

## **Patient information on a thickened Nuchal translucency measurement**

### **What is a nuchal translucency (NT) measurement?**

This is a measurement taken of the thickness of the skin at the back of the your baby's neck. It has to be taken with your baby lying in a specific position and it must be taken when the head to bottom measurement, the crown rump length, measures between 45 and 80 mm. This is equivalent to 11 to 13 weeks and 6 days.

### **Why is it measured?**

It is known that increased NT measurements are associated with an increased risk of the baby having a chromosome problem eg Down's syndrome. The measurement can be entered into a computer program along with your age and if taken results of a specific blood test to calculate a risk. If you consider the risk to be too high you will be offered an invasive test such as an amniocentesis or chorion villus biopsy.

### **I have had an invasive test and it was normal: what does it mean now?**

In the majority of babies with an "abnormal" NT, the thickness will only have been slightly above normal. If the chromosomes are normal and the NT measurement less than 3.5 mm then it is likely that all will be well. This is because any measurement has a normal range. For example a women who is six foot tall is well outside of the normal range but that does not make her abnormal. Therefore if your NT measurement was less than 3.5mm you will be offered a 20 week ultrasound scan, back in your own hospital, when we will again check the baby's anatomy as carefully as they can. If all appears well at this time no further investigations will be performed.

### **What does an NT measurement of greater than 3.5mm mean?**

A NT measurement of greater than 3.5 mm can suggest a number of other potential problems, in addition to the increased risk of a chromosome anomaly. These include a structural abnormality such as a diaphragmatic hernia. (where there is a hole in the diaphragm that allows the stomach and other organs to slide into the chest and squash the developing lung) or a problem with the baby's movement which is usually possible to spot on ultrasound. In addition we know that the thicker the NT the more likely the baby is to have a heart defect and that thickened NT measurements can be seen in a variety of genetic disorders. Finally it can be a sign of a pregnancy which will miscarry and if this is going to occur it will usually happen before 20 weeks.

I

**f my baby’s NT is above 3.5mm will I be offered further tests or watched more closely?**

Yes. We will offer you another scan 2-3 weeks later. This will usually be between 14 and 16 weeks. This gives us an opportunity to check that the baby is growing as it should. It lets us check on the thickening, which may have disappeared increased or have stayed the same. It also allows us to check that the baby is developing as normally as possible. If we find a problem then this will be discussed with you along with your options.

At 19-21 weeks you will be offered a detailed ultrasound scan when we will again check the baby to see if we can find any evidence of a physical problem. In addition we will ask our colleagues in the paediatric cardiology department to check the baby’s heart as they are experts at picking up subtle heart defects.

If the chromosome test was normal, the anatomy and heart scans are all normal then we will be very reassuring. However, it must be remembered that not all problems can be seen on ultrasound.

**Is there evidence that a thickened NT will mean that my baby has a mental handicap (developmental delay)?**

Thickened NT measurements can be associated with a variety of disorders some of which may as part of the condition have either physical or mental problems as part of the condition. The available evidence on what happens to babies with an isolated thickened NT measurements is very limited. To date there is no convincing data to link an isolated thickened NT measurement with developmental delay.

**How high are the risks that my baby will have a problem?**

Overall outcome based on NT measurement at 11 – 13 weeks 6 days.

NT	Chromosomal defect	Fetal Death	Major fetal anomaly	Alive and well
<95 <sup>th</sup> centile	0.2%	1.3%	1.6%	97%
95-99 <sup>th</sup> centile	3.7%	1.3%	2.5%	93%
3.5-4.4mm	21.1%	2.7%	10%	70%
4.5-5.4mm	33.3%	3.4%	18.5%	50%
5.5-6.4%	50.5%	10.1%	24.2%	30%
>6.5mm	64.5%	19%	46.2%	15%

The above table gives a crude estimate of the likely outcome depending on the NT measurement. You will notice that the percentages do not add up to 100. Over all the exact numbers are less important than the concept that the thicker the NT measurement the more likely you baby is to have a poor outcome. HOWEVER, even when the NT measurement is over 6.5 mm 15% (one in eight) of these children will have a good outcome in the pregnancy and be alive and well.

**What are my/ our options?**

The options are always to continue with the pregnancy, and either carrying on regardless or accepting the additional investigation described above. Some women / couples will consider a termination, which is an option either at the time of diagnosis or at a later stage in the pregnancy, for example if an abnormality is found at the mid pregnancy scan.

The doctors and midwives looking after you are here to help. We realise that we cannot provide you with all the answers you need. We appreciate that this increases your anxiety / apprehension however we will try to do what ever we can. If you have any questions at any time please ask and we will do our best to answer it.