



Annual Health Check Information for GPs

Orthopaedic Problems

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Below is Down's syndrome specific information. The information is for GPs and is to be used alongside DSA's Adult Health Book.

Introduction

The musculo-skeletal problems occurring in Down's syndrome arise mainly from the abnormal Collagen type 6 that forms part of the structure of ligaments and other musculo-skeletal tissues. This results in excessive joint laxity and, together with the muscle hypotonia which is also characteristic, has particular effects on the Hip, Knee, feet and spine (Diamond et al 1981, Cristofaro et al 1986).

The Hip

Laxity, subluxation and dislocation

Capsular insufficiency, ligamentous laxity & muscle hypotonia produce hip instability in Down's syndrome with a reported incidence of between 2 and 5% (Bennet et al 1982, Diamond et al 1981 and, Shaw & Beals 1992). Under the age of 2 years, it is thought that hips are often stable but hypermobile, and may then pass through phases of subluxation and dislocation, leading eventually to fixed dislocation and osteo-arthritis.

Dislocations may be acute, or habitual, where they dislocate without trauma and spontaneously reduce. This can be concerning for the carers as it can look quite dramatic, however these are usually not painful.

Recurrent posterior dislocations can affect the growth and shape of the acetabulum, (dysplasia) which then contribute to the dislocation tendency.

In adulthood, hip abnormalities may be found in up to 28% of adults with Down's Syndrome, with instability worsening with time and correlating with walking ability (Hresko 1993). Occasionally instability begins after skeletal maturity. With life expectancy in Down's syndrome increasing, this relatively high incidence of long-term problems is concerning.

Management

While capsular reefing is possible, any such tightening procedure tends to stretch out, and more permanent solutions attempt to stabilise the joint by altering the alignment of the proximal femur or the acetabulum by osteotomies (reconstructive surgery). Once there is a fixed dislocation, or there is evidence of osteo-arthritis, a total hip replacement is usually required.

Reconstructive surgery is major surgery requiring several months of healing and rehabilitation, and the benefits need to be carefully weighed against the risk of complications, which include infection, non-union and redislocation.

For these reasons the management of these patients should be individualised, with functional impairment being the most important indication for surgery.

There are a number of other conditions affecting the hip which occur with increased frequency in children and adolescents with Down's syndrome and which may leave the hip with deformity and pain in adulthood, for example Slipped upper femoral epiphysis (SUFE) occurs with an increased incidence in adolescents with Down's syndrome, believed to be associated with the increased incidence of hypothyroidism and obesity. Perthes disease and avascular necrosis of the hip are also thought to be slightly commoner in children with Down's syndrome, for reasons which are unclear.

As a result of all of these problems affecting the hip, or due to instability arising in adulthood, the hip joint in patients with Down's syndrome may become arthritic and painful and adversely affect function and independence.

Management should follow the same broad principles as in the general population with osteo-arthritis of the hip, including weight reduction if possible, simple analgesia including paracetamol and the use of walking aids where necessary.

Eventually, total hip replacement may be considered. Theoretical concerns regarding Total Hip Replacement in Down's syndrome include a higher than normal potential risk of dislocation (due to capsular laxity and muscle hypotonia), and a higher than normal risk of infection. However a recent article (Zywiell 2013), elegantly summarising the current evidence, suggests that these risks may not necessarily be as high as once thought. In 42 patients from four studies, standard components were used, although constrained liners and extra acetabular screws were often needed to enhance stability of components. Survival rates varied between 81 and 100% at a mean follow up of 105 months. This review suggested that "while THR in patients with Down's syndrome does present some unique challenges, the overall clinical results are good, providing these patients with reliable pain relief and good function", which is encouraging for a population of patients in whom life expectancy is increasing.

Patellofemoral Instability

Patello-femoral instability can develop in the knee, as in the hip, whereby the patella subluxates or dislocates.

This is probably very common, although not necessarily related to walking ability (Diamond et al, Dugdale 1986, Mendez et al 1988).

The main indication for surgical treatment is functional impairment limiting independence but this should be considered very carefully, and treatment should be individualised. Although results are encouraging, only small case series describing a variety of different procedures are reported in the literature (Dugdale 1986, Mendez et al 1988, Bettuzzi et al 2009, Kocon H 2012). There was a suggestion in one study that surgery might be more successful in patients with more severe disability (Bettuzzi et al 2008).

The Foot in Down Syndrome

The main problems encountered in the foot in Down's syndrome are Hallux valgus and flat foot. Clubfoot is thought to occur with an increased incidence in Down's syndrome, and the residual effects of this may be seen in adulthood.

Metatarsus Primus varus and hallux varus

Metatarsus primus varus describes a deformity where the first metatarsal deviates towards the midline. This can produce a hallux valgus or bunion deformity and is often associated with a flat foot. A hallux varus deformity describes the big toe deviating towards the midline. Either of these deformities can cause problems with shoe fitting and pain across the front of the foot, which can often be treated with appropriate footwear. If symptoms require surgical correction then this will usually involve resection of the bunion if present, with first metatarsal osteotomy to correct the metatarsus primus varus. There are currently no studies documenting outcome or recurrence rates in patients with Down's syndrome.

Flat Feet

Flat feet are common in children and adults with Down's syndrome, with increased tissue laxity predisposing. Often they may not cause a problem, but if arch pain is a problem then insoles may help. Surgery to relieve pain in flat foot is theoretically possible, and may be an option in patients with severe pain that is unresponsive to insoles, however as with all surgery in Down's syndrome there is a significant risk of complications, particularly infection.

Deformity/Problems Secondary to Clubfoot

In newborns, the Ponseti technique is usually very successful in treating the vast majority of babies with clubfeet nowadays, so that anything but minor surgery is rarely needed. However, adults with residual deformity following clubfoot surgery in the past are still being seen, and these are often also stiff feet which are frequently painful.

Sometimes if there is a clear deformity, correction with osteotomies is possible, otherwise, for pain or deformity affecting the subtalar joint, a triple fusion or subtalar fusion may be advocated. This can convert a stiff, deformed and painful foot into a stiff, plantigrade and painfree foot and is often a good treatment option in these cases.

Associated Issues

Some general problems encountered in Down's syndrome may predispose to, or exacerbate orthopaedic conditions and make treatment more difficult.

By increasing the load passing through weightbearing joints, obesity exacerbates pain in joints damaged by a variety of disorders. It also makes surgery technically more difficult, and can prejudice the outcome of Total hip replacement.

Several studies have suggested that low bone mineral density may be relatively common in Down's syndrome (Wu J 2013, McKelvey et al 2013) predisposing to fractures, and osteopenic bone frequently makes fixation with orthopaedic implants very challenging.

Finally, the increased rate of infection in individuals with Down's syndrome individuals means that any surgical procedure needs to be performed with caution and with appropriate antibiotic prophylaxis.

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The Down's Syndrome Association (DSA) is the only organisation in England, Wales and Northern Ireland which supports people with Down's syndrome at every stage of life.

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