Down's syndrome

Down's syndrome (also known as Down syndrome) is a genetic chromosome problem. Someone with Down's syndrome has an extra copy of chromosome 21 in the cells of their body.

This causes the typical features of the condition. All people with Down's syndrome have some degree of learning disability. They are also at increased risk of having various other medical problems. Down's syndrome may be detected before birth (prenatally) or after birth (postnatally). It cannot be cured but treatment and support can help someone with Down's syndrome to lead an active life. A number of advice and support groups are available for people with Down's syndrome and their families and carers, such as the National Down Syndrome Society.

Understanding chromosomes and cell division

Chromosomes are found in the centre (nucleus) of a cell. They carry genetic information in the form of genes.

In general, each cell in your body contains 46 chromosomes arranged in 23 pairs. One chromosome from each pair is inherited from your mother and the other is inherited from your father. One of these chromosome pairs is known as the sex chromosomes because this pair determines our sex. Females have two of the same kind of sex chromosome (XX). Males have two different sex chromosomes (XY). The Y chromosome contains the male determining genes. So, a normal female is 46, XX and a normal male is 46, XY.

However, egg and sperm cells only have 23 chromosomes. This is so that, when an egg and a sperm meet during conception, the child that is produced also has 46 chromosomes in each cell.

New body cells are produced for growth and repair by cells dividing, producing two 'daughter' cells. This process is called mitosis. Each chromosome can duplicate an exact copy of itself so that each new cell that is formed has a full, identical set of chromosomes.

Cell division that produces reproductive cells (egg and sperm cells) is slightly different. The process is called meiosis. Four 'daughter' cells are produced that have half the number of chromosomes of the original cell - so 23 chromosomes (one of each pair).

If cells do not divide properly, either during mitosis or meiosis, one of the 'daughter' cells produced may receive two copies of a chromosome while the other cell receives none. So, two abnormal cells are produced. This error in cell division is called nondisjunction.

If nondisjunction occurs during meiosis, this can lead to an egg or a sperm having either an extra chromosome, or one less chromosome. This means that when it meets a normal egg or sperm at conception, the embryo produced (that will go on to develop into the fetus or baby) will have either one extra copy of that chromosome, or one less copy of that chromosome, in each of their body's cells. One extra chromosome is known as trisomy. One less chromosome is known as monosomy.

What is Down's syndrome?

Down's syndrome is a genetic chromosome problem. It was first described by John Langdon Down in 1866. He recognised the typical features that someone with Down's syndrome has (see below). There is no cure for Down's syndrome. It will affect someone throughout their life. Someone with Down's syndrome can usually be recognised by their typical facial features. It also causes learning disability and there are some medical problems that someone with Down's syndrome has an increased risk of having.

What causes Down's syndrome?

Someone with Down's syndrome has an extra copy of chromosome number 21 in the cells of their body. This is also known as trisomy 21. Trisomy refers to the fact that there are three copies of chromosome 21. Because there is an extra chromosome 21, there is extra genetic material in the body. This causes the typical features of Down's syndrome. There are different ways that trisomy 21 can occur.

Full trisomy 21

About 94 in 100 people with Down's syndrome have full trisomy. That is, all of the cells in their body have an extra copy of chromosome 21. Full trisomy 21 can occur in an embryo due to nondisjunction during meiosis as described above. The chromosomes of the mother and the father are normal. In this case, Down's syndrome is caused by the inheritance of an extra chromosome (either from the mother or the father) that occurs by chance at the time of conception.

Mosaic Down's syndrome

Mosaic Down's syndrome can also occur. About 2 in 100 people with Down's syndrome have mosaic Down's syndrome. In this case, only some of the cells in the body have an extra chromosome 21. People with mosaic Down's syndrome tend to be more mildly affected and have less obvious features of Down's syndrome. The degree of problems depends on the number of cells in the person's body that are affected by trisomy.

Translocation Down's syndrome

In about 4 in 100 people with Down's syndrome, it is due to a translocation. This means that one of the affected child's parents carries a translocated chromosome. That is, part of one of their chromosome 21s becomes attached to another chromosome. (It is often chromosome 14 that is affected.)

The parent is healthy and has no signs or features of Down's syndrome because they are not carrying any extra chromosome 21 genetic material. They have one normal chromosome 21, one chromosome 21 that has a piece 'broken off' or missing, one chromosome 14 that has an extra bit of chromosome 21 attached to it, and another normal chromosome 14.

However, the extra chromosome 21 material can be passed on to their offspring, so producing a child with Down's syndrome.

What are the features of Down's syndrome?

There are some facial and other physical features that are typical in people with Down's syndrome. There are also certain medical problems that can affect someone with Down's syndrome.

However, not everyone with Down's syndrome has all of these features or problems. Some people may only have a few and be mildly affected, while others can have many more. So, the severity of Down's syndrome can vary from person to person. Everyone with Down's syndrome is different. Features associated with Down's syndrome can include the following.

