



**Down's Syndrome
Association**

A Registered Charity No. 1061474

Health Series



Orthopaedic issues



The
Down Syndrome
Medical Interest
Group

A Down's Syndrome Association publication

Our resources and Information Team are here to help

Please see our website for up-to-date information: www.downs-syndrome.org.uk

If you would like to talk about any of the issues raised in this resource, then please get in touch with our helpline by calling 0333 1212 300 or by emailing us on info@downs-syndrome.org.uk.

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The Down's Syndrome Medical Interest Group (DSMIG)

This resource has been produced in collaboration with the Down's Syndrome Medical Interest Group (DSMIG).

DSMIG was launched in 1996 and is a registered charity. It is a network of healthcare professionals – mainly doctors – from the UK and Republic of Ireland whose aim is to share and disseminate information about the medical aspects of Down syndrome and to promote interest in the specialist management of the syndrome.

Musculoskeletal problems are commonly seen in people with Down's syndrome due to low muscle tone, lax ligaments and hypermobile joints.

This is due to the presence of atypical collagen, which is an important supporting structure in muscles and joints. This is believed to be caused by the overexpression of the genes for collagen type VI, COL6A1 and COL6A2, encoded on chromosome 21.

Collagen type VI is present in almost all connective tissues i.e. ligaments, muscle, tendons, bone and cartilage.

This resource focuses on the orthopaedic issues commonly seen in people with Down's syndrome except neck instability which is covered in depth by another DSA resource.

Spinal problems

Scoliosis

Scoliosis is the abnormal rotation of the spine causing a sideways curvature of the back. Severe scoliosis can cause back pain and can compromise lung function. In the general population the incidence is between 1.5 – 3%, in people with Down's syndrome it occurs more commonly with an incidence between 7 - 9%. It is more commonly seen in children who have undergone open heart surgery or a thoracotomy. It commonly develops in late childhood to early adolescence and tends to progress during periods of skeletal growth, although it may develop later in adulthood. The management is similar to the general population, with monitoring, the possibility of bracing in some cases and surgery reserved for more severe cases.

Cervical spondylosis

Cervical spondylosis or arthritis in the joints of the cervical spine (neck bones) is present in over 80% of people above the age of 60. In people with Down's syndrome due to premature ageing, lax ligaments and hypermobility symptoms may appear much earlier by the age 40 or earlier. The person may complain of neck pain or stiffness, unsteadiness whilst walking, sometimes headaches and visual disturbances. It is important to recognise the signs and symptoms as often the diagnosis and appropriate management is delayed. The treatment is similar to that of the general population which includes physiotherapy, management of pain, soft collar and if required steroid and anaesthetic block injection to the affected joints.

Lumbar Spondylolysis and Spondylolisthesis

Lumbar spondylosis is the term given to a stress fracture in the bones of the spine, this most commonly affects the 5th and sometimes the 4th lumbar vertebrae (lower back region). If the bones become misaligned and shift out of position, it is called spondylolisthesis. This may result in low back pain, stiffness, difficulty in maintain posture and difficulty in walking.

Lumbar spondylolysis is reported to be present in 3 to 6 percent of the general population with 2.7 to 8.4 percent affected by spondylolisthesis. A case series in people with Down's syndrome reported 18.2 percent had spondylolysis and 34.6 percent, spondylolisthesis. It is postulated that an increased body weight, decreased bone mineral density and laxity of ligaments contributes to the increased incidence. It is important to recognise and manage the condition early to prevent irreversible long term complications.

Hip problems

Hip instability

The stability of the hip depends on the bony anatomy, the capsule and ligaments around the hip joint. Excessive capsular laxity together with low muscle tone predisposes children with Down's syndrome to an increased incidence of hip subluxation and dislocation. Hip instability is reported to be present in about 2 and 5 percent of children

with Down's syndrome. Under the age of two years the hips are usually stable, but as the ligaments around the hip joint are lax and hypermobile, the ball at the upper end of the femur (the thigh bone) may move in and out the joint (subluxation). This is usually painless and becomes "habitual" and may in the long term lead to fixed dislocation, where the head of the femur lies completely out of the hip joint.

In the authors' experience this happens gradually and is very unlikely to be sudden and painful. Children with Down's syndrome usually do not require a general anaesthetic for reduction. Reconstructive hip surgery can be done but does not address the underlying abnormality, i.e. excessive capsular laxity. The aims of surgery are to realign the bony anatomy to enhance stability and keep the femoral head contained in the acetabulum. There is some evidence that simply realigning the proximal femur is enough, but any surgery in people with Down's syndrome needs to be carefully balanced with its benefits and risks; as surgery is not without risks, particularly with complications related to poor healing, infection and recurrent dislocation. The rehabilitative period may be prolonged requiring immobilisation for up to 3 months.

