

Down's Syndrome Scotland

AGES AND STAGES OF DOWN'S SYNDROME

What is Down's syndrome?

An English doctor, John Langdon Down, first described Down's syndrome in detail in 1866. It is a congenital condition, which randomly affects about 1 in 700 babies born throughout the world, male and female alike.

'Syndrome' means a group of recognisable characteristics occurring together. A 'congenital syndrome' is one that is present at birth that cannot be 'caught' later on.

Down's syndrome is caused by an extra chromosome appearing on the 21st pair. (There are 47 chromosomes in each cell). This additional chromosome, because of the genes it contains, causes an excessive amount of certain proteins to be formed in the cell, this in turn disturbs normal growth in the body of the foetus.

All children with Down's syndrome have the extra critical portion of chromosome 21 in their cells, but the amount of chromosome present, and the way in which the error comes about, can take one of three forms.

Trisomy 21 – The most common form of Down's syndrome (95%). There is an extra, whole chromosome 21 in every cell of the body. This type is not hereditary, although the chances of having a second child with Down's syndrome are 1 in 100, which is higher than the general population.

Translocation Down's syndrome - This type occurs when the extra chromosome attaches itself to another chromosome, in every cell. Each cell contains 46 chromosomes, but one is much larger, as it has the extra chromosome attached. Translocation Down's syndrome affects 3-4% of people with Down's syndrome and can be hereditary.

Mosaic Down's syndrome - This is the rarest form of Down's syndrome, affecting 1 – 2%. Some cells contain the extra chromosome, and some do not. It is impossible to check how many cells are affected but there is evidence to suggest that the fewer cells affected, the lesser the degree of learning disability will be.

Physical features associated with Down's syndrome

There are as many as 120 features described in Down's syndrome but no one individual will carry them all, indeed most people have only 7 or 8.

Face – usually rounded with a flatter profile.

Head – back of the head is slightly flattened (brachycephaly).

Eyes – slant slightly upwards. There is often a small fold of skin that runs vertically between the inner corner of the eye and the bridge of the nose (epicanthic fold) – this can also be seen in babies who do not have Down's syndrome. The fold becomes less prominent as the child gets older. Eyes often have white or yellow spots around the iris (Brushfield spots) – these do not affect vision and can also be seen in other children – again, they often disappear in later life.

Neck – usually short and stocky, there may be excess skin over the back of the neck in newborn babies but this is often disappears as the baby grows.

Mouth – mouth cavity is smaller than average and the tongue is usually thicker and larger, which leads to the tongue hanging out of the mouth. Children are taught to hold their tongue in their mouth once they have the control to do so.

Hands – broad with short fingers, the little finger often has only one joint and curves inwards. There is only a single crease running across the palm.

Feet – tend to be broad and short, toes are widely spaced.

Tone – limbs and necks of infants with Down's syndrome are floppy – this improves as the child gets older. Low tone does not mean muscle weakness.

Body size – babies are usually smaller than average at birth and therefore weigh less, children are smaller than their peers. Adult stature is at the bottom end of the normal range.

Despite these common characteristics, it is important to remember that people with Down's syndrome will more closely resemble their parents and siblings in appearance, than other people with Down's syndrome.

Health problems associated with Down's syndrome

In the 1950's, the life expectancy for a person with Down's syndrome was 15 years – this has increased to 60+ years. This is due to increased knowledge about the health problems associated with Down's syndrome, the development and use of heart surgery, immunisation programmes and the use of antibiotics.

The Heart – around 1 in 3 babies with Down's syndrome will have a congenital heart defect. Although some defects will cause no problems and eventually heal themselves, most require surgical intervention and long term monitoring of the condition. Newborn babies are checked for heart problems and then examined again at 6 weeks.

Hypothyroidism – (under active thyroid) causes lethargy, lack of concentration, weight gain, dry coarse skin, memory impairment and intolerance of cold. Newborn babies are tested and ongoing screening should be done at least every 2 years throughout life. Treatment is thyroid supplement in tablet form – however if left untreated the condition can lead to heart disease and mental disturbances.

