Nuchal Translucency or NT

Patient Information Series – What you should know, what you should ask.

What is Nuchal Translucency (NT)?

NT is the name given to the black **area seen by ultrasound** at the back of the fetal head/neck **between 11 - 14 weeks** of gestation. The NT represents a normal accumulation of fluid, but, if too thick (usually above 3-3,5mm), it is a sign that something may not be going well with the development of your baby.

The most common problem is that the baby has a chromosomal anomaly, such as Down syndrome (extra chromosome 21), or a structural problem in another organ, such as the heart. For this reason, a thickened NT is called a 'marker' for fetal disorders. The magnitude of the risk of your baby having a problem is estimated by determining the 'risk calculation'. There is a screening test, called the **combined test,** that determines this risk calculation. The test combines the measurement of the NT, the length of the baby, your age, and the measurements of two hormones in your blood.

If the doctor has not yet done so, you can request this test and risk calculation. The risk tells you what is the chance that your baby has a problem with the chromosomes and can be reported as very low, such as 1 in 1000, or rather high such as 1 in 100. The doctor will discuss with you what the risk means for you and help you to interpret the test result. If the risk is low, you do not need other tests and will be reassured about the fact that it is unlikely that your baby has a problem. A risk of 1:100 is usually considered high risk. This means that if 100 women have been given the same risk, only one of them will have a baby with a problem and 99 will not.

Ultimately, interpretation of risk is highly individual and only you can decide your risk tolerance and whether you wish to follow up a positive screening test with a confirmatory, diagnostic test such as taking a small amount of tissue from the placenta or some fluid from around the baby (chorion villous sampling or amniocentesis).

How does the NT happen?

It is not clear why the fluid accumulates behind the fetal neck of all babies. When the NT is thin, it likely represents regular fluid accumulation during normal development. When the NT is thickened, above what is generally considered normal for the baby at that gestational age (usually more than 3-3,5mm), there is an increased risk for the baby of having a chromosomal anomaly (Down syndrome or others).

A thickened NT is seen in about 1 in every 20 fetuses, however not all these fetuses have a problem. About 1 of 10 fetuses with a thick NT has a chromosomal anomaly or another problem, usually a heart anomaly or other anomalies, but the others do not have any problem. Generally, a confirmatory test, called a diagnostic test, is required to distinguish these fetuses with a positive NT screening test.

How are chromosomes relevant to a thickened NT?

Chromosomes are where most of our genetic information is kept. We usually have 46 of them matched in pairs: 23 come from one parent and the other 23 come from the other parent. For example, people with Down syndrome have an extra chromosome number 21. Babies with a thickened NT have a higher chance of having a chromosome extra, usually nr. 21 or 18 or 13. It is also possible that the genetic defect is very small, at the gene, or part of a gene, level. That is why your doctor may suggest doing an even deeper examination of the genetic material of your baby.

Should I have more tests done?

Many women with a thick NT will choose to have more tests done to know more about the condition of the baby. Tests to ask about include:

- A **chorionic villous sampling** or **amniocentesis** to look for problems with the number of chromosomes and some of the problems within the chromosomes. This is done by removing a small amount of placental tissue or of amniotic fluid surrounding the fetus. After this test, the genetic material can be analysed in various ways. Depending on the test used, small genetic defects and genetic syndromes can be found. The most frequent genetic syndrome involving a single gene, that is found in a baby with a very thick NT is called Noonan syndrome.

- An **advanced fetal scan** including **fetal echocardiography** - a specialised ultrasound of all of the baby's organs and heart to analyze the structures of the fetus for structural anomalies.

- These **scans** can also be repeated **later in pregnancy at 20 and 28-32 weeks** gestation.

What are the things to watch for during the pregnancy?

If the tests are all negative it means that no problems have been found with your baby and your pregnancy can be considered normal, so you do not need any further special care during pregnancy. If the NT was very large it can cause a miscarriage and loose the pregnancy, even if no anomalies are found. In this case, the thick NT was a sign that your baby could not survive, due to unknown problems.

If the pregnancy continues normally, you may occasionally notice that your belly has grown very fast over the last week. This may be because too much amniotic fluid (water) surrounds the baby. This condition is called **polyhydramnios**. You should discuss this with your doctor and request a scan. A big baby with a lot of fluid may be a sign of Noonan syndrome.

What does a thickened NT mean for my baby after it is born?

If no problems or anomalies have been found with the tests the baby has no extra risks of abnormal development compared with a baby with a normal NT measurement. If anomalies have been found, the outcome will depend on the nature and severity of that anomaly. In very rare cases additional subtle findings, not suspected at prenatal ultrasound, may be detected after birth, increasing the risk of a genetic syndrome.

Will it happen again?

Usually, a thick NT will not occur again in the next pregnancy, but if the baby had a chromosomal anomaly or a heart anomaly the risk may be slightly increased in your next pregnancy. There are also women who had a thickened NT in more than one pregnancy, but all the babies were normal. It seems that these women may be more prone to have fetuses with extra fluid in the neck.

What other questions should I ask?

- Does this look like a severe case of thick NT?
- What is the chance of my baby having a chromosomal anomaly?
- Can the doctor do a combined test and give me an exact risk?
- Which invasive test should I choose for chorion villous sampling or amniocentesis?
- Which risks carry these tests?
- Which genetic investigations will be performed?
- Will these include diagnosis of small genetic problems and of Noonan syndrome?
- Will the doctor organize extra specialised scans in the pregnancy to check if the NT has disappeared and if there are other anomalies?

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