Taking the combined test – screening for Down’s syndrome

This leaflet is about the combined test, which is a screening test for Down's syndrome in pregnancy. It is available to you between 11 weeks and 2 days and 14 weeks and 1 day of your pregnancy, when your baby’s Crown Rump Length (the length from the top of the head to the bottom of the buttocks) is between 45mm and 84mm. You can choose whether you wish to have the screening for Down’s syndrome or not. Your midwife or GP will discuss the test with you to help you decide whether you want to have it. For further information you can contact the Fetal Medicine Unit (see the back of this leaflet for contact details).

What is Down’s syndrome?

Down's syndrome is a condition some babies are born with. It affects about one in every 700 babies. Children can have a wide range of learning difficulties and some medical problems, such as heart defects, bowel problems and chest and sinus infections. Some adults with Down’s syndrome are able to get jobs and live fairly independent lives. However, most people with Down’s syndrome need long-term support and ongoing care.

Down's syndrome is caused by the presence of an extra chromosome (number 21) in the cells of a developing baby. Usually it is not inherited and so a baby can be affected even if there is no family history of Down's syndrome.

We do not know exactly why babies get Down’s syndrome. Although older women are more likely to have a baby with the condition, any woman can have a baby with Down's syndrome. It is not caused by anything parents have done or not done.

What is the combined test?

This test combines information from an ultrasound scan of the baby and a blood test from the mother.

The scan (sometimes called a nuchal translucency test) measures the small collection of fluid at the back of a baby’s neck. All babies have this fluid, but it may be increased in babies with Down’s syndrome.

The blood test measures two substances in your blood. These are hormones called PAPP-A (pregnancy associated plasma protein-A) and free B-hCG (B-human chorionic gonadotrophin). On some occasions, because of the mother’s health or the baby’s position, we may be unable to perform the test. If this is the case we will discuss alternative options with you.

What are the benefits of having the combined test?

The combined test is a screening test that predicts the chance of your baby having Down’s syndrome. It cannot tell you whether or not your baby is definitely affected. Combined
screening picks up 85 to 90% of babies with Down’s syndrome (more than four out of five). This means that 10 to 15% of Down’s babies are not picked up by this test (less than one in five). In 2.5% of cases (about two out of every 100 tests), the test result may indicate that your baby has Down’s syndrome, when he/she doesn’t. This is called a false positive test result.

What will happen if I have the test?
When you arrive in the Fetal Medicine Unit a member of the team will take a blood sample. The blood takes approximately 40 minutes to be processed. After this, you will go for your ultrasound scan, which takes around 20 minutes. The sonographer (person performing the scan) will discuss the results with you and you will be given a printed report to keep in your maternity notes.

You should expect to be in the Fetal Medicine Unit for between one and two hours.

You do not need to have a full bladder for the scan, but you should avoid going to the toilet just before the scan, as this can help the quality of the image.

Are there any risks to me or my baby?
The scan and the blood test are safe and will not harm you or your baby.

How is the risk of Down’s syndrome calculated?
The risk of Down's syndrome following the combined test is based upon a combination of:
- your age
- the measurement of nuchal translucency
- the length of your baby
- the level of the two hormones (PAPP-A and free B-hCG) in your blood.

We use a ‘risk cut-off’ of one in 100 at the time of the scan, to identify babies that we consider as having a higher risk of Down’s syndrome. This means that if your combined test result indicates a risk of between one in two and one in 100, we anticipate that your baby is at increased risk of having Down’s syndrome. If the results show a risk of one in 101 or more, this is classified as a lower risk result, meaning that your child is less likely to have Down’s syndrome.

What happens if I have a high risk result?
Most babies at increased risk will not have Down’s syndrome. If the chance of your baby having Down’s syndrome is higher than one in 100 (at the time of the scan), you will be offered the opportunity to discuss diagnostic testing, such as amniocentesis (carried out from 16 weeks) or chorionic villus sampling (CVS) (carried out at 11–14 weeks). These tests can give a clearer indication of whether your baby has Down’s syndrome or not. However, a diagnostic test carries a risk of miscarriage of around 1% above the usual risk of miscarriage. If you have difficulty deciding what to do and would like further counselling, the fetal medicine midwives are available to discuss your options.

