Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive

Cork University Maternity Hospital,
Health Service Executive,
Wilton
Cork.

CUMH prenatal screening : Combined Test

This leaflet has been provided to help answer some of the questions you may have about the COMBINED TEST

What is combined testing?
Combined testing is a screening test to determine your risk of having a baby with Down syndrome. This test involves an ultrasound scan (measuring the baby's nuchal translucency or neck thickness) plus a blood sample and is performed between 11^{+2} and 14^{+1} weeks. The measurements are then integrated together with your age, weight, weeks of pregnancy (gestational age), family origin and smoking details to estimate your chance of having a baby with Down syndrome.

The nuchal translucency scan (NT) is no longer available as a stand alone screening test in the CUMH, as its detection rate is only 80% with a false positive rate of 5%.

What is Down syndrome?
Down syndrome is caused by an extra copy of chromosome number 21 in the baby's cells. Anyone can have a baby with Down syndrome, regardless of age or family history. It is usually not inherited. However, a woman's risk of having a baby with Down syndrome increases with age. Down syndrome occurs in one out of every 700 babies and is the most common cause of learning disabilities. Babies with Down syndrome are also at greater risk of heart defects and other medical problems. Babies with Down syndrome often miscarry, and about 10% die in infancy. Life expectancy in the rest is around 40-60 years, but the severity of the condition varies.

How good is this screening test?
The Combined test will correctly identify between 85 and 90% of babies with Down Syndrome. Around 1 in every 50 results from this test will identify the baby as being at increased risk. Most pregnancies with a "high risk" result will not be affected by Down syndrome. The false positive rate for combined screening is 2.0%.

Why does combined testing have 2 steps?
Combined testing merges the strengths of ultrasound and blood testing. The blood test measures 2 substances, human chorionic gonadotrophin (hCG) and pregnancy-associated plasma protein (PAPP-A), which are passed from your baby to you. If you are carrying a baby with Down syndrome you are likely to have abnormal levels of these substances in your blood. Integrating measurements from both steps into one result makes the test more accurate, meaning it identifies more affected pregnancies as high risk, and more normal pregnancies as low risk.

What happens during the test?
The test is best performed between 11^{+2} -14^{+1} weeks. The NT scan measures (in millimetres) the thickness of skin at the back of the baby's neck. We will also collect the blood sample at this visit. A computer program in the laboratory will then use the results from the blood sample combined with the N.T. measurement and the mother's age, weight, weeks of pregnancy, family origin and smoking details to calculate the overall risk of Down syndrome.

What about the results?
Results are given as a '1 in … chance' of the baby being affected, rather than a 'yes' or 'no'. We will receive most of the results within 5 working days.

If your risk is less than 1 in 150, you will be considered low risk and no further tests will be offered. You will receive the results in the post usually within 5-7 working days.

If your risk is 1 in 150 or greater we will contact you by phone within 5 working days and you will be referred for a discussion with a Fetal Medicine Specialist. You will be offered a diagnostic test called an amniocentesis. Amniocentesis will give a definite 'yes' or 'no' answer about Down syndrome or other major chromosome abnormalities, but is associated with approximately a 1% (1 in 100) risk of causing a miscarriage.

How do I make an appointment?
This screening test is not available at your routine booking visit at 12 weeks or at your dating ultrasound between 10-14 weeks. It is a separate clinic that you can book if you are interested in prenatal screening.

Please contact our prenatal screening clinic which is held on Monday afternoons in the Aislinn Suite on (021) 4920550 at CUMH. You can make an appointment by ringing this number between 2-4pm on Monday-Thursday. There are a limited number of appointments available for this clinic..

What are the costs?
This test is not funded by the Health Service Executive or the CUMH. It is available to you for a cost of €150. This fee includes administration costs, the laboratory charges for the blood tests and data analysis, the scan, as well as the courier charges to the laboratory in Birmingham.

How do I pay?
On the day of your appointment, please bring a cheque or bank draft for €150 with you. This should be made payable to "HSE South".

Who can I contact with questions?
If you have any questions about combined screening or your test results, please contact the Aislinn Suite from Monday to Thursday (2-4pm) and ask to speak to one of the midwives working in the unit.