Leukaemia – around 1 in 100 children with Down's syndrome develop leukaemia, onset is usually between the ages of 1 and 4 years.

Upper respiratory tract infections – Children with Down's syndrome are prone to more coughs, colds and ear infections due to their relatively narrow ear and nasal passages – antibiotic treatment may be needed. Older people are also more susceptible to chronic respiratory conditions.

Atlanto-axial instability – there is increased mobility of the 2 bones directly under the skull (atlas and axis), which in extreme cases can become compressed. Cord compression can happen gradually or rapidly causing dislocation and even paralysis. The annual physical examination should include a check for signs of cord compression (neck pain, restricted neck movement, unsteadiness in walking and deterioration in bowel and bladder control). Children should not be barred from sporting activities **except;** intensive trampolining, intensive horse riding and high diving. When undergoing any treatment requiring anaesthesia a collar should be used to keep the neck stable. Collars should also be used if a person with Down's syndrome is involved in a road traffic accident, until examination has taken place.

Ears – children with Down's syndrome are very prone to ear infections particularly 'glue ear'. Treatment is by insertion of grommets, the problem is often recurrent throughout childhood. Hearing should be monitored regularly (at least every 2 years) throughout life.

Eyes – squints are very common and need to be corrected, by wearing glasses or surgically – if left untreated the good eye will take over and the lazy

eye can become blind. Cataracts occur in 46% of adults and people with Down's syndrome are prone to Keratoconus (where the cornea becomes cone shaped) – cataracts are treatable if identified early enough and regular eye checks are advised.

Muscle tone – very poor in infants and can cause problems with sucking and weight gain.

Dental – the milk teeth erupt later and stay longer, suffering more wear and tear – this in turn makes gum disease more common. Adult teeth may be irregularly spaced. Regular dental check ups are advised.

Screening and Testing

Various tests can give pregnant women more information about the baby they are carrying. Maternal serum screening is offered to all women at about 15 – 16 weeks into the pregnancy. A blood sample taken from the mothers arm can be screened for Down's syndrome.

The blood sample is tested for certain proteins and hormones, usually alpha fetoprotein (AFP), human chorionic gonadotrophin (hCG) and oestriol (uE3). Levels of these, assessed with the mother's age, indicate the 'risk' of the baby having Down's syndrome.

An 'increased risk' or 'positive screening' means that there is a greater than expected chance of the baby having Down's syndrome. It is still only a possibility and the likelihood is that the baby will not have Down's syndrome.

A 'low risk' or 'negative screening' result only means that it is very unlikely that the baby will have Down's syndrome, it does not rule out the possibility.

A definite diagnosis of Down's syndrome can be made through further testing such as amniocentesis or chorionic villus sampling (CVS). For amniocentesis, a needle is passed into the womb to collect some of the fluid surrounding the baby. For CVS, cells are collected from around the placenta. Both procedures carry around 1 in 100 risk of miscarriage.

Results can take up to three weeks, although most hospitals try to inform the parents as soon as possible. There is a new technique called FISH (fluorescence in situ hybridisation), which is based on genetic engineering methods. A slide of amniotic fluid is analysed using chromosome specific probes. FISH can provide results in 24 hours.

Development

All children with Down's syndrome will have some degree of learning disability, mild in some, more marked in others. It is impossible to predict at birth the extent of a baby's learning difficulty.

In people with Down's syndrome, the brain is formed differently, making the process of learning less efficient. Children need to be encouraged to develop to their full potential, but not driven beyond their capabilities, or put under undue pressure to achieve.

Children with Down's syndrome always go forward in their development – physically, intellectually and emotionally – becoming more competent with each passing year. However, progress is at a slower rate to other children.

Children with Down's syndrome vary in their rate of development; they go along at a slower speed than other children and arrive at each stage of development later, staying at that stage for longer. It does not matter how far behind other children a child with Down's syndrome is developing, what is important is that they are maintaining a constant rate of development, however fast or slow that may be.

The development of children with Down's syndrome proceeds in a series of stops and starts. Stops are when children do not appear to be learning any new skills. These lulls in development are very important – they are when the child is consolidating what they have learnt by practising their new skill. It is a time when they are beginning to develop the components for the next skill. The milestones that children reach are the tip of the iceberg with far more development going on unseen below the surface.

