Down Syndrome in Children: The Role of the Orthopaedic Surgeon

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Abstract

Down syndrome, the result of trisomy of chromosome 21, is one of the most common chromosomal abnormalities. Patients have a characteristic facial appearance, variable levels of intelligence and self-care skills, and a variety of associated medical conditions. Orthopaedic manifestations occur frequently; most are related to hypotonia, joint hypermobility, and ligamentous laxity. Atlanto-occipital and atlantoaxial hypermobility, as well as bony anomalies of the cervical spine, can produce atlanto-occipital and cervical instability. Methods of screening for this instability, particularly with regard to participation in sports, are a subject of controversy. Scoliosis, hip instability, slipped capital femoral epiphysis, patellar instability, and foot deformities are other musculoskeletal conditions found in patients with Down syndrome that can be challenging for the orthopaedic surgeon to treat.

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John Langdon Down reported the syndrome that bears his name in 1866. Down syndrome is considered to be the most common chromosomal abnormality in humans. Manifestations include several characteristic phenotypic features and orthopaedic anomalies primarily related to ligamentous laxity and joint hypermobility.

Epidemiology, Genetics, and Phenotypic Features

Down syndrome is a common pattern of malformation, occurring in 1 in 660 live births. Risk increases with maternal age, from 1 in 1,500 for mothers age 15 to 29 years to 1 in 50 for mothers older than age 45 years.[1] Although the number of Down syndrome fetuses conceived is thought to have increased as the average maternal age has increased, the number of terminated pregnancies with Down syndrome also has increased. This has resulted in an overall decrease in the prevalence of Down syndrome births.[2]

Diagnosis of Down syndrome is made by chromosome analysis, either prenatally or after birth. Currently, screening begins with a blood test that is offered to all pregnant women, followed by cytogenic diagnosis with chorionic villus sampling or amniocentesis, if needed. After birth, a karyotype can be performed with either a blood or tissue sample. Complete trisomy 21 occurs in 95% of patients, translocation of a portion of chromosome 21 is found in 3%, and mosaicism of normal and trisomy 21 cells is found in 2%. The general likelihood of recurrence in parents is 1% for complete trisomy and higher for translocation.[1]

Patients with Down syndrome exhibit a variable phenotype, including some degree of mental impairment and hand abnormalities, characteristic facial features, generalized ligamentous laxity, and hypotonia (Table 1).

Associated Medical Conditions

A variety of medical conditions is found in patients with Down syndrome (Table 2). Screening for these conditions, managing associated medical problems, and educating families can significantly improve their level of function.[2]

Patients with Down syndrome have variable degrees of developmental delay. The IQ range is approximately 25 to 50. However, these patients often perform better socially than would be expected for their mental age.[1] They meet motor milestones later than do children without Down syndrome; for example, children with Down syndrome often have poor coordination and on average walk a year later than do children without Down syndrome. Awareness of these delays can simplify the evaluation of musculoskeletal problems.

Early onset Alzheimer's disease affects patients with Down syndrome; this dementia can complicate evaluation and care of musculoskeletal problems in adulthood. Symptom onset of dementia occurs at roughly 40 years of age, but symptoms vary. They can include seizures, changes in personality, focal neurologic signs, and changes in speech.[3] Several risk factors have been investigated, including increasing age (>40 years), female sex (inconclusive evidence), preexisting level of mental retardation (inconclusive evidence), genotype for apolipoprotein E, and decreased level of activity.[4]

Life Expectancy

Although patients with Down syndrome once had a short lifespan, life expectancy has increased as treatment of associated problems (particularly cardiac anomalies) has improved. The overall median age of death in patients with Down syndrome in the United States in 1983 was 25 years; this had increased to 49 years by 1997.[3] In patients with congenital heart defects, survival to 1 year is 76%; this decreases to 50% by age 30 years. In patients without congenital heart defects, survival to 1 year is 90%, dropping to only 79% by age 30 years.[1]

Table 1

Common Phenotypic Features in Down Syndrome[1-3]