Typical facial features

It is usually quite easy to recognise that someone has Down's syndrome by their typical facial features which include:

- An oval-shaped face.
- Eyes that slant upwards and have an extra skin fold on the upper eyelid, known as an epicanthal fold. This skin fold covers the inner corner of their eye next to their nose.
- A small mouth and a tongue that often protrudes.
- The back of their head (called the occiput) is often flatter than normal.
- Their nose may be small and have a flat and low bridge.
- Ears may also be small and low-set.

Other outward physical features

These can include:

- A single crease across the palm of the hand (single palmar crease).
- Short fingers and a little finger that can curve inwards.
- Joints that seem 'looser' than normal.
- A bigger than normal space between the first and second toe (the 'sandal gap').
- Muscles that lack tone and can be quite floppy (this is often most noticeable in babies).
- Babies may have a low birth weight.

Problems with physical and mental development

Everyone with Down's syndrome will have some degree of learning difficulty. However, the level varies from person to person and some are more severely affected than others.

Delayed speech development and delay in motor development (sitting, crawling, walking, potty training, etc) can occur.

Feeding difficulties are also very common for children with Down's syndrome.

Heart defects

Around half of babies with Down's syndrome will have a congenital heart defect (that is, a heart problem that they are born with). The most common problem is an atrioventricular septal defect (AVSD). This is a hole in the muscle that usually separates the different chambers in the heart. There can also be problems with the valves in their heart. If someone has an AVSD, it means that the blood flow around their heart becomes abnormal.

An AVSD can usually be fully corrected with surgery if it is detected early. Therefore, all babies with Down's syndrome should have a thorough examination of their heart shortly after they are born. If there is any suspicion of a heart problem, doctors will usually refer the baby for an ultrasound scan of their heart, called an echocardiogram. Other heart problems can sometimes develop later in life in those with Down's syndrome. Therefore, a regular check-up of the heart is needed.

Typical behaviour

People with Down's syndrome tend to be naturally very warm, cheerful and gentle. They can be quite spontaneous in their behaviour. Sometimes this type of behaviour can make them quite vulnerable and put them at risk. However, it should be noted that people with Down's syndrome can experience moods and feelings just like everyone else.

Other problems

Someone with Down's syndrome is also at increased risk of developing other medical problems or conditions. These can include:

- Vision problems including short-sightedness, long-sightedness and abnormal alignment of the eyes (squint).

- Ear infections and hearing problems. It is important that hearing problems be detected and treated, as they can affect a child's ability to learn.
- Growth, teeth and bone development may all be affected.
- Leukaemia is more common than average in children with Down's syndrome. This is a type of cancer that affects the white blood cells that usually fight infection.
- Obesity can often develop during adolescence.
- Frequent infections such as coughs, colds and eye infections are common in people with Down's syndrome. They may be due to problems with immune system development.
- Thyroid problems including an underactive thyroid gland.
- Diabetes.
- Premature ageing can occur leading to the formation of cataracts and age-related hearing problems. Early-onset dementia can occur. People with Down's syndrome don't have an increased risk of developing dementia; its just that if they do develop dementia, it tends to develop 20 to 30 years earlier than it normally would.
- Fertility is usually reduced in someone with Down's syndrome.
- Oesophageal or duodenal atresia can occasionally affect some babies born with Down's syndrome. In this condition, the gullet (oesophagus) or the first part of the intestine (duodenum) is like a blind-ending pouch. That is, the oesophagus does not connect as it normally would to the stomach, or the duodenum does not connect as it normally would to the rest of the intestine. Feeding problems occur as milk collects in the blind-ending pouch. Pneumonia can occur if fluids flow over into the lungs.
- Hirschsprung's disease can sometimes affect a baby with Down's syndrome. There is a problem with the muscles in the wall of the bowel. Nerves usually make these muscles contract to help to push stools (faeces) along but they are missing in Hirschsprung's disease. About 2 in 100 babies with Down's syndrome have Hirschsprung's disease.

How common is Down's syndrome?

Down's syndrome is the most common and best known chromosome problem in humans. About 1 in 1,000 babies born in the UK have Down's syndrome. It is thought that around 60,000 people with Down's syndrome are living in the UK. Down's syndrome is the most common cause of learning disability in the UK.

There is no test that you can have before conception that will predict whether you will conceive a baby with Down's syndrome. Anyone can have a baby with Down's syndrome but a woman's risk increases as she becomes older. Having said that, most Down's syndrome babies are born to younger women. This is because, compared with older women, younger women more often have babies.

Large studies have been carried out to look at the risk of Down's syndrome related to a mother's age and have shown that:

- A 20-year-old woman has a 1 in 1,500 chance of having a baby with Down's syndrome.
- A 30-year-old woman has a 1 in 800 chance.
- A 35-year-old woman has a 1 in 270 chance.
- A 40-year-old woman has a 1 in 100 chance.
- A 45-year-old woman has a 1 in 50 chance or greater.

However, it should also be remembered that the risk of *conceiving* a baby with Down's syndrome is actually higher than the above figures. This is because about three quarters of embryos or developing fetuses with Down's syndrome will never actually develop fully and so a miscarriage will occur before a baby is born.

