

SCREENING, TESTING

and Down's Syndrome

INTRODUCTION

Screening is an extremely emotional issue. Expectant parents therefore need to have clear information about each stage of the screening process and what the possible outcomes may be. Screening should be seen as a means of assisting parents to make informed choices about their pregnancy.

Screening and testing for Down's syndrome (and Spina Bifida) is now common practice. Many women enter into the screening process without much explanation as to why they are being screened and what the possible implications may be for

their pregnancy. Many accept screening as routine, something that will reassure them that all is well.

There has always been recognition that the risk of having a baby with Down's syndrome increases with maternal age. In the past, screening was only offered to women over a certain age. However, the majority of children are born to women in their twenties and early thirties, and 75% of babies with Down's syndrome are born to women in this age group. In recent years, screening has increased to include women of all ages.

MATERNAL SERUM SCREENING

Maternal Serum Screening (MSS) is offered to all women at about 15-16 weeks into pregnancy. A blood sample is taken from the mother's arm. The blood test is called the double test or triple test depending on which hormones are used as part of the test. It is sometimes called the Bart's test.

The sample is sent to the lab and tested for certain hormones and proteins. These are AFP (alpha fetoprotein) and hCG (human gonadotrophin) with or without Oestriol. The test measures the levels of the proteins as a percentage. This is used in conjunction with the mother's age, weight and exact gestation, to assess the chance, or risk, of the baby having Down's syndrome.

The results of the test are expressed as either a risk value,

e.g. 1 in 300, or as a positive or negative screening. Having a positive screening (increased risk) does not mean that the baby will definitely have Down's syndrome. Having a negative screening (low risk) is not a guarantee that the baby will not have Down's syndrome.

A risk value of 1 in 300 is an indication of the chance that the baby will have Down's syndrome. It means that 299 people with this risk will not have a child with Down's syndrome and only one person will.

The sensitivity rates for serum screening vary. 40% of Down's syndrome pregnancies will not be detected. For every one that is identified, 60 unaffected pregnancies will be given anxiety provoking false positive results.

ULTRASOUND SCAN

The ultrasound scan is part of the screening process. Scans are usually performed at around 12 weeks and again at 18 weeks. The scan is not simply a chance for parents to have a look at their baby. The radiographer will use the scan to look for thickening or swelling at the back of the neck. This is called nuchal translucency. The scan will also look for shortened length of long bones and a sandal gap between the first and second toe. Ultrasound cannot diagnose Down's syndrome, or exclude it. However, if the above markers are seen, the possibility of the baby having Down's syndrome is increased.

Many hospitals have an automatic cut off point from offering a

woman further screening. This is generally based on a risk value of 1 in 250 because this figure is close to the spontaneous miscarriage rate caused by amniocentesis. Most parents would not consider the risk of miscarriage worthwhile if it is significantly greater than the chance of Down's syndrome being diagnosed.

Parents who have been told that there is a high risk of their baby having Down's syndrome need to have time to gather their thoughts and decide on what steps to take. Some parents will decide against any further testing. Others will feel that they need to know for definite, whether or not their baby has Down's syndrome.

AMNIOCENTESIS AND CHORIONIC VILLUS SAMPLING

Amniocentesis and Chorionic Villus Sampling (CVS) are both invasive tests that can give a definite diagnosis that the baby has Down's syndrome.

Amniocentesis involves a very fine needle being passed into the womb, under guidance by the ultrasound. A sample of the amniotic fluid is taken from around the baby. This test is done under local anaesthetic and carries a risk of miscarriage believed to be between 1 – 4%. This means that for every baby that is diagnosed as having Down's syndrome, up to 4 babies who do not have Down's syndrome are miscarried.

In Chorionic Villus Sampling, cells are collected from around the placenta. CVS carries the same risk of spontaneous miscarriage as

Amniocentesis and there is currently some debate about whether the procedure can lead to other birth defects. CVS and Amniocentesis results can take up to three weeks, a time of great anxiety for the parents.

There is also a technique called FISH (fluorescence in-situ hybridisation) based on genetic engineering methods. A slide of amniotic fluid is analysed using chromosome specific probes. Results can be available in a few days. Women still have to have an amniocentesis, as this is the method by which the amniotic fluid is collected.

Although Amniocentesis and CVS will tell parents for definite whether their child does or does not have Down's syndrome, the tests cannot identify all conditions, or guarantee a healthy baby.

WHAT HAPPENS NEXT?

No one can tell parents what to do or how to feel. The right decision for one family might be very different for another. Some people feel that they need to know for sure whether their baby has Down's syndrome and choose to have further testing, while others decide to wait and see.

For those who find out that they are carrying a baby with Down's syndrome, there are several options. Some families use this time to prepare for the birth of their baby with Down's syndrome. Down's Syndrome Scotland can help with information at this stage. Some babies are adopted. There are always families willing to adopt

children with Down's syndrome. Another choice is to have a termination.

Whilst each individual's right to make their own choice is respected, Down's Syndrome Scotland hope that parents will gather as much information as possible before making their decision. Down's Syndrome Scotland do not endorse the termination of pregnancy simply on the basis that the baby has, or may have, Down's syndrome.

Parents who would not consider a termination of the pregnancy should Down's syndrome be detected, do not usually undergo the amniocentesis as they would

not want to risk losing the baby. In some instances, people have a definitive test so that they can prepare themselves and their families for the birth of a baby with Down's syndrome. These parents will have weighed up the possibility of losing the child against the need to know for definite.

Around 80% of women who have an increased risk after Maternal Serum Screening go on to have an amniocentesis.

10% of those who have a positive amniocentesis continue with their pregnancy comfortable in the knowledge that their baby has Down's syndrome.

There can be the feeling of urgency about making decisions during pregnancy. People are often expected to respond quickly about how they want to proceed. Expectant parents should not feel that they are being rushed and should be given all the relevant information that is available to help them. Time should be taken in making any decisions. Another twenty-four hours may not change initial thoughts, but it will allow time to ensure that the decision is right for them.

Counselling should be available through the hospital. Down's Syndrome Scotland also offers support and further information.

A REMINDER OF THE MEDICAL TERMS AND THEIR MEANINGS

Screening – A blood sample or a scan which gives a risk factor of the baby having Down's syndrome. Screening is not conclusive and only further testing can give a definite diagnosis.

Testing – A procedure that can give conclusive evidence of Down's syndrome.

Invasive – Procedures which involve passing a needle into the womb to collect a sample of the amniotic fluid.

Maternal Serum – The mother's blood.

Amniotic fluid – The fluid contained in the amniotic sac which surrounds the baby.

Alpha fetoprotein – A protein produced by the baby that is present in the amniotic fluid.

Human chorionic gonadotrophin – A hormone produced by the placenta.

Oestriol – One of the female sex hormones produced by the ovaries.

False positive – Results of maternal serum screening indicate that the baby has Down's syndrome when in actual fact it does not.

False negative – Results of maternal serum screening indicate that the baby does not have Down's syndrome when actually it does.

Sandal gap – The space between the big toe and the second toe is wider than usual.

Chromosomes – Threadlike structures in the cells that contain the hereditary genes.

For further information contact

Down's Syndrome Scotland

158-160 Balgreen Road, Edinburgh EH11 3AU

Telephone: 0131 313 4225 Fax: 0131 313 4285

Email: info@dsscotland.org.uk Website: www.dsscotland.org.uk