Facial features
Flat nasal bridge
Epicanthal folds
Upward-slanting palpebral fissures
Open mouth
Hand abnormalities
Small finger hypoplasia
Small finger clinodactyly
Single, deep palmar crease (simian crease)
Characteristic pelvis with lateral flare of iliac wings
Joint hypermobility
Ligamentous laxity
Hypotonia
Short stature
Mental impairment

Table 2

Medical Conditions and Frequencies in Patients With Down Syndrome[1-3]

Condition (Frequency [%])
Cardiac abnormalities
Congenital heart disease (50)
Atrioventricular septal defect (45)
Ventricular septal defect (35)
Patent ductus arteriosis (7)
Tetralogy of Fallot (4)
Leukemia
Leukemia (1)
Acute lymphoblastic leukemia (0.33)
Acute myeloid leukemia (0.33)
Otolaryngologic abnormalities
Hearing loss (75)
Otitis media (50 to 75)
Obstructive sleep apnea (50 to 75)
Ophthalmologic disorders
Refractive errors (35 to 75)
Strabismus (27 to 57)
Congenital cataracts (NA)
Gastrointestinal disorders
Gastrointestinal atresias (12)
Celiac disease (7)
Hirschsprung disease (1)
Skin disorders (87)
Neurologic and psychiatric disorders
Mental impairment (NA)
Seizures (8)
Alzheimer's disease (in adulthood) (75)
Disruptive behavior disorders (17)
Depression (in adulthood) (6)
Endocrine disorders
Hypothyroidism (15)
Diabetes mellitus (1)

NA = not available

Musculoskeletal Conditions

Joint Hypermobility and Ligamentous Laxity

A number of musculoskeletal abnormalities are seen in patients with Down syndrome. Many of these are related to generalized ligamentous laxity and joint hypermobility. Cervical spine instability and atlantooccipital instability can have the most severe consequences. Other manifestations that likely result from laxity or hypermobility include hip instability, patellar instability, and foot deformities. Although the natural history for most of these disorders is not fully defined, they may cause pain and disability in adulthood when untreated.

Arthropathy of Down Syndrome

An arthropathy similar to juvenile rheumatoid arthritis, known as arthropathy of Down syndrome, can

develop in patients with Down syndrome. This rare condition, estimated to occur in 1.2% of patients,[5] is marked by joint subluxations, has a progressive course, and is polyarticular in nature. Affected joints can include the cervical spine, metacarpophalangeal joints, patellofemoral joints, ankles,and hips. Diagnosis is frequently delayed because the symptoms of pain and changes in activity can be confused with behavior problems.[5] Coordinated care with rheumatologists is important for medical management; special attention should be paid to the cervical spine for evidence of subluxation.

Occipitocervical and Cervical Spine Instability

Cervical spine instability, primarily of the upper cervical levels, is a concern in children with Down syndrome. This instability can occur not only at the atlantoaxial joint (C1-C2) but also at the occiput-C1 junction. Instability of the upper cervical spine was first described nearly 50 years ago; it is an issue of primary concern in children with Down syndrome because of the neurologic sequelae that can result.

Atlantoaxial Instability

Stability at the atlantoaxial joint is provided by bony and ligamentous structures. Bony integrity is supplied by the odontoid process and the anterior arch of C1. Another primary stabilizer is the transverse ligament that holds the odontoid against the anterior arch of C1. The paired alar ligaments that connect the odontoid to the occipital condyles, together with the apical ligament, which runs from the odontoid to the foramen magnum, act as secondary stabilizers and checkrein ligaments for the C1-C2 joint during rotation.[6]

In children with Down syndrome, hypermobility and frank instability of the atlantoaxial joint can result from decreased support from the ligamentous structures secondary to ligamentous laxity. Atlantoaxial instability is attributed primarily to laxity of the transverse ligament and C1-C2 joint capsules. Atlantoaxial instability in patients with Down syndrome has been variably defined radiographically on lateral, neutral, and flexion-extension plain radiographs by a minimum atlanto-dens interval (ADI) of 4 to 5 mm and has been noted in 10% to 30% of patients.[7-12] These studies include both children and adults. In infants and toddlers, portions of C1 and C2 are not yet ossified; thus, meaningful plain radiographic measurements are not possible. The space available for the cord has been suggested as a better radiographic measurement for instability at C1-C2, but ultimately, supervised flexion-extension cervical magnetic resonance imaging (MRI) allows for more direct assessment of cord compression in pediatric patients who have a suggestion of instability on plain radiographs or clinical symptoms of instability.[7,9,10,12,13]

