Frequently Asked Questions about Prenatal Screening for Downs Syndrome and Neural Tube Defects.

Q. What are Antenatal Screening Tests?
A. "Antenatal" or "Prenatal" just means "before birth" and the aim of screening at this stage is to pick out pregnancies where the baby may be affected by a particular type of rare abnormality before birth.

Q. What antenatal screening tests are available?
A. The most commonly offered screening tests are carried out between about 15 and 20 weeks of pregnancy and are designed to identify pregnancies where the baby has spina bifida or Down's syndrome or other similar types of abnormality.

Q. Can the screening test detect all affected pregnancies?
A. No. About 4 out of 5 spina bifida pregnancies and 2 out of 3 Down's syndrome pregnancies are identified. Occasionally, other, rarer abnormalities are picked up by the screening test.

Q. What is spina bifida?
A. Babies with spina bifida have an opening in the bones of the spine and the nerves to the lower part of the body are damaged. This can result in difficulties with walking and bowel and bladder control.

Q. How common is spina bifida?
A. Happily, most pregnancies are perfectly normal, but occasionally a developing baby may have an abnormality such as spina bifida. Spina bifida affects around 1 or 2 in every 1000 pregnancies.

Q. What is Down's syndrome?
A. Down's syndrome is a chromosomal abnormality. Chromosomes carry genes which pass certain characteristics from parents to their children. Abnormalities can occur when there are too many chromosomes. People with Down's syndrome have three chromosomes 21 instead of the normal two. This is why Down's syndrome is sometimes called "trisomy 21". Children with Down's syndrome will have a learning disability and may have other abnormalities such as heart defects.

Q. How common is Down's syndrome?
A. On average, about 1 in every 700 pregnancies is affected by Down's syndrome, although the frequency in individual pregnancies is related to the mother's age. Down's syndrome is more common in older mothers. For example, the risk of having a baby with Down's syndrome is about 1 in 1500 at age 20 years and that rises to about 1 in 100 at age 40 years.

Q. Can spina bifida and Down's syndrome be cured?
A. No. People with spina bifida or Down's syndrome will have varying degrees of disability but with help and support many sufferers can often lead fulfilling and rewarding lives. However, if it is shown through prenatal testing that a pregnancy is affected, the couple then have a difficult decision, - whether to prepare for the birth of a handicapped child or choose a termination of pregnancy.

Q. Who should have the screening tests?
A. All pregnant women are offered screening but it is up to individual couples to choose whether or not to have screening. These tests are available on the NHS and there is no charge for testing.

Q. How do the screening tests work?

A. There are two stages to testing. The first stage is the screening test and depending on the results of the screening test a second, follow-up or diagnostic test will be offered. It is very important to realise that the screening test is not conclusive, and a positive screening result does not mean that the baby is affected, it just means that there is an increased chance that the baby may be affected. A second, definitive test is required to confirm or exclude the presence of a abnormality in the baby.

Q. How is the screening test for spina bifida carried out?

A. For spina bifida screening, a substance called alphafetoprotein (AFP) is measured in a small sample of the mother's blood taken between 15 and 20 weeks of pregnancy. 85% of pregnancies with spina bifida have higher than normal levels of AFP, but so also do some perfectly normal pregnancies. Therefore the AFP test cannot distinguish completely between normal and affected pregnancies. Positive identification of affected pregnancies requires the use of a detailed ultrasound scan.

Q. How is the screening test for Down's syndrome carried out?

A. The same blood sample, taken between 15 and 20 weeks for the spina bifida test can be used for the Down's syndrome screening test. In addition to measuring AFP, another substance produced during pregnancy called hCG is also measured. If the baby has Down's syndrome, there is a tendency for the AFP level to be lower than normal and for the hCG level to be higher than normal. These results are converted to a probability or chance that the baby has Down's syndrome and that chance is combined with the mother's age risk. This screening test can identify about 65% of pregnancies with Down's syndrome.

Q. What happens if the screening result shows a high risk of Down's syndrome?

A. If the combined risk is considered to be high for example 1 chance in 50, a second follow-up or diagnostic test is offered. About 1 in 20 women will have screening results with risks high enough to be offered a diagnostic test. For Down's syndrome the follow-up test is amniocentesis which involves inserting a needle into the pool of fluid surrounding the baby, removing a small quantity, and growing the cells floating in the fluid in the laboratory to show the baby's chromosomes. The amniocentesis test will show whether the baby has normal chromosomes or is affected.

Q. What happens if the screening result shows a low risk of Down's syndrome?

A. Low risk does not mean "no risk" but as there is only a small chance that the baby is affected, the amniocentesis test is not recommended. 19 out of 20 women will be low risk.

Q. As amniocentesis is a more accurate test for Down's syndrome, why is amniocentesis not offered to all pregnant women instead of screening?

A. Because there is a risk (about 1 in 100) that the amniocentesis procedure might cause a miscarriage and it is therefore best to use it only where screening has shown that there is an increased chance that the fetus may be affected.

Q. Both of these screening tests are carried out at 15-20 weeks. Can they be carried out earlier in the pregnancy?

A. Testing for Down's syndrome before 15 weeks is possible, but requires the use of a different test. With regard to spina bifida, AFP measurements in the mother's blood before 14
weeks do not show up spina bifida pregnancies and it is best to carry out AFP testing at 15-20 weeks or a detailed ultrasound examination later in the pregnancy, around 20 weeks.

Q. How is the early screening for Down's syndrome carried out?

A. New tests have been developed which use both ultrasound measurements of the baby and laboratory measurements of factors in the mothers blood. This is called CUB screening, which is short for "Combined Ultrasound and Biochemical" screening and is carried out at 11-14 weeks of pregnancy.

Q. What is the ultrasound test in CUB screening?

A. This is a very accurate ultrasound measurement of the fluid which gathers at the back of the baby's neck. This area is called nuchal translucency and it has been shown that the thickness of the nuchal translucency is increased at 11-14 weeks of pregnancy in many babies with Down's syndrome or other types of abnormality.

Q. What is the biochemical test in CUB screening?

A. The second part of the CUB test is the laboratory measurement of two pregnancy factors in the mother's blood called free beta hCG and PAPP-A. If the baby has Down's syndrome, there is a tendency for free beta hCG levels to be higher than normal and PAPP-A levels to be lower than normal.

Q. How are the ultrasound and biochemical tests combined?

A. Both the ultrasound measurement and the biochemical measurements can be converted to risks or probabilities and combined with the mother's age risk, - in much the same way as for the established test which is offered later in pregnancy. The combined result then gives the chance that the baby is affected. If high chance, the mother can be offered amniocentesis or a different type of follow-up test called chorionic villus sampling which can be carried out before 15 weeks, to check the baby's chromosomes.

Q. What are the advantages of CUB screening over the later test for Down's syndrome?

A. It has been shown that the CUB test is more accurate. Over 80% of Down's syndrome pregnancies can be picked up by CUB screening with the added advantage that the result is available earlier in pregnancy.

Q. Is CUB testing available to all women on the NHS?

A. Women, booked for their antenatal care at the Queen Mother’s Hospital are routinely offered CUB screening. A few centres are beginning to offer this new type of test on a selective basis but it will take perhaps 2-3 years before CUB screening is offered to all pregnant women in Scotland.