Does the combined test check for other problems?
The measurements used as part of the combined test can also identify pregnancies at high risk of Edwards syndrome (also known as trisomy 18) or Patau syndrome (sometimes called trisomy 13). Edwards and Patau syndrome are rare but serious conditions and affected babies
can have a range of severe medical problems. Sadly, most babies with Edwards and Patau syndrome die before the end of pregnancy or soon after birth.

During the scan, the sonographer will also check your baby’s physical health. As the baby is still very small, most problems cannot be detected. This is why we always recommend that you have a further scan in mid-pregnancy (the detailed scan) when the baby will be bigger.

**Attending your appointment**

Appointments for combined screening are in demand due to the limited period of time in which they can be performed. If the appointment you are sent is not convenient or if for any reason you are unable to keep it, please telephone **020 7188 8003** and let us know.

If you are late for your appointment, we may not be able to perform your scan or give you another appointment.

It is important that the sonographer is able to concentrate during the scan, so we ask that there are only one or two people in the room during the ultrasound examination. Taking pictures or films during the ultrasound is strictly forbidden.

Guy’s and St Thomas’ is a teaching hospital, and so we may wish to use images from your scan to help students learn. Please speak to the sonographer if you have any questions or concerns, or if you do not want images from your scan to be used in teaching.

**Are there any alternatives to the combined test?**

There are other screening tests available, which can be carried out at different stages in your pregnancy:

**Serum screening – quadruple test**

If your pregnancy is too advanced (more 14 weeks plus 1 day) for the combined test to be carried out, we will offer you another blood test. This is called the quadruple test. This test can be done between 14 weeks and 2 days and 22 weeks plus 6 days (ideally between 15 weeks and 20 weeks and 6 days). The quadruple test picks up around 80% of babies with Down’s syndrome. This means that 20% of Down’s babies will not be picked up by this blood test. We have a separate leaflet for the quadruple test – please ask if you would like a copy. If you have a combined test, we will not offer you a quadruple test, as it is less specific and less helpful.

**The detailed 20 week anomaly scan**

Around half (50%) of babies with Down’s syndrome will have features that can be picked up by this scan. However, as the other 50% of Down’s babies do not have these features, scan findings may appear completely normal, even if your baby does have Down’s syndrome.

**Non-invasive prenatal testing (NIPT) – Verifi test**

NIPT is not available on the NHS and is only available privately in the UK at present. We are in a position to offer you this test for a fee of £460.

Verifi has a detection rate of 99.9% for Down’s syndrome, 97.4% for Edward’s Syndrome and 87.5% for Patau Syndrome. The results take seven working days.

Like the combined test, Verifi is a screening test, and a diagnostic test like a CVS or an amniocentesis will still be needed to confirm a positive result.
If you would like more information, please contact the Fetal Medicine Unit midwives on 020 7188 2321.

Decline screening
The screening for Down’s syndrome is optional. If you decide not to have any screening, we will still date the pregnancy and check your baby’s physical health and progress. We will also offer to check the PAPP-A level, as this information can be useful at other stages of the pregnancy. If a low PAPP-A is detected, we will organise further scans to look at the growth of the baby.

Further information
You can get more information about screening from the following organisations:

Antenatal Results and Choices
t: 020 7631 0281
w: www.arc-uk.org

Fetal Medicine Foundation
t: 020 7034 3070
w: www.fetalmedicine.com

National Screening Committee
Antenatal and Newborn Screening Programmes
w: www.screening.nhs.uk

National Institute for Health and Clinical Excellence
w: www.nice.org.uk

Contact us
If you have any questions or concerns about the test, please contact the Fetal Medicine Unit on 020 7188 2321 and ask to speak to the fetal medicine midwives.

The Fetal Medicine Unit is open between 9am and 4.30pm, Monday to Friday.

For more information leaflets on conditions, procedures, treatments and services offered at our hospitals, please visit www.guysandstthomas.nhs.uk/leaflets

Pharmacy Medicines Helpline
If you have any questions or concerns about your medicines, please speak to the staff caring for you or call our helpline.
t: 020 7188 8748 9am to 5pm, Monday to Friday

Patient Advice and Liaison Service (PALS)
To make comments or raise concerns about the Trust’s services, please contact PALS. Ask a member of staff to direct you to the PALS office or:
t: 020 7188 8801 at St Thomas’ t: 020 7188 8803 at Guy’s e: pals@gstt.nhs.uk

Language Support Services
If you need an interpreter or information about your care in a different language or format, please get in touch using the following contact details.
t: 020 7188 8815 fax: 020 7188 5953