Most children with Down syndrome and instability at C1-C2 are asymptomatic.[8,10,11,14] Long-term studies demonstrate that only minor changes in the ADI develop over time in most patients with a C1-C2 abnormality.[10] In their 3-year study of patients with Down syndrome, Ferguson et al[7] found that an increase in ADI developed in none of the patients.

Symptomatic atlantoaxial instability is estimated to occur in 1% to 2% of patients with Down syndrome.[8,14,15] Symptomatic patients often present between ages 5 and 15 years and may report neck pain, sensory deficits, or difficulty with bladder control. More subtle symptoms include a decreased tolerance for activities, changes in gait, or loss of ambulatory skills. Specific questions about these historic details should be directed to the caregivers. On physical examination, a variety of signs can be present, including wide-based gait, neck posturing, decreased neck range of motion, hyperreflexia, abnormal Babinski reflex, ankle clonus, or weakness. We have found that most children with Down syndrome are very cooperative, pleasant, and easy to examine. However, if the child is upset, a helpful technique (besides patience on the part of the examiner) is to have the child sit on a parent's lap during the examination.

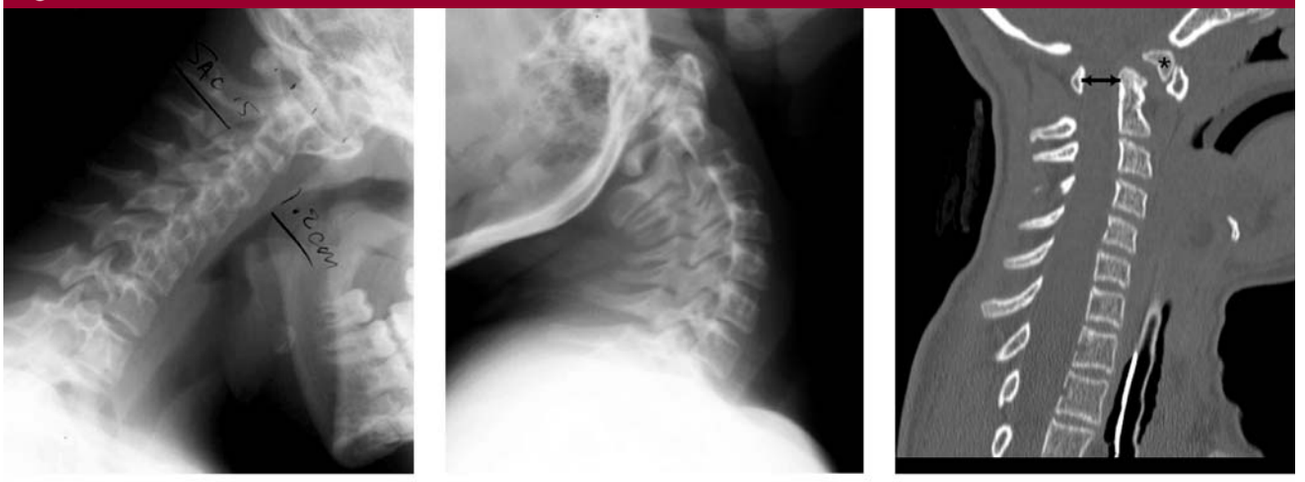
Less commonly, instability of the atlantoaxial joint can result from loss of bony integrity of the dens, as with os odontoideum (Figure 1). Children with Down syndrome have a notably greater number of osseous anomalies of the upper cervical spine than do age- and sex-matched normal, healthy children.[8,9] Os odontoideum, persistent dentocentral synchondrosis of C2, spina bifida occulta of C1, and ossiculum terminale are the most frequently noted anomalies.[9] These anomalies can make interpretation of radiographs more difficult. Furthermore, children with Down syndrome and asymptomatic atlantoaxial instability are more likely to have additional upper cervical spine anomalies than are Down syndrome children without C1-C2 instability.[8]

