

A practical introduction to nuchal translucency screening

What nuchal translucency screening screens for

Nuchal translucency screening, or NT screening, is an ultrasound test. It screens for Down syndrome (trisomy 21, meaning an extra copy of chromosome 21) and other disorders that are caused by extra copies of chromosomes (trisomy 13, trisomy 18), as well as congenital heart defects.

Fetuses who have an extra chromosome may have more fluid at the base of their necks — a spot known as the nuchal fold — and this can make their necks larger. This fluid can be measured on a sonogram during weeks 11 to 14 when the base of the neck is still transparent. Timing is crucial, because the nuchal fold becomes less transparent as your baby grows. NT measurements are not conclusive, so the NT screening test can't tell you for sure whether your child has a chromosomal disorder, but it can be combined with other data (from blood tests and from population studies) to provide a statistic about the likelihood of such a disorder. This information can be helpful to parents who are trying to decide whether to have more invasive genetic tests, such as amniocentesis or chorionic villus sampling (CVS).

Who nuchal translucency screening is for

NT screening is available for any pregnant woman. It can be especially helpful for older moms-to-be who are at greater risk for having a baby with a chromosomal disorder, and who may be hesitant about more invasive genetic testing, like CVS and amniocentesis, because of the increased risk of miscarriage associated with them. Other likely candidates for NT screening are mothers with a history of problems in prior pregnancies or a family history of chromosomal disorders. The accuracy of the test is, however, dependent on the skill of the sonographer performing it and the sensitivity of the equipment. It is internationally recommended that NT screening be performed only at specialized and both certified centers.

How nuchal translucency screening is done

This procedure is done during an ultrasound. The sonographer will first measure the baby from crown to rump to ensure that the fetal age is accurate. Then the nuchal fold is located and measured. This may take some time, since the baby must be measured in a standardized mid-line fashion from head to toe.

When nuchal translucency screening is done

Since NT screening must be done between weeks 11 and 14, an accurate date of conception is important for an accurate result.

Additional tests - Combined NT Test

NT screening can be combined with blood tests for more definitive results. For example, your first trimester blood test measures free beta-hCG (a certain type of human chorionic gonadotropin hormone) pregnancy protein called PAPP-A (pregnancy associated plasma protein A). Low levels of PAPP-A early in pregnancy indicate increased risk of genetic abnormalities, heart problems, and preterm birth. The results of these two blood tests plus the NT screening, known as the first trimester combined screening, can be combined into one number that represents the likelihood of having a child with a genetic abnormality.

Risks

Unlike more invasive genetic tests, there are no increased risks of miscarriage for the NT screen (or the accompanying first and second trimester blood tests). The biggest problem might be the anxiety they can provoke. False positives are common. So are false negatives. But try to keep it in perspective: The odds of having a completely healthy baby are overwhelmingly in your favor. The NT screen is best thought of as a decision-making tool. Before you decide whether to have this procedure, talk to doctor about genetic counseling to help you determine which genetic tests are right for you.