Prenatal and postnatal diagnoses of Down's syndrome were found to have increased from 1989 to 2008 in England and Wales. This is despite little difference in birth rates. However, the numbers of babies born with Down's syndrome did fall slightly due to improved and more widely used prenatal screening. This screening has led to increased rates of detection of Down's syndrome during pregnancy and, as a result, termination of pregnancy. Without this improved screening, because women are generally having babies at a later age, it is thought that the numbers of live births with Down's syndrome would have otherwise increased by almost half.

If you have previously had a baby with Down's syndrome because of trisomy 21, your risk of having another affected baby is increased. About 1 in 100 women with a previous baby with Down's syndrome will have another.

How is Down's syndrome diagnosed?

Down's syndrome can either be diagnosed before birth (prenatally) or after birth (postnatally).

Prenatal diagnosis

If you are pregnant, there are various Down's syndrome screening tests available during your early pregnancy to look at your risk of having a baby with Down's syndrome. All pregnant women should be given the option, or choice, of having one of these tests. However, these tests are not compulsory.

Screening tests for Down's syndrome usually involve a blood test and a special ultrasound scan called a nuchal translucency ultrasound scan. This scan is carried out in a similar way to other ultrasound scans that you will have during pregnancy. However, it focuses on measuring the thickness of fluid that collects behind your baby's neck while they are developing in your womb. It is normal for all babies to collect some fluid here but babies with Down's syndrome tend to collect more fluid than normal.

It is important to understand that these screening tests do not give a definitive answer as to whether your baby does have Down's syndrome - they just give a risk. If this risk is deemed to be high, a diagnostic test can be done prenatally to determine if a developing baby actually does have Down's syndrome or not.

There are two main prenatal diagnostic tests that can be done: amniocentesis and chorionic villus sampling. However, these diagnostic tests do carry a small risk of miscarriage. See separate leaflets called Down's Syndrome Screening (Prenatal Screening), Amniocentesis and Chorionic Villus Sampling (CVS) for more details.

Postnatal diagnosis

Down's syndrome may be suspected shortly after birth because of the typical features that a baby with Down's syndrome has (see above). Diagnosis is confirmed by genetic tests to look at the karyotype of your baby. This is essentially their genetic make-up. It involves your baby having a blood test. The chromosomes in their blood can then be examined to look for an extra chromosome 21.

Are any further investigations needed?

If your baby is diagnosed with Down's syndrome, various other tests may be suggested by their doctor to look for medical problems associated with the condition. For example, an ultrasound scan of their heart (echocardiogram) can show if they have any heart defects. A hearing test known as a brain stem auditory evoked response can detect hearing problems. Eye tests can look for visual problems.

What is the treatment for Down's syndrome?

There is no treatment that can cure Down's syndrome. It is something that you have for life. Treatment is aimed at monitoring for any complications and treating these where possible. Most of the complications associated with Down's syndrome are treatable. With treatment and support, someone with Down's syndrome can lead a very active life.

Early intervention programmes from birth are important for a baby born with Down's syndrome. Such programmes can include speech and language therapy, physiotherapy, etc. They can help with the learning and development of a child with Down's syndrome. The earlier that such help and care are given for any problems, the better the long-term outcome is likely to be and the more likely that person is to reach their full potential.

A number of different healthcare practitioners are likely to be involved in the care of someone with Down's syndrome. The people involved will depend on the problems that are faced by the individual with Down's syndrome. For example, the team of people involved can include a doctor specialising in the care of children (a paediatrician), your GP, a heart specialist (cardiologist), an eye specialist (ophthalmologist), a speech and language specialist, a physiotherapist, an occupational therapist, a social worker, etc.

Someone with Down's syndrome will need regular medical follow-up. In babies and children it is important to monitor their growth and development. When their teeth develop, they will also need regular dental checks. Monitoring for heart problems, hearing problems and eye problems is also important. A healthy, balanced diet is needed and exercise should be encouraged to reduce the risk of weight gain and obesity.

Will someone with Down's syndrome be able to lead an independent life?

To some extent, this does depend on how severely someone with Down's syndrome is affected. While some children with Down's syndrome may need to go to a school that caters for their special needs, many are able to go to a mainstream school. Likewise, as they become older, many people with Down's syndrome are able to cope with some kind of job. People with Down's syndrome are also able to make friends and have relationships.

Many people with Down's syndrome will never be able to live completely independently. For some, arrangements such as sheltered housing with close availability of carers and support may be possible. However, other people with Down's syndrome may need much higher levels of support and care.

What is the outlook (prognosis)?

Of babies born with Down's syndrome, life expectancy is improving due to better detection of medical problems and advancements in medical treatments. At least half of people with Down's syndrome now live into their 50s and 60s.

Congenital heart problems (heart problems a person is born with) are often the cause of death in those who do not survive.

Support for parents and carers

Having a baby with Down's syndrome can be a difficult thing for some parents to cope with. However, you should never feel that you are alone. Great support and advice for parents who have a child with Down's syndrome are available through the Down's Syndrome Association and a number of other support groups detailed below.

Further reading

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