Atlanto-occipital Instability

Stability at the atlanto-occipital junction is provided by the cup-shaped joints between the occipital condyles and the superior articular facets of C1, as well as their capsular ligaments. The tectorial membrane, a continuation of the posterior longitudinal ligament, also provides considerable support. White and Panjabi[6] described normal motion in the atlanto-occipital joint as 13° of total flexion-extension, no axial rotation, and 8° of lateral bending. Flexion is limited by the contact of the anterior sphenoid on the dens. Extension is limited by the posterior aspect of the occiput contacting the atlas and by the tectorial membrane.[6]

Instability at the occiput-C1 level in patients with Down syndrome likely results from laxity of the atlanto-occipital joint capsule, the

Figure 1



A B C

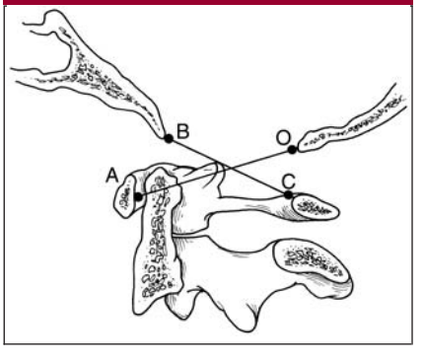
Flexion **(A)** and extension **(B)** lateral radiographs of the cervical spine demonstrating os odontoideum and atlantoaxial instability in a symptomatic 12-year-old patient with Down syndrome. **C,** Sagittal computed tomography scan in neutral. The os odontoideum is labeled with an asterisk; the space available for the cord is marked with arrows. This patient underwent the maximum reduction that could be achieved with positioning in the operating room and occiput-to-C2 fusion, using iliac crest bone grafting with sublaminar wires and halo vest placement. Although reduction was incomplete, it was sufficient; solid arthrodesis was achieved with resolution of neck pain.

tectorial membrane, and the anterior and posterior atlanto-occipital membranes.[16] In skeletally mature patients, the assessment of occiput-C1 instability can be made from lateral cervical spine radiographs using several techniques, including the Power ratio (Figure 2) and the Weisel-Rothman technique. The Power ratio offers the advantage of being unaffected by patient size or magnification. In addition, the axis is not involved in the measurement, which eliminates the possibility of any C1-C2 instability confusing the assessment.[17] A Power ratio >1.0 is indicative of anterior occiput-C1 dislocation. The Weisel-Rothman method measures the translational motion between the occiput and C1 based on the difference between two points on both the flexion and extension films.[18] The distance should not change by more than 1 mm. Karol et al[19] examined these measurement techniques for occiput-C1 instability specifically in children with Down syndrome and found that they were not easily reproducible. They recommended confirmation of instability with MRI in children with Down syndrome when plain radiographs suggest abnormal motion.[19]

Although patients with symptomatic occiput-C1 instability present with neurologic signs and symptoms similar to those of C1-C2 instability, the true incidence of instability at occiput-C1 in Down syndrome is unknown. A study of radiographs of both adults and children with Down syndrome found hypermobility (>1 mm of motion) at the occiput-C1 level by the Weisel-Rothman method in 79% of patients with Down syndrome compared with 21% of control subjects.[20] Thirty-seven of the 38 patients with Down syndrome in the study were asymptomatic. Radiographic views of atlanto-occipital instability in a child with Down syndrome are shown in Figure 3.

