

How many babies are found with an increased nuchal translucency?

About 1 in every 100 to 1 in every 20 women (1-5%) who has an ultrasound between 11 and 14 weeks will be told her baby has an increased nuchal translucency. This number may be higher if you belong to a higher risk group, such as those women that are over the age of 35.

What problems could be associated with an increased nuchal translucency for my baby?

In many cases, an increased nuchal translucency occurs in typical pregnancies and in healthy babies. However, an increased nuchal translucency measurement is sometimes caused by an underlying problem with the baby's development. In general, the larger the nuchal measurement, the greater the chance that an abnormality is present.

A common cause of an increased nuchal translucency is a chromosome problem, such as Down syndrome or Turner syndrome. In order to understand a possible chromosomal cause or genetic syndrome cause, it is helpful to know a little about chromosomes. Chromosomes are packages of genetic information that carry the instructions (genes) necessary for our growth and development. Typically, there are 23 pairs of chromosomes in each cell of our bodies. One copy of each pair is from our mother and one copy of each is from our father. The first 22 pairs of chromosomes are the same in males and females. The last pair is known as the sex chromosomes and they determine our gender. Females typically have two X chromosomes and males usually have one X and one Y. It is important to have the correct amount of genetic material to have typical development. Sometimes there is an imbalance of genetic material that may cause birth defects. The imbalance may be caused by an extra or missing whole chromosome or sometimes only by extra or missing pieces of chromosomes. Some genetic conditions are caused by changes in one or more genes that result in a particular set of characteristics or features (also called a syndrome). A chromosome imbalance or genetic syndrome can produce birth defects and physical changes such as an increased nuchal translucency.

Other possible causes include birth defects and other rare genetic syndromes. The most common birth defects in babies with an increased nuchal translucency are heart defects. The likelihood of any of these problems depends on several factors, including the mother's age, the gestational age, the size of the nuchal translucency, previous pregnancy history, and family history.

In a minority of cases, babies with an increased nuchal translucency will have extra fluid in other parts of the body (called hydrops). This is usually a sign that the baby is not doing well.

What further testing may be offered and what will it tell me?

Depending on your current gestational age, a number of additional tests may be offered to you. If an increased nuchal translucency is found during a routine ultrasound, a more detailed ultrasound will usually be performed to look carefully at the baby's anatomy. A level II ultrasound may also be offered to you at approximately 20 weeks' gestation to look for any other physical features associated with an increased nuchal translucency.

There are two additional types of prenatal tests: screening tests and diagnostic tests. Screening tests can modify your risk for a chromosome abnormality such as Down syndrome and involve no risk of complications to your pregnancy. A screening test will not determine if your baby has Down syndrome, but it may help you decide whether or not you would like to pursue a diagnostic test. Diagnostic tests can evaluate the number and structure of your baby's chromosomes. They are 99% accurate in detecting extra or missing chromosomes. Unfortunately, all diagnostic tests have a risk of complications including the potential for pregnancy loss.

Screening Tests

There are multiple screening tests to determine the chances for a pregnancy to have a chromosome condition. These screening tests cannot diagnose or rule out the presence of a chromosome condition but may be used to help you decide whether or not you would like to pursue a diagnostic test.

Non-invasive prenatal testing, or NIPT, is a screening test for certain chromosome conditions. During pregnancy, some of your baby's chromosome material is in your blood, along with your own chromosomes. NIPT is a blood test that measures the amount of chromosome material in your blood to determine if your baby could have an extra chromosome, such as in Down syndrome where there is an extra 21st chromosome. This test also screens for two other more severe chromosome conditions, trisomy 13 and trisomy 18, and may screen for some less severe conditions that are caused by different numbers of the X or Y chromosome. NIPT is a highly accurate screen; however, it is not a diagnostic test. The detection rate for these chromosome conditions is typically between 90-99%.

Other screening blood tests (often called a triple, quad, penta, or first trimester screen) may also be offered. Your genetic counselor or health care provider can discuss these various screening options in more detail.

Diagnostic Tests

Depending on your gestational age, two diagnostic testing options may be available including chorionic villus sampling (CVS) or amniocentesis. These are diagnostic tests in which a small sample of placental tissue or amniotic fluid is obtained to examine the baby's chromosomes. Because these procedures are considered invasive procedures, there is a risk, likely less than 1%, for complications that can lead to miscarriage.

If you choose to pursue a CVS or amniocentesis, you may be offered an additional test called a chromosome microarray (CMA.) It is used to identify small missing or extra pieces of the baby's chromosomes that may be associated with genetic conditions. CMA cannot detect all genetic conditions and may detect variations in the chromosomes that have uncertain clinical significance. Parental blood samples may help to clarify the meaning of a variation, but effects of these changes may not be known until after the baby is born. CMA may also detect information such as non-paternity and close relationships between parents. In addition to CMA, you may also be offered testing for specific genetic syndromes to look for changes in one or more genes.

The risks, benefits, and limitations of screening and diagnostic testing options should be discussed with you by your genetic counselor or other health care provider. Follow-up counseling and referrals for support can be made if a chromosome condition is detected prenatally. As with all situations in which prenatal testing is discussed, it is your decision whether or not this test is done.

If no additional abnormalities are seen on ultrasound or echocardiogram, and if all chromosome studies, including CMA and possibly other single gene tests, are normal, the chance of a good outcome is as high and similar to general population. However, it is important to remember that prenatal screening and testing cannot detect or rule out all genetic conditions. After the baby is born, if additional birth defects or developmental delays are noted, an examination by a pediatric geneticist may be recommended. If a chromosome analysis was not performed by amniocentesis, the geneticist may recommend that a small amount of blood be taken from the baby to evaluate for chromosome or genetic problems. This examination and possible additional tests are important, since an accurate diagnosis will provide you with the best information regarding the prognosis for your baby, the cause of the ultrasound abnormality, the potential recurrence risks to future children and that of other family members, as well as to determine appropriate tests to be offered to you in future pregnancies.

What do I do now?

This information is only intended as an introduction to some of the terms and tests that you have already heard or will be hearing about from your genetic counselor and healthcare provider. We hope that this information sheet will be helpful as you begin to understand more about increased nuchal translucency. We understand that any time something of concern is found through prenatal screening and testing, parents are going to be worried. Please don't hesitate to contact your genetic counselor with any questions or concerns you have. We are here to help you and your baby.

The decision to have one or more of these tests or to do no additional testing is a difficult and personal one. There is no one right decision. Some people will decide to have no further testing because they do not feel they need to know if their baby will have any of these conditions before delivery. Other people feel that they want more information as soon as possible. After gathering all of the information you need about an increased nuchal translucency, you should make the decision that is right for you and your family. Please keep in mind that not all babies with an increased nuchal translucency will have the complications described in this information sheet. Though it is never possible to predict the long-term prognosis for a child with even an isolated increased nuchal translucency, many babies will be born without significant medical complications.