Coombe Women & Infants University Hospital

Ultrasound and Fetal Medicine Centre
Information for Women on Nuchal Translucency Screening (NTS) at 11-13^{+6} weeks

Some facts...

- *The vast majority of babies are normal at birth*
- *All women, whatever their age, have a small chance (3-5%) of delivering a baby with a physical or mental handicap*
- *In some cases, the birth defect is due to a chromosome problem such as trisomy 21, which causes Downs Syndrome*

Risks for Trisomy 21 in relation to maternal age

Maternal Age (yrs)	*Chance of Downs Syndrome At 12 weeks*	*At birth*
20	*1 in 1070*	*1 in 1530*
25	*1 in 950*	*1 in 1350*
30	*1 in 630*	*1 in 900*
35	*1 in 250*	*1 in 360*
36	*1 in 200*	*1 in 280*
38	*1 in 120*	*1 in 170*
40	*1 in 70*	*1 in 100*
42	*1 in 40*	*1 in 55*
44	*1 in 20*	*1 in 30*

Screening for Downs Syndrome and diagnostic testing

The only way to be certain whether the fetus has Downs Syndrome or not is by doing a **diagnostic test** – an amniocentesis or chorionic villus sampling (CVS).

A CVS is performed between 11 and 13^{+6} weeks and involves taking a small amount of tissue from the placenta, whilst an amniocentesis involves taking fluid from around the baby and is usually done after 15 weeks. Both provide a sample that contains tissue

that has the same genetic make-up as the baby and will allow the baby's chromosomes to be looked at in detail.

But the problem with these invasive tests is that they can cause a miscarriage, even if the baby is entirely normal. The risk of miscarriage is about 1% (1 in 100).

Therefore, the decision as to whether to have an invasive test or not must be taken on the basis of a non-invasive assessment of the likelihood (risk) that the baby has Downs Syndrome. The most accurate way of estimating this risk is by combining the information from the following:

- The mother's age
- Nuchal translucency (amount of fluid behind the neck of the fetus on ultrasound) carried out at 11 – 13^{+6} weeks
- Level of two proteins in your blood (PAPP-A and bHCG)

The nuchal scan is an ultrasound scan performed between 11 and 13^{+6} weeks of pregnancy, during which the fluid at the back of the baby's neck (the nuchal translucency) is measured. All babies will have some fluid here, but, in babies with Downs Syndrome, the fluid tends to be increased.

By combining this information we can identify about 9 out of 10 pregnancies (90 %) in which the baby has Down Syndrome. This is based on studies of more than 100,000 pregnancies.

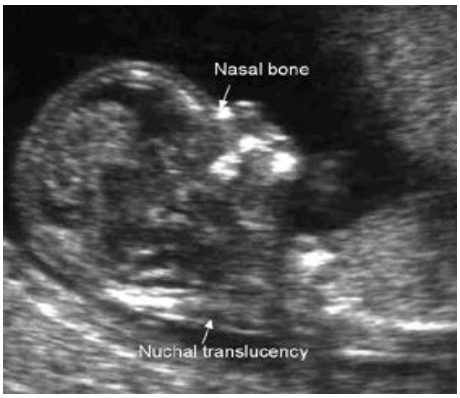
What else does the scan do?

- Confirm the dates of your pregnancy
- Provides reassurance that the baby is developing appropriately at this stage
- Detect some fetal abnormalities
- Diagnose a multiple pregnancy and if twins, whether they are identical or not

How is the examination performed?

The procedure is the same as for the routine ultrasound scan.

After an image has been obtained, a measurement is taken of the thickness of the skin at the back of the baby's neck. This information, together with your age and blood results, will be used to calculate an individual estimate of risk by using an authorised software package which is produced by the Fetal Medicine Foundation.



Nuchal Translucency Screening is performed by trained accredited staff who have achieved a certificate of competence.

The results

As your blood sample is sent away for analysis, the full result of your screening test may not be available for a number of days.

Depending on the estimated risk, you may be happy to continue with your pregnancy without further testing. This is the outcome for the vast majority.

If the estimated risk is high for your age, you may want a definitive answer by having a diagnostic test, such as a chorionic villus sample or an amniocentesis. This decision is balanced against the risk of miscarriage with a diagnostic test of about 1% (1 in a 100). This is a personal decision which only you can make. You will have the option of choosing a definitive test if you prefer.

Please see separate information factsheets on diagnostic tests - CVS and amniocentesis.

Cost of test Private/Semi-Private/ Public patients €250;

Booking an appointment If you would like to book an appointment, or you have any queries, please contact the Ultrasound Department on 01 4085578 (fax 01 4085574)

We hope the information supplied in this factsheet has been useful. We are constantly striving to improve information and communications with our expectant parents and if you have any suggestions on how this leaflet could be enhanced we would be delighted to hear from you.

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