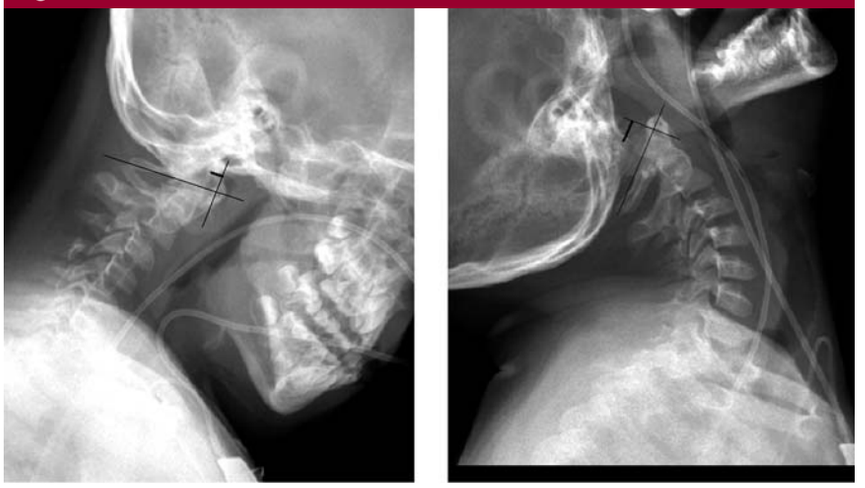
Atlantoaxial and atlanto-occipital instability may coexist in patients with Down syndrome. The effects of concurrent occiput-C1 and C1-C2 instability may be additive, leading to overall symptomatic instability even though motion at each individual segment is small. Thus, when evaluating cervical spine radiographs in

Figure 2



The Power ratio is calculated by drawing a line from the basion (B) to the posterior arch of the atlas (C), and a second line from the opisthion (O) to the anterior arch of the atlas (A). The length of line BC is divided by the length of OA. (Reproduced from Copley LA, Dormans JP: Cervical spine disorders in infants and children. *J Am Acad Orthop Surg* 1998;6:204-214.)

Figure 3



A B

The Weisel-Rothman technique. Flexion **(A)** and extension **(B)** lateral radiographs of the cervical spine demonstrating significant translation of the occiput on C1 in a symptomatic 8-year-old patient with Down syndrome. The difference in length (in **A** versus **B**) between the heavy lines represents the translation.

patients with Down syndrome, it is important to consider instability at both the occiput-C1 and C1-C2 levels. In multilevel instability, absolute displacement values are not as important as is overall space available for the spinal cord.[16] A supervised flexion-extension cervical MRI scan allows for assessment of cord compression because of hypermobility at multiple levels in pediatric patients with myelopathic symptoms.[13]

Screening for Upper Cervical Spine Instability

Radiographic screening of patients with Down syndrome for upper cervical spine instability is controversial because of undefined efficacy and cost effectiveness. Currently, screening lateral flexion and extension radiographs are required by Special Olympics before patients with Down syndrome can participate in sports viewed as high risk, such as diving and gymnastics.[21] However, in 1995, the Committee on Sports Medicine and Fitness of the American Academy of Pediatrics published a position paper questioning the value of lateral plain radiographs as a screening test in detecting patients at risk for spinal cord injury that occurs during sports participation.[22] The Committee members contended that symptomatic instability in patients with Down syndrome is neither common nor severe enough to justify the work and cost of detection in the asymptomatic patient. The members argued that lateral radiographs have low reproducibility, leading to decreased sensitivity and specificity in detecting instability. They also asserted that asymptomatic instability has not been proved to be a risk factor for symptomatic instability. Finally, consistently effective, low-risk treatment is not available to prevent progression from asymptomatic to symptomatic instability.[22] After review of the literature, the Committee members determined that screening radiographs for upper cervical spine instability do not meet the criteria for an effective screening test and are not warranted in asymptomatic individuals.

Other experts disagree on the value of screening radiographic evaluations. Pueschel[23] favors screening radiographs of the cervical spine in patients with Down syndrome. He cites several reasons for screening, including the increased frequency of upper cervical spine instability in patients with Down syndrome and the risk of progression to neurologic symptoms, especially with an injury to the head or neck. In an editorial, Cohen[14] presented a counterargument, suggesting that evidence for the usefulness of screening remained inconclusive and that further study and a consensus meeting were needed to help clear the issues and provide the best care to patients with Down syndrome.

