



Nuchal Translucency Screening

What is the Nuchal Translucency Screening Test?

This screening test uses ultrasound to measure the clear (“translucent”) space in the tissue at the back of your developing baby’s neck. This measurement can help your practitioner give you an assessment of your baby’s risk for Down Syndrome (DS) and other chromosomal abnormalities. Babies with abnormalities tend to have more fluid accumulated at the back of their necks during the first trimester, causing this clear space to be larger. While it will not give a definite diagnosis as compared to more invasive tests like CVS and amniocentesis, it can help you decide whether you want to undergo more diagnostic testing. And, unlike invasive diagnostic tests, it is painless and involves no risk to you or your baby.

How is the screening done?

An ultrasound must be done between 11 and 14 weeks of pregnancy by a physician certified to perform the test. An ultrasound confirms the date of the pregnancy and measures the nuchal thickness. The measurements are used along with the maternal age to calculate the baby’s chances of having a chromosomal abnormality, based on statistical probability. In addition to the ultrasound, two blood tests improve the risk assessment increasing the accuracy of the risk assessment. The blood tests measure two proteins: free B-HCG (the free beta subunit of human chorionic gonadotropin) and PAPP-A (pregnancy-associated plasma protein-A). The blood samples are collected from your finger stick.

What do the results mean?

You’ll be given your results in the form of a ratio that expresses your baby’s chances for having a chromosomal problem (based on your age, the baby’s age, and the nuchal fold measurement). For example, if you’re going to be 35 when you deliver, your baby’s average risk for a chromosomal abnormality is 1 in 178. (This risk gets higher as you get older.) If your baby’s nuchal fold measurement is found to be average for his age, your baby’s risk stays the same: 1 in 178. If it’s thicker than the average, your baby’s risk goes up, and your baby is considered at a higher risk for an abnormality. If the nuchal fold is thinner than the average, the baby’s risk goes down.

This test does not directly test for chromosomal problems; it just gives better indication of your baby’s statistical risk of having a problem. A normal result (sometimes called “screen negative”) is not a guarantee that your baby is normal, but it suggests that a chromosomal problem is unlikely. Nor does an abnormal result (sometimes called “screen positive”) mean that the baby has a chromosomal problem—just that he has an increased risk of one. (Even so, most “screen positive” babies still end up being normal.)

Based on your risk, you then will have to decide if the results indicate a high enough risk that you want to have more testing for a definitive diagnosis - that is, to see if your baby really does have a chromosomal defect. Individual parents-to-be have different feelings on what is an “acceptable” risk for them. Generally, we offer an invasive test if the risk of Down syndrome is

1 in 300 or worse. Tests that can diagnose a chromosomal defect include chorionic villus sampling (CVS) and amniocentesis.

What does it mean that this test is “91% accurate?”

You may have read that the results of this test are 91% accurate in detecting your risk of having a baby with Down Syndrome. That means that if you’re carrying a baby with Down Syndrome, there’s an 91% chance that the test will pick that up and give you a “screen positive” result that indicates further testing is recommended. It also means there’s a 9% chance that the test will miss the Down Syndrome and give you a “screen negative” result and diagnostic testing won’t be recommended. This does NOT mean that a “screen positive” baby has an 91% chance of having DS. It just means that 91% of babies who have DS will have screening results that are suspicious enough to recommend diagnostic testing. And 9% of babies who have DS will be shown to be at normal risk—that is, the results will be falsely reassuring.

This screening test also has up to a 5% false positive rate. (A “false positive” result is when a test suggests there may be a problem when, in fact, there is no problem.) In this case, a 5% false positive rate means that 5% of all the babies with normal chromosomes who are tested will be “screen positive”—meaning that the test will show them to be at an increased risk even though they’re normal. Based on this “false positive” result, their mothers may opt for invasive diagnostic testing that they otherwise might not have done.

What are the advantages to nuchal fold and Ultra-screen blood tests?

The advantage to these screening tests is that they can give you a better estimate of your baby’s risk for chromosomal problems at an early date without subjecting you to the small risk of miscarriage from a more invasive diagnostic test like CVS. If the risk is low, you can find out as soon as possible and may be relieved. If the risk is high you can decide whether to have CVS (done between 10 and 12 weeks), or amniocentesis, which can tell you for sure whether your baby has a problem while you are still in your first trimester or second trimester. The nuchal fold test is non-invasive and carries no more risk than an ordinary ultrasound. And even if you forgo diagnostic testing, you can get more information about your baby’s health and development by following up with a routine second trimester ultrasound at 18 to 20 weeks that looks for “soft markers” of chromosome disorders, such as short limbs, a bright dot in the heart, a bright bowel, cysts in a portion of the baby’s brain, and certain problems in the kidneys.

What’s the downside of these screening tests?

Like any screening test, they are not diagnostic—that is, they cannot tell you definitively if your baby has normal chromosomes. In some cases they will cause additional intervention and, in other cases they will be incorrectly reassuring. The nuchal fold test does not detect neural tube defects, such as spina bifida and other anomalies that may be indicated by the multiple marker test (done at 15 to 18 weeks), but the second trimester ultrasound done at 18 to 20 weeks should be able to detect these problems at least as well as the multiple marker test and with less false positives.

What is the cost of the various components of these tests, and what if my insurance does not pay?

Because this procedure is separate and additional from your global obstetric services, it may not be a covered benefit. Call your insurance carrier to determine your benefits. The current procedure code for most pregnancies is 76813 or (76814 for twins). The diagnosis code is 655.83.

How do I schedule the nuchal translucency screening test?

You must call a perinatology office (high-risk obstetrician) to schedule an appointment for genetic counseling and testing. Please check with your insurance carrier to determine whether a physician or facility is contracted and how it is covered. Locally, we recommend the Obstetrix Medical Group at (408) 371-7111, or Stanford perinatology department at (650) 725-7030.

How can I find out more information about the test?

An educational video can be viewed under first trimester screening at www.Obstetrix.com/sanjose. Click on the first trimester screening tabs.

Do I need to do an AFP test?

If you are over 35 and will have a Level II ultrasound, you do not need the AFP test. If you are under 35 and had a nuchal screening test or did a CVS, we recommend the AFP only test.