Slipped Upper Femoral Epiphysis (SUFE)

This is a rare condition of the hip that usually occurs during the adolescence growth spurt, but may occur in younger children, if there are associated endocrine abnormalities, e.g. thyroid disorders. Obesity is an additional risk factor. The top of the femur (thigh bone) above the growth plate slips in a backward direction, hence the term Slipped Upper Femoral Epiphysis.

The diagnosis is often delayed as the person often presents initially with knee pain as referred pain from the hip. In young people with Down's syndrome diagnosis may be even further delayed if there are difficulties of effectively communicating with the young person during the clinical examination.

The mainstay of management is early surgical intervention within the first 24 - 48 hours to prevent any further slippage of the head of the femur. A delay in treatment can result in a more complex management. However, in young people with Down's syndrome there is higher complication rate of infection, avascular necrosis, i.e. the blood supply to the head of the femur is permanently compromised.

Perthes Disease

Perthes disease is a rare condition of the hip occurring due to the disruption of the blood supply to the femoral head (the ball at the upper end of the thigh bone). This causes a cycle of bone death, repair, and eventually remodelling, however during this period the femoral head collapses and distorts. Over time as the blood supply normalises the bone repairs and remodels itself. The cause is unknown; it is more common in boys. The exact incidence of Perthes Disease in children with Down's syndrome is not known, though it is believed to be increased.

It can occur during anytime in childhood, more commonly between 4-10 years and the outcome is much better at a younger age. Children present with pain and limitation of movement in the hip, relieved upon resting.

The aim of treatment is to keep the hip joint moving to allow the femoral head to remain in the acetabulum, a process known as “containment”. This may involve simple physiotherapy, plaster casts, or surgery. The management depends on the age of the child and the stage of the disease and this does not differ in children with Down’s syndrome.

Total Hip Replacement (THR) in Down’s syndrome

The end stage of hip disorders can result in severe osteoarthritis, with severe pain and immobility. About 28% of adults with Down’s syndrome are reported to have hip problems. Total hip replacement should be carefully considered with the goals to restore independence and improve quality of life. Recent evidence suggests that outcome of surgery in people with Down’s syndrome is reasonable, though not without its challenges.

Knee problems

Knee cap instability (Patellofemoral Instability)

In people with Down’s syndrome due to ligamentous laxity, low muscle tone and possibly due to an increased incidence of knock knees, the knee cap can move away from the front of knee joint (Patellofemoral Instability), occurring in about 10 - 20% of people, producing habitual subluxation of the patella. Often this does not cause any symptoms or require any treatment. Treatment is difficult; surgery is often unsuccessful and should be considered on an individual basis.

Foot problems

Flat feet

The majority of typically developing children up to the age of five have the appearance of flat feet; as they grow the arch develops. In Down’s syndrome, ligamentous laxity may predispose to flat feet. If pain free, they do not require any treatment. As with any child or adult with a flat foot, insoles are only indicated if flat feet are painful. Very rarely, in severe cases only, will surgery be of benefit

Bunions

In people with Down’s syndrome, commonly the first bone in the foot (first metatarsal) along with the big toe may turn towards the midline. This is called a bunion. This may cause pain and difficulties in fitting of shoes. Treatment should begin with modification of shoe-wear. Early advice from a podiatrist may help. Occasionally if there is disabling pain, surgical intervention may be required.

Club Feet

Congenital talipes varus or club feet is more commonly present in children with Down’s syndrome. This responds very well to the Ponseti technique used to treat club feet in the general population.

Bone Health and osteoporosis

Bone health in people with Down's syndrome can be affected from an early age (e.g. 7-10 years). As in the general population, bone health can be affected by obesity, decreased activity levels and limited exposure to sunshine. In addition to these factors people with Down's syndrome are more likely to suffer from certain health conditions (e.g. autoimmune disorders, coeliac disease, diabetes and hypothyroidism and medications to treat epilepsy) which may cause poor bone health.

The mainstay of improving bone health is to ensure taking optimum levels of calcium and vitamin D and maintaining adequate levels of exercise i.e. at least 1 hour of moderate exercise a day in children and 2.5 hours of moderate exercise per week in adults. Adults should be offered screening for osteoporosis and managed appropriately.

Annual health checks for people with Down's syndrome (aged 14 years plus)

In the past people with learning disabilities have not had equal access to healthcare compared to the general population. This, amongst other reasons, has given rise to poorer mental and physical health and a lower life expectancy for people with learning disabilities. Free annual health checks for adults with learning disabilities, with their GP, were introduced in 2008 as a way to improve people's quality of life.

The annual health check for people with learning disabilities is a Directed Enhanced Service (DES). This is a special service or activity provided by GP practices that has been negotiated nationally. Practices can choose whether or not to provide this service. The Learning Disability DES was introduced to improve healthcare and provide annual health checks for adults on the local authority learning disability register. To participate in this DES, staff from the GP practice need to attend a multi-professional education session run by their local Trust. The GP practice is then paid a sum of money for every annual health check undertaken.