Many children benefit from early intervention – this is the term used to cover the physiotherapy, occupational therapy, speech and language therapy and special educational help, given to pre-school aged children.

These professionals work with the child and parents, devising programmes that are followed in the home. Any areas where the child is having specific difficulties are targeted to prevent possible delays with future development.

Nowadays, most children with Down's syndrome attend mother and toddler groups, nursery, play groups and other clubs alongside their peers. These activities have many benefits; the child will develop coordination and strong muscles as well as learning good social skills, communication and becoming more independent from their parents. Most importantly, they are taking their place within their community.

More and more children with Down's syndrome are attending mainstream school. With the correct type, and level of support many will develop academic skills.

Young People

Whilst some children continue with mainstream schooling in their secondary school, this is often the age at which the gap in development between a child with Down's syndrome and other children starts to widen, for this reason

some young people with Down's syndrome choose to go into more specialised education.

Young people should not be pressurised into continuing with mainstream schooling if they do not feel that it will be right for them – putting pressure on a child to attain academic skills may be fruitless and result in the young person losing confidence in their abilities and self-worth. Neither should a young person be deterred from continuing with mainstream if they believe that it is the right thing for them.

Adolescents with Down's syndrome go through the changes associated with puberty at the same time and in the same order as their peers. Sometimes their lack of emotional maturity makes this a very difficult time and they need plenty of information, support and guidance to help them through. Information about relationships and sex, appropriate and inappropriate behaviour, hygiene and bodily changes needs to be presented in an effective way that the young person will find easy to understand. Using drawings, pictures, books and video's will help to illustrate difficult concepts.

At this time, young people with Down's syndrome become aware that they are different and will need help to understand how Down's syndrome has affected their development. A positive self-image is very important, clothing and hairstyles should be up to date and age appropriate.

Activities and social lives need to reflect the age of the young person; more opportunities to have some independence from the family and to be an individual, should be encouraged – young people may need more support to access social facilities, but once there, parents need to stay in the background.

Information about keeping safe is important to ensure that individuals are not exploited or abused. Taken gradually, with plenty of opportunities for practise, young people will not face situations unprepared, and risks will be minimised.

From the age of 14 years, the future needs assessment process will begin – involving the young person, their parents and school staff. At school leaving age, the young person needs support to make decisions about what to do next. Many people with Down's syndrome stay on at school until they reach 18, after which options include college, youth training, work experience or day resources.

Further education can offer the chance to increase 'life skills' and independence. Some colleges have a good range of integrated courses where people with Down's syndrome can study alongside other students. Other courses are more specifically geared towards people with learning disabilities.

Many older adults with Down's syndrome attend resource centres (day centres); these are changing so that they can provide people with a range of activities and settings throughout the week, which reflects the abilities and interests of the individual.

For many people with Down's syndrome, getting satisfying employment is extremely important. There are schemes around which offer on the job training so that a person with learning disabilities can receive help and support on the job until they have mastered all the skills necessary to do the job independently.

Unfortunately, there are issues around claiming benefits if working and this does prevent people from being able to hold down full time employment. For this reason, many people do a variety of activities throughout the week such as part time work combined with a college course.

Adulthood

Within our society, people with Down's syndrome have the same rights as everyone else. The right to vote, the right to marry, the right to work, the right to access good medical care, the right to housing etc. People with Down's syndrome have the same dreams and aspirations about how they want to live their lives, as anyone else. These dreams can be achieved if each individual is given the right type and amount of support that they need. Some people require constant assistance in their daily lives, others manage perfectly well with very little help.

People with Down's syndrome have a lot more choices available to them nowadays (although there are still areas for development), in order to make these choices, information has to be presented in an appropriate manner and experiences offered. Adults with Down's syndrome continue to learn new skills throughout life. Making informed choices takes practise, particularly if someone else has always made the decision on their behalf.

Important issues for adults with Down's syndrome are; relationships, where to live, who to live with, what type and amount of support they need, whether to go to college or work (or both), how to spend their free time, having enough money, being with friends, having holidays and being able to enjoy the lifestyles that they have chosen.

