Medicine for Managers

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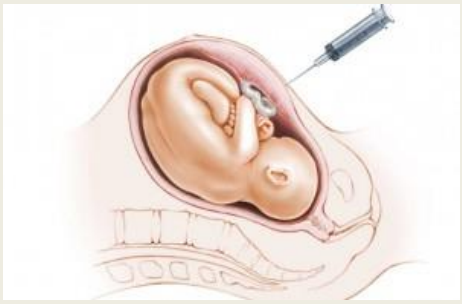
Down's Syndrome

There seem to be new, exciting and ground-breaking health discoveries every day. Last week, the brilliant Hospital for Sick Children at Great Ormond Street announced a blood test to assist in the diagnosis of Down's syndrome. This Non-Invasive Prenatal Test (NIPT) reduces the need for amniocentesis by 80% and is a simple blood test which identifies fragments of foetal DNA.

About 770 women a year give birth to a child with Down's syndrome in the United Kingdom and women, in whom the risk is greater than one chance in 150, is offered an amniocentesis.



This is a procedure in which a needle is inserted into the abdomen and through the wall of the amniotic cavity, which contains amniotic fluid which surrounds the developing foetus.

A sample of the amniotic fluid is withdrawn and which can be tested for genetic material defects through which a diagnosis can be made. One problem with the technique is that the intrusion into the amniotic cavity with the needle stimulates a miscarriage in about one woman in 200, a proportion of which were in women whose child had no genetic abnormality in any case. The blood test is 99% accurate for Down's syndrome.

The test is expected to be introduced into the NHS later this year because, although expensive, it is cost-effective because it reduces the need for the expensive and more risky amnniocentesis.

Currently all women are offered screening but it does not guarantee identifying Down's syndrome. Between eleven and fourteen weeks of pregnancy, a blood test, which is about 80% effective, together with a scan of the nuchal region of the skull (a nuchal translucency ultrasound scan – a nuchal translucency is a collection of fluid) can be offered.

Between fourteen and twenty weeks, which is too late for the combined test, a blood test can still be offered but is less accurate than the combined test. It is essential that the nature of any test, the possible results and the implications of such results should be discussed before they are undertaken. Following testing the parents will be notified. The test may show

that it is unlikely that the child will have Down's syndrome and the pregnancy proceeds as before. If parents are told that the baby is likely to have Down's syndrome then genetic counselling is essential to discuss the possible impact, options and to give the opportunity for referral to specialists who can provide more information about the condition and answer any questions.

So, what is Down's syndrome. In essence it is a genetic condition which usually results in some degree of learning disability and the development of characteristic physical features.

The child with Down's syndrome has an abnormal ***karyotype*** (that is the number of chromosomes which the child has). A patient with a conventional karyotype has 46 chromosomes which are distributed in 23 pairs (22 pairs called ***autosomes*** and one pair of sex chromosomes, which are either XX (for a girl) or XY (for a boy). In Down's syndrome about 93% exhibit ***trisomy 21***, the individual has forty-seven chromosomes because there are three of chromosome 21 instead of two. This is the most common chromosomal abnormality in new born infants. In about 5% extra chromosomal material is attached to one of the existing chromosomes but there is no separation. In this situation, called ***translocation***, there are therefore 46 chromosomes but the amount of genetic material is broadly the same as in trisomy 21. In about 2% the individual exhibits ***mosaicism*** which is where only some of the cells have the extra chromosome.

More information and support is available from:

The Down's Syndrome Association
2A Langdon Park
Teddington, Middlesex, TW11 9PS
Telephone: 0845 230 0372
www.downs-syndrome.org.uk/

Developmental delay is less marked in such children.

The physical variations associated with Down's syndrome are considerable and may be marked or minimal. However, most people with Down's do share a number of physical characteristics and developmental problems. These include a general floppiness and weakness which is the result of poor muscle tone, a protruding tongue, a flattened nose, a flattening of the back of the head and a low weight and length at birth. Other features may include broad hands and short fingers, a single palmar crease and widening of the space between the first and second toe.

All children with Down's syndrome will exhibit a degree of developmental slowing and learning disability. The usual milestones will be reached later with delays in sitting, standing, walking and talking. Each child needs to be assessed individually because virtually all will need additional help at school.

Any mother can give birth to a child with Down's syndrome but the risk of doing so increases with advancing age. A mother aged 20 has a one in 1,500 chance, whereas a mother over the age of 45 has a risk of one in 50 or greater.

Having a previous child with Down's syndrome or carrying affected genes adds further to the risk and if one of the parents has Down's syndrome then the risk is one in two.

In some cases the diagnosis is only made after birth and is often first suspected simply by the physical characteristics of the child.

Additional and specialised help may be required as the child grows but much of the time the child can integrate with the family to ensure that he or she has the same loving attention as any other child.

Children with Down's syndrome, like any other child, can be bright and happy and enjoy 'fun' activities. Education may commonly occur in a conventional school but, in some cases, more directed learning may be required.

In additional such children may need rather more regular physical checks-up because about 10% of children develop thyroid deficiency and heart problems may also occur (around half have a congenital heart problem).

Other problems include a range of ocular complications (long sight, short sight, cataracts, glaucoma), hearing disorders including glue ear and increased risk of infections. Gut problems such as constipation or diarrhoea and coeliac disease may also occur. Growth should also be monitored. However, many children are well and have no health problems.

Once adult many people with Down's syndrome can live independent productive lives, walking, talking, dressing and feeding themselves. Significant numbers work for a living. Others need more support in accommodation such as assisted housing with home wardens. For some, greater levels of support are needed.

People with Down's syndrome fall in love and enter meaningful relationships. For them it may be necessary to counsel carefully about the possibility of having a child and the likelihood of that child having Down's syndrome. Although the fertility rate is reduced in both sexes, pregnancies can occur successfully. Advice on effective contraception may also be important. Miscarriage is an increased risk.

The discovery by any parent that their child may have or does have Down's syndrome will initially be very frightening when they do not understand the implications and the decisions generated by the counsellors will be of major impart.

Those fears will revolve around the challenges of the child, how adulthood will be managed and what will happen when the parents are no longer there to provide support. Yet the children, who so often have a very happy demeanour and who love to have fun, can bring so much joy and happiness to a family.

Many do form a stable and successful relationship and live long and contented lives.

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