Another situation in which patients with Down syndrome require cervical spine evaluation is before receiving anesthesia for surgical procedures. Neither evidence-based guidelines nor a consensus exists in the anesthesia literature regarding the inclusion of cervical spine radiographs in the preoperative assessment of patients with Down syndrome. A search of the English-language literature and of the American Society of Anesthesiologists Closed Claims Database by Hata and Todd[24] revealed eight possible cases of cervical spine injury associated with anesthesia in patients with Down syndrome. They recommended thorough preoperative assessment by the anesthesiologist, who should look specifically for signs and symptoms of cervical cord compression. When the assessment is suspicious for neurologic abnormality or when the patient has had abnormal flexion-extension cervical radiographs in the past, the preoperative assessment should be followed by referral to a pediatric orthopaedic or neurology specialist.[24] The authors suggested that cervical spine radiographic evaluation be done when manipulation of the head and neck is required during the surgical procedure. In the case of asymptomatic instability or hypermobility identified before an elective surgical procedure, care is individualized and

coordinated between the orthopaedic surgeon, anesthesiologist, and operating surgeon.

Management of Upper Cervical Spine Instability

In our experience, most children with Down syndrome present initially to the orthopaedic surgeon between ages 5 and 10 years. The type of imaging ordered at presentation depends on the age of the patient and the information obtained from a careful history and physical examination. However, our practice is to obtain lateral cervical spine radiographs taken in neutral, flexion, and extension on most patients at initial presentation. These radiographs should be evaluated for atlanto-occipital and atlantoaxial as well as lower cervical spine instability. When no instability exists and the rest of the evaluation is normal, we educate the family on activities and on symptom surveillance. We instruct parents to watch for and to return if symptoms such as neck pain, torticollis, changes in bowel or bladder control, and gait abnormalities occur. In asymptomatic patients, we repeat the clinical evaluation every 2 to 3 years, and we repeat lateral cervical spine radiographs in neutral, flexion, and extension when symptoms or signs of instability are found in the history or on physical examination.

When hypermobility or instability is identified, patients with Down syndrome who have asymptomatic atlantoaxial instability should be followed with symptom surveillance, neurologic examination, and further radiographic evaluation.[7,11] When abnormalities are detected on plain radiographs, we recommend the use of more advanced imaging, such as flexion-extension MRI scans. We emphasize educating the parents in the avoidance of activities that put the child at increased risk for cervical spine injuries, such as gymnastics, football, wrestling, tumbling, and other vigorous sports activities (eg, downhill skiing). When asymptomatic instability exists, we perform an annual neurologic examination and radiologic evaluation of the patient, with lateral cervical spine radiographs. Asymptomatic patients with grossly increased ADI are followed more frequently, with exact treatment decisions made on an individual basis.

Surgical Treatment of Upper Cervical Spine Instability

Patients with Down syndrome who have symptomatic upper cervical spine instability should be strongly considered as candidates for a stabilization procedure.[7,9,11] Based on their study of adult patients with Down syndrome, Ferguson et al[7] recommended fusion for patients with progressive or acute onset of neurologic symptoms, ≤14 mm of space available for the spinal cord on plain radiographs, and MRI or computed tomography (CT) evidence of cord compression. Preoperative assessment of all potential levels of instability should be performed with advanced imaging, and fusion may include the occiput-C1 level, C1-C2 level, or both, when warranted. In our experience, factors that would tip the balance in favor of surgery include neck pain, deterioration of motor activity, loss of or failure to meet motor development milestones, or neurologic findings related to brainstem or upper cervical spinal cord compression.

In the absence of neurologic symptoms, surgical treatment of upper cervical spine instability is rarely indicated. Arthrodesis of the upper cervical spine in these patients can be very difficult to achieve and has a high complication rate. Noted complications include infection and wound dehiscence, development of junctional instability, nonunion, loss of reduction, resorption of bone graft, and neurologic deterioration.[9,12,25] In all cases, emphasis on education of the family and patient is integral to the decision for surgery.