Until more recent years, many adults with Down's syndrome continued to live at home with their parents. Nowadays, young people often prefer to live independently from their families, although they may move away from home at a slightly later age than other young people.

There are various housing options available - many homes are owned and staffed by housing associations. People can chose to live within a smaller or larger group and various levels of support are offered, depending on individual needs. Some people chose to live alone, for example in council provision, others buy their own home.

With the direct payments scheme (where Local Authorities give an individual the money to purchase services, rather than providing that service), people have more chance to buy the best and most appropriate support for them as an individual. Hopefully, more people with learning disabilities will be able to take advantage of this scheme in the near future.

With varying degrees of support, adults with Down's syndrome are becoming increasingly independent and leading fuller and more active lives. Unfortunately, there is still a tendency for people to talk to them as if they were children, this is wrong and hurtful – they are adults and should be treated accordingly, with respect and dignity.

Growing older

People with Down's syndrome are now living longer with a life expectancy of 50 – 60+ years (compared with 15 years in the 1950's). There is a misconception that people with Down's syndrome age prematurely, but there is no firm evidence to support this. Much of the research was carried out on people who had spent their whole lives in large institutions where they had very little physical or mental stimulation. It is understood that people who lead busy and active lives age more slowly.

People with Down's syndrome in the 50 – 60 age bracket show the same typical ageing pattern as the general population – a decline in memory, vision, hearing and motor function. After the age of 60, there is usually deterioration in vision (cataracts become common), hearing and skin. Other health problems that are more common in older people with Down's syndrome are chronic respiratory conditions, arthritis, epilepsy, diabetes and hypothyroidism.

Another feature of ageing in adults with Down's syndrome is the onset of epileptic seizures – at the age of 50, 1 in 10 people will have had a seizure – medication will be prescribed to control the epilepsy. Depression is also common in older adults and is usually a result of bereavement or changes in living situations.

At this time, there are still a lot of older people with Down's syndrome who live at home with their parents (and therefore leading the lifestyles of people much older than themselves). Sometimes this is for practical reasons such as the adult taking on a caring role within the home. Often, the reason is that there was never a right time for the person to move away from home.

Inevitably, these adults will experience bereavement and will require a lot of support to come to terms with their loss. For people who have remained at home there may be the additional loss of having to leave the family home – this will be even more stressful if they have never spent any time away from home.

Preparation work, with the individual and their family is extremely important. Although it is very upsetting for all involved, some information about death

and thought about what the person will do afterwards, will help when the time comes. If parents are involved they can help to put any fears that their son or daughter may have to rest. Decisions about the adult's future home and care needs can be arranged and finances put in order.

When adults with Down's syndrome have had the opportunity to make plans about what will happen once their parent is not there, they feel much more secure. Individuals who are not given this opportunity say that they are 'confused' and 'terrified' about what the future holds for them.

Dementia

As life expectancy has increased, the link between Down's syndrome and dementia has emerged. Research has shown that nearly all adults with Down's syndrome have Senile plaques and neurofibrillary tangles in their brains, which are similar to those seen in Alzheimer's type dementia.

Although most people with Down's syndrome have this neuropathology, only 50% will go on to develop dementia – the reason for this is not yet known. Diagnosis is difficult because there can be numerous other medical reasons why a person with Down's syndrome may show some of the signs of dementia (i.e. confusion, withdrawal, disorientation, irritation). Possible factors being hypothyroidism, deteriorating vision, depression.

In the general population, memory changes are the first outward sign that a person may have dementia, in many cases the person themselves is aware that something isn't right. For most people with Down's syndrome, the first signs are changes in behaviour – for example, a normally placid person gradually becomes more and more angry and aggressive.

Changes due to dementia are likely to happen over a period of time with carers becoming gradually aware that something is going on. When changes occur over short period, it is likely to be a physical problem that can be treated.

Dementia progresses much more rapidly in people with Down's syndrome than in the general population. Early diagnosis is very important so that people are given the opportunity to try current medication and treatment. Modern medication can slow the progress of dementia but it works best if it is prescribed in the initial stages.

