

SCREENING IN PREGNANCY

This document has been written to provide you with information about the tests available to screen and test for possible foetal abnormalities in pregnancy. Whilst reading this, please remember that the vast majority (98%) of babies are normal. All women, whatever their age, have some risk of having a baby with physical and/or mental disability.

In some cases the disability is due to chromosomal abnormalities such as Down's Syndrome (trisomy 21). The only way to be certain if a baby has a chromosomal abnormality is to have an invasive test such as Amniocentesis or CVS (see over). Both tests increase the risk of miscarriage by 1%.

SCREENING TESTS

Screening tests do not carry any risk to the pregnancy and are designed to identify women at higher chance of having a baby with a chromosomal abnormality, Down's Syndrome is the commonest cause of learning disability, but screening may also show other chromosomal variations of differing significance.

Screening does not tell you for sure whether or not the pregnancy is affected, but it may help you decide if you want a diagnostic test.

DIAGNOSTIC TESTS

These tests give a definitive answer about whether or not the baby has a chromosomal abnormality. If the result is abnormal, we will provide counselling, support and information within 36 hours so that you can decide whether or not to continue with the pregnancy.

	RISK OF DOWN'S SYNDROME		
	Age	At Birth	At 12 weeks
<i>This table shows how the risk of delivering a baby with chromosome abnormality increases with age. At 12 weeks the risk is higher because many fetuses with chromosome abnormalities die naturally during pregnancy.</i>	20	1: 1527	1: 950
	25	1: 1352	1: 898
	30	1: 895	1: 795
	34	1: 446	1: 262
	36	1: 280	1: 165
	38	1: 167	1: 98
	40	1: 97	1: 57
	42	1: 55	1: 32
	44	1:30	1: 18

TESTS AT 11 – 13 WEEKS – NUCHAL TRANSLUCENCY SCAN

This is a screening test. You will be offered an early pregnancy scan which may identify babies who are more likely to have a chromosomal abnormality. The baby is seen with ultrasound during a scan lasting 15 – 20 minutes. The size of the baby is measured to accurately date the pregnancy. At the same time, it is possible to measure the amount of fluid behind the neck of the baby. By combining the risk based on your age, with the information from the scan, we can give you an individual risk of chromosomal abnormality in this pregnancy.

If the scan is not reassuring, you may wish to have further tests such as Amniocentesis or a CVS to get a definite answer. If this is normal, it is still important to consider having a detailed scan at 20 weeks to exclude other defects that are not due to chromosomal abnormalities. **Some women may choose not to have this test. The choice is entirely yours.**

CHORIONIC VILLUS SAMPLING (CVS)

This is a diagnostic test. Local anaesthetic is used and a thin needle is passed through the abdomen to take a sample from the placenta. Both baby and placenta (afterbirth) develop from the same cell and so the chromosomes present in the placenta can be used to check the chromosomes in the baby. CVS is not performed before 11 weeks. The procedure takes 3-4 minutes, is performed under scan guidance and the results are available in about two weeks. The extra risk of miscarriage associated with this test is 1%. In approximately 1% of cases, the test will need to be repeated because the cells will not grow in the laboratory or the results are inconclusive.

TEST AT 15 – 19 WEEKS – BLOOD TEST

DOUBLE TEST/SERUM SCREENING

This is a screening test. A sample of the mother's blood is taken at about 16 weeks of pregnancy. For accurate results, the pregnancy should be dated by ultrasound. By analysing two different substances in the blood, it is possible to estimate the risk of Down's Syndrome. The results take approximately one week. If you are found to be at high risk (screen positive), there is still only a small chance that your baby is affected. If this test is not reassuring, you may wish to consider a diagnostic test (Amniocentesis) to give you a definite answer. If however you prefer not to have an invasive test, we would offer to carry out a detailed scan at 20 weeks. **Some women may choose not to have this test. The choice is entirely yours.**

We strongly urge you not to have this test if you have had the nuchal translucency test at 11 – 14 weeks, otherwise you will significantly increase the risk of having a false positive result and therefore an unnecessary diagnostic test.

AMNIOCENTESIS

This is a diagnostic test. A fine needle is passed through the abdomen and amniotic fluid around the baby is taken and tested for abnormalities. The procedure is very quick, taking less than 2 minutes. It is performed under scan guidance and the results take about 2 weeks. The extra risk of miscarriage is 1% and in approximately 1% of cases, the test will need to be repeated, because the cells will not grow in the laboratory or the results are inconclusive.

TEST AFTER 20 WEEKS – DETAILED ANOMALY SCAN

All women will be offered a detailed scan at 20 weeks. This scan looks for major physical abnormalities such as spinal bifida, kidney and heart problems. In addition, the scan will also check to see if there are any features of chromosomal abnormality. Measurements of the babies size will be made. In the majority of cases, this scan will be normal and reassuring. Occasionally an abnormality may be detected. You will be told after the scan of any abnormal findings and a further scan will be arranged if necessary.

LIMITATIONS OF SCANNING

Even if your scans are normal, it is impossible to totally exclude Down's Syndrome and every foetal abnormality. However, if no abnormal features are present, the risk of chromosomal

abnormality is reduced. Although very reliable, 20 week anomaly scanning cannot detect every physical and mental abnormality.

CORDOCENTESIS

This is a diagnostic test. A thin needle is passed through the abdomen to sample the babies blood (from the umbilical cord). Local anaesthetic is used. The procedure takes 2 – 3 minutes, is performed under scan guidance, and the results are available in 3 days. The extra risk of miscarriage is 1%. **This test is not performed before 20 weeks.**

SUMMARY

Gestation (weeks)	Screening Test	Diagnostic Test
11 - 13	Early Scan	CVS
15 - 19	Blood Test	Amniocentesis
20 PLUS	Detailed Scan	Cordocentesis

Whilst note of the screening tests will detect every abnormality, the 11-13 week scan will give you your risk earlier in pregnancy. We do not advise you to have both screening tests.