Regarding surgery, most authors suggest autologous bone graft and instrumentation[9,10,26,27] and prolonged halo-ring and vest immobilization to increase the likelihood of successful bony union. The type of instrumentation should be selected carefully. When cervical stenosis is noted on preoperative studies or is evident in the patient's signs and symptoms, there is a potential risk of neurologic injury with the passage of sublaminar wires. Careful positioning and radiographic confirmation at the beginning of the surgical procedure, to reduce any mobile subluxation and maximize the space available for the cord, can minimize this risk. In the case of irreducible subluxation, surgical decompression may be required in combination with fusion and instrumentation.[9] With the advent of lateral mass plates and advanced internal fixation techniques of the upper cervical spine, additional options are now available for stabilization. Doyle et al[12] recommended that patients with Down syndrome be followed indefinitely after upper cervical fusion; they reported late onset of upper motor neuron signs in the face of solid fusion as well as late adjacent motion segment instability in the subaxial spine.

Scoliosis

Diamond et al[28] originally reported scoliosis in 50% of 107 institutionalized patients with Down syndrome; however, only 15% of the population in the study had a curve >7° by radiographic examination. A more recent study yielded an incidence of scoliosis of 7% of patients by physical examination; this study included patients living at home as well as in institutions.[15]

Despite the reported increased incidence of scoliosis in Down syndrome, little has been published regarding its natural history, treatment, or outcomes. In their report on results of surgical treatment of progressive scoliosis in Down syndrome, Lerman et al[29] noted that successful fusion occurred in 6 of 7

patients who underwent spinal arthrodesis for progressive scoliosis; however, complications occurred in 5 of the 7 patients. Notable problems included pseudarthrosis, instrumentation failure, postoperative wound infections, and delayed wound healing.[29] No neurologic problems were reported. This led the authors to conclude that the goals of arthrodesis and cessation of curve progression could be attained in patients with Down syndrome, although complications are frequent.

With the paucity of information on bracing and outcomes in scoliosis in Down syndrome, our treatment recommendations are conservative and similar to those for idiopathic scoliosis. Examination for scoliosis is incorporated into the physical examination in grade school–aged children with Down syndrome, and it should be part of the regular examination in the primary care physician's office. We recommend bracing in skeletally immature patients with scoliotic curves of 25° to 30° and continuing close follow-up. Compliance with brace wear can be difficult or frankly unsuccessful with some patients. We reserve surgery for progressive scoliosis and Cobb angles >50° to 60°, following open discussion with families and consideration of the patient's other health problems.

Hip Instability

Hip instability has a 2% to 5% incidence in Down syndrome[28,30,31] and is considered to be multifactorial. The bony structure of the hip joint is altered in Down syndrome, characterized by a deeper and more horizontal acetabulum as well as decreased acetabular anteversion and increased femoral anteversion.[31] These bony changes seemingly increase hip stability; however, capsular laxity, hypermobility, and increased external rotation appear to play a large role in hip joint instability, including dislocation. Dysplastic changes occur over time, including posterior acetabular deficiency, which can be identified with the aid of three-dimensional CT.[32]

Unlike patients with developmental dysplasia of the hip, patients with Down syndrome who have dislocation typically have stable hips before walking age; the subluxation and dislocation develop later. Bennet et al[30] divided the natural history of hip dislocation in Down syndrome into four distinct phases: initial, dislocation, subluxation, and fixed. The initial phase includes patients with stable but hypermobile hips before the onset of walking, or at approximately age 2 years. The dislocation phase is subdivided into acute and habitual dislocation groups. Children in the acute dislocation group often present at age 7 or 8 years with sudden onset of limp or refusal to walk. Their hips are dislocated but can be reduced closed with the patient under anesthesia. Children in the habitual dislocation group have hips that dislocate without trauma and spontaneously reduce, starting in early childhood, sometime after walking age. Some of these hips may stabilize without treatment during this phase. In the subluxation phase, acetabular dysplasia develops. In the fixed dislocation phase, which occurs in early adulthood, patients remain ambulatory with a limp. In a child with Down syndrome, episodic hip dislocation can result in permanent damage to the femoral head (Figure 4).