When dementia is confirmed, it is better if the person is not automatically removed from their living environment – gradually adapting the place to meet the needs of the person works the best until the very last stages of dementia when specialised nursing care may be necessary.

A – Z of the professionals and how they can help

From birth, people with Down's syndrome will have contact with a variety of professionals from health, education and social services. Some people require very little intervention and prefer to get on by themselves, others need physical assistance and ongoing support.

Although some of these professionals will have a great deal of experience of working with people with learning disabilities, others will have had very little. However, more and more professionals are coming into contact with people with Down's syndrome as access to primary care and mainstream services continues to improve.

Audiologist – Usually has a clinic within a hospital. It is recognised that traditional testing methods are not always reliable and many clinics employ new techniques in order to accurately assess hearing.

Cardiologist – A doctor who specialises in disorders of the heart.

Community Nurse Learning Disabilities – A registered nurse in the field of learning disability, usually attached to community learning disability teams. They give information, support, assessment and therapy.

Dentist / Orthodontist – Some areas have specialist dental services but the current impetus is for people with Down's syndrome to be seen by a local dentist whenever possible, with input from a specialist if problems occur.

Dietician – Usually based in hospitals although occasionally in health centres. Provide advice and information about cooking and feeding – people with Down's syndrome tend to become overweight and children are prone to constipation.

Educational Home Visitor – A trained teacher attached to the regional psychological service. Works in the home planning an educational programme for the parents to undertake with their child.

Educational Psychologist – Works on the areas of learning and behaviour.

G.P. – Offer a general medical service and can be a source of support.

Genetic Counsellor – There are 4 regional genetic centres in Scotland. Many new parents of babies with Down's syndrome seek genetic counselling before having further children.

Health Visitor – A nurse with specialist training in advising families with babies and young children, who works closely with the G.P. Health Visitors carry out development checks.

Occupational Therapist – Breaks difficult tasks down into smaller, more manageable steps. The steps are then built up gradually until the whole activity is completed. Works on the activities of daily living.

Ophthalmologist – Eye specialist.

Paediatrician – A doctor specialising in the care of children. Will monitor the baby's development and general health. Will undertake the tests and check ups that are routinely done for all children with Down's syndrome.

Physiotherapist – Works with posture and movement to develop muscle tone and balance.

Speech and Language Therapist – Works on ways of promoting the development of communication and listening skills.

Social Worker – Helps the person with Down's syndrome and their family in areas such as accessing respite or long-term housing, applying for benefits, getting a day resource place etc.

Teacher – Many children with Down's syndrome attend mainstream school and often receive help from the learning support assistant. In special educational schools, teachers have usually had specialist training.

Summary

The general public is now much more aware about and accepting of, people with Down's syndrome. There has been a lot of publicity about Down's syndrome over the years and people with Down's syndrome are taking a more active place within their local communities.

Some myths and misconceptions still remain; 'All people with Down's syndrome are musical', 'they are all affectionate', and 'they are always happy' – not true, Individuals with Down's syndrome are as diverse as other people, anyway, who do you know that doesn't like music!

There is a tendency for the public to regard them as child-like, this leads to over familiarity and patronising behaviour towards an adult with Down's syndrome. People with Down are syndrome want to be treated with respect, as adults. They want to be able to make choices, exercise their rights and lead fulfilling lifestyles.

In their daily lives, people with Down's syndrome are encouraged to behave appropriately towards other people, to mind their manners and not do anything that will draw any attention to them. If anything, they are expected to behave like model citizens so that society will accept them, and yet, it is still highly unlikely that a group of adults with Down's syndrome would be

tolerated in a pub, unless there was a 'normal' person with them. It is important to remember, that people with Down's syndrome have the right to access places independently and to do so without fear of negative reactions from the general public.

Attitudes are changing and will continue to change. Organisations such as Down's Syndrome Scotland, are working hard to educate the public about Down's syndrome, the Scottish Parliament is producing new legislation to benefit people with learning disabilities and there is a great deal of collaboration and cooperation between the relevant agencies to help ensure that people with learning disabilities lead full and enjoyable lives.

Every person with Down's syndrome is a unique individual, full of potential.