Who can have one?

Annual health checks have been extended to include anyone with learning disabilities aged 14 years or above. So anyone with Down's syndrome aged 14 years or over can have an annual health check.

The benefits of annual health checks

- additional support to get the right healthcare
- increased chance of detecting unmet, unrecognised and potentially treatable health conditions
- action can be taken to address these health needs.

How to get an annual health check

- The GP may get in touch with the person with Down's syndrome to offer an annual health check but this doesn't always happen.
- A person with Down's syndrome and/or a supporter can ask their GP for an annual health check. You do not need to be known to social services to ask for an annual health check.

Not all GPs do annual health checks for people with learning disabilities but they should be able to provide details of other GPs in your area who offer this service.

What happens next?

- The GP practice may send out a pre-check questionnaire to be filled out before the annual health check takes place.
- The GP may arrange for the person with Down's syndrome to have a routine blood test a week or so before the annual health check.

Who attends the annual health check?

If the person with Down's syndrome (age 16 years or over) has capacity and gives their consent, a parent or supporter can attend the health check as well.

How long should an annual health check be?

Guidance from the Royal College of GPs suggests half an hour with your GP and half an hour with the Practice nurse.

What areas of health should be looked at as part of the annual health check?

We have produced a check list for GPs which contains information about what should be included as part of a comprehensive and thorough annual health check. This includes a list of checks that everyone with a learning disability should undergo as part of an annual health check and a list of checks specific to people with Down's syndrome. You can find the health check list at the 'annual health checks' section of our website under 'families and carers and 'health and wellbeing'.

What happens after the annual health check?

Your GP should tell you what they and the nurse have found during the annual health check. You should have a chance to ask any questions you have. Your GP may refer you to specialist services for further tests as appropriate. Your GP should use what they have found during your annual health check to produce a health action plan. This should set out the key actions agreed with you and (where applicable) your parent or carer during the annual health check. Your GP has to do this as part of the annual health check service.

Information about health issues for GPs

There is information at our website for GPs about some of the more common health conditions seen in people with Down's syndrome. You will find this information at the 'annual health checks' section of our website under 'families and carers and 'health and wellbeing'.

GPs learning disability register

People with learning disabilities experience poorer health compared to the rest of the population, but some ill health is preventable. Over one million people in the UK have a learning disability but only 200,000 are on their GPs learning disability register.

We know that people with a learning disability often have difficulties accessing health services and face inequalities in the service they receive. The Government is asking parents and supporters to speak to their GP and ensure their sons/daughters or the people whom they support are registered. It is hoped that this drive will ensure better and more person centered health care for people with learning disabilities.

The Learning Disability Register is a record of people with a learning disability who are registered with each GP practice. The Register is sometimes referred to as the Quality Outcomes Framework (QOF) Register.

If you are not sure you are on the Register, you can ask the receptionist at your GP Practice to check for you.

The doctor may have made a note on the record that a person has Down's syndrome but this does not automatically mean they have been put on the Register. When you speak to the GP about being registered, the needs and support of the person in health settings can be discussed. This information can be entered on the person's Summary Care Record (SCR) so that all health professionals at the practice know about their needs and how best to support them.

If the person is over 16 years of age or older, they must give their consent (see section in this resource about the Mental Capacity Act 2005):

- for information about their support needs to be added to their SCR
- to which information can be shared and with whom

It's never too early (or late) to join your GP's Learning Disability Register; you can join at any age. It's a good idea for children with a learning disability to join the learning disability register at an early age. This means adjustments and support can be put in place before they reach adult services.

Reasonable adjustments in health care

You may have heard of the term 'reasonable adjustments' and wondered what it meant. Since the Disability Discrimination Act (1995) and the Equality Act (2010) (this does not apply to Northern Ireland) public services are required by law to make reasonable adjustments to help remove barriers faced by people with disabilities when trying to use a service. The duty under the Equality Act to make reasonable adjustments applies if you are placed at a substantial disadvantage because of your disability compared to people without a disability or who don't have the same disability as you.

So for people with physical disabilities reasonable adjustments may include changes to the environment like ramps for the ease of wheelchair users. For people with learning disabilities 'reasonable adjustments' may include easy read information, longer appointments, clearer signs at the practice, help to make decisions, changes to policies, procedures and staff training.

If a patient with Down's syndrome is NOT on their GP's Learning Disability Register then reasonable adjustments to care for that person cannot be anticipated and made.

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The Down's Syndrome Association provides information and support on all aspects of living with Down's syndrome.

We also work to champion the rights of people with Down's syndrome, by campaigning for change and challenging discrimination.

A wide range of Down's Syndrome Association publications can be downloaded free of charge from our website.

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