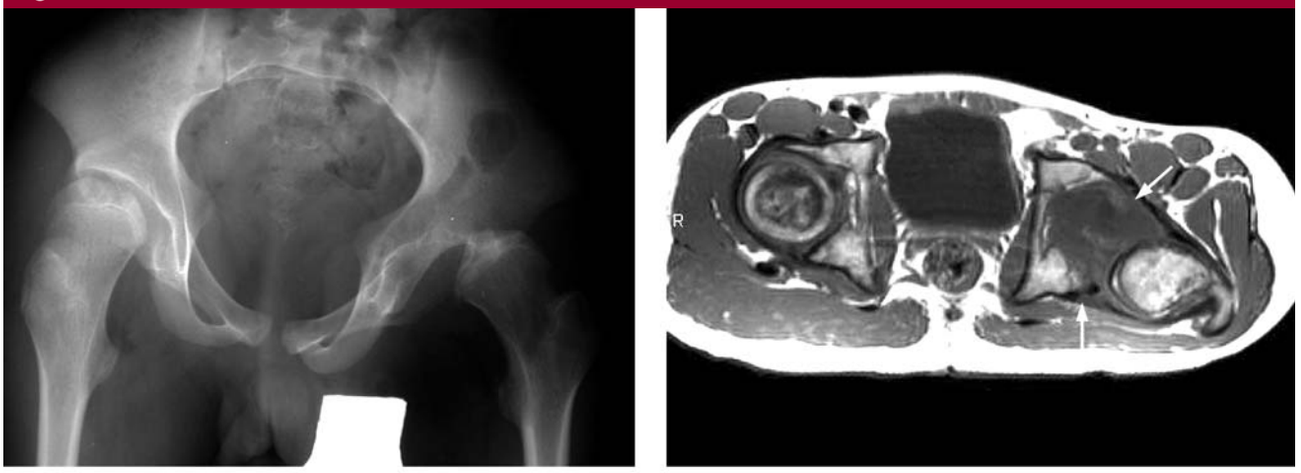
As the life expectancy of patients with Down syndrome increases, the incidence of painful arthritis in adulthood is also rising. Hresko et al[33] reported hip abnormalities in 28% of adult patients with Down syndrome. These patients were much more likely to use a wheelchair or to ambulate in the household only. Although they found 6% of patients with osteoarthritis of the hip, 18% of patients had dysplasia, subluxation, or dislocation.[33]

Treatment of the subluxating or dislocating growing hip in Down syndrome is focused on minimizing late degeneration. Treatment is based on the pattern and chronicity of the dislocation, the presence of associated pain and disability, and patient factors such as age and remaining growth. An initial trial of nonsurgical management may be successful. Greene[34] described two young children, one with acute and the other with habitual dislocation of the hip, who were both treated with closed reduction and prolonged immobilization for 10 to 12 months. At 2.5- and 10-year follow-ups, there was no pain, limp, or recurrent dislocation in either patient.

When immobilization is unsuccessful, surgical intervention can be undertaken to address the redundant capsule with plication or repair.[30] Additionally, in patients with a normal-appearing acetabulum, femoral varus derotation osteotomy has been advocated.[30,35] When the acetabulum is deficient, an acetabular osteotomy that addresses the acetabular insufficiency should be performed. Several authors[30,31] have described use of the Salter innominate osteotomy and caution against a Chiari innominate osteotomy, which has a high associated rate of redislocation. A reverse Pemberton osteotomy, which provides augmentation for the posterior acetabulum, has been suggested for those patients with posterior acetabular deficiency.[32] Katz et al[36] reported good results, clinically stable hips, and symptomatic improvement in skeletally mature patients with Down syndrome and hip dysplasia treated with the modified Bernese periacetabular osteotomy.

Complications of surgical management of hip dislocation in Down syndrome are frequent. Bennet et al[30] noted complications in 9 of 14 patients treated surgically in their study. The complications included wound infection, redislocation, proximal femur fracture following an osteotomy, and osteomyelitis. Katz et al[36] reported postoperative subluxation, labral tear, ischial nonunion,

Figure 4



A B

A, Anteroposterior pelvic radiograph of a 10-year-old boy with Down syndrome who had recurrent episodes