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Childhood Seizures

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Rapid Prenatal Diagnosis For Pregnant Women - The Next Day Test

With today's technology, the results of a blood test to check our haemoglobin level are available within the hour. Why then are parents-to-be currently expected to wait up to 4 weeks for the results of their prenatal diagnostic tests and to find out if their baby is normal, or has Down's Syndrome?

At NUH, our solution is the Next Day Test.

Down's Syndrome in older and younger mothers

Most of us are aware that the risk of Down's Syndrome in the fetus rises as the mother gets older, but what is less known is that even young mothers are at risk of delivering a baby with Down's Syndrome. In fact, more babies with Down's Syndrome are born to mothers under the age of 35 than to mothers older than 35. This is because 4 out of 5 mothers are younger than 35 years of age.

Screening and Diagnostic tests

Ultrasound has been used for many years to examine the fetus for abnormalities such as club feet, spina bifida and a hole-in-the-heart. This hasn't changed much over the years. On the other hand, the screening test for Down's Syndrome has traditionally been a maternal blood test (Triple test), taken at about 15 weeks of pregnancy. The result takes a week or two to be ready, and is only 60 to 70% accurate. Thereafter, if the mother is classified as having a 'high risk for carrying a Down's Syndrome baby', an amniocentesis is organised and performed, and the results of this diagnostic test takes up to 3 weeks to be ready. In all, it could take up to 4 to 6 weeks for the results to be informed to the parents. This means 42 days of anxiety for the couple.

Today, technology allows us to produce the results the very next day.

The Combined Test – Ultrasound NT + Maternal blood biochemistry

A new test, that combines ultrasound and a maternal blood test, can be performed as early as 11 weeks. This is the Combined Test, and is typically performed at 12 weeks of pregnancy. This test has many advantages over the older tests. During this test, we can accurately determine the age of the fetus (date the pregnancy), confirm that the fetus is alive and viable, that it is a single baby, and by measuring the nape of the fetus inside the womb (nuchal translucency, NT), we can determine the likelihood of the fetus being affected by Down's syndrome with 80% accuracy. This can be improved to 90% when combined with the maternal blood test done at the same time. If the Test is performed early in the morning, the results will be available by noon. Couples will know if they are at low or high-risk of carrying a baby with Down's syndrome.



Figure 1. Nuchal Translucency (NT) measurement

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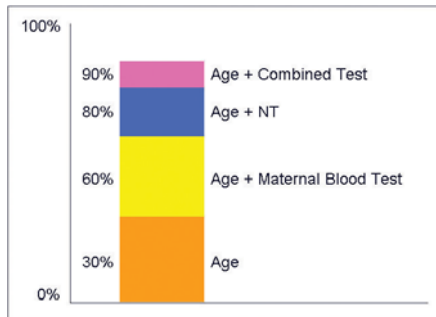


Figure 2. Accuracy for detection of Down's Syndrome increases using the Combined Test

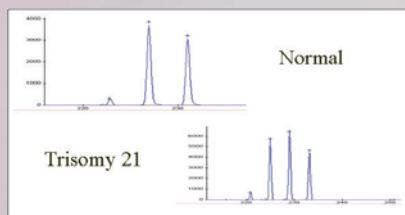
Amniocentesis, Chorion Villus Sampling and the Full Chromosome Result

Conventionally, the amniocentesis has been used to test mothers at high-risk of carrying a baby with Down's syndrome. These days, the chorion villus sampling (CVS) is gaining popularity among pregnant women as it can be performed as early as 11 weeks, and give accurate results about the fetal genetic condition. In amniocentesis, a small sample of amniotic fluid is collected for analysis whereas in CVS, a small sample of the placental tissue is collected to study the fetal chromosomes. Both tests are diagnostic in that the results will reveal if the baby has, or does not have, Down's syndrome.

Ordinarily, once the amniocentesis or CVS has been performed, the fetal cells are grown (cultured) in a Petri dish and their chromosomes studied. This Full Chromosome Result (also known as karyotype) takes between 10 - 21 days to be ready depending on the quality of the sample available. But recently, we have developed 2 rapid molecular tests that allow us to be able to give the couple the results within 48 hours, usually by the next day.

The PCR test

The first of these tests uses a technology known as polymerase chain reaction, or PCR. In this test, we obtain the DNA of the fetal cells from the amniocentesis or CVS sample, and amplify short segments of the genes within the Down's syndrome chromosome, chromosome number 21. The same can be done for other



chromosomes that are commonly affected e.g. chromosomes number 13, 18, and the sex chromosomes X and Y. This test usually takes 48 hours.

Figure 3. The PCR Test result from a normal and a Down's Syndrome fetus

The Next Day Test



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The FISH test

The other test, known as fluorescence in situ hybridisation or FISH for short, checks the chromosomes directly within the fetal cells obtained from the amniocentesis or CVS sample. In this test, there is no need to extract the DNA from the fetal cells, and can usually be performed faster. This test can confirm Down's syndrome within 24 hours, and we can release the results to the couple the very next day after amniocentesis or CVS.

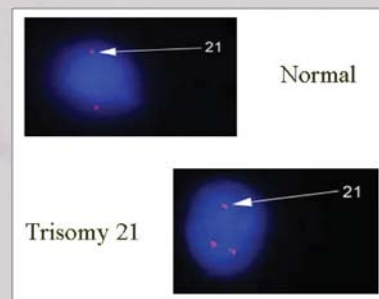


Figure 4. FISH test showing cells from a normal fetus and a Down's Syndrome fetus (Trisomy 21)

The Next Day test

All these tests have now been integrated at the NUH Antenatal Diagnostic Centre (ADC), into the Next Day Test. The Combined Test is performed early in the morning, and if the mother is in the high-risk category, she can choose to have a CVS on the same day. Apart from the Full Chromosome Result, she can also choose to have either the PCR test, or the FISH Next Day Test.

Contact us

For more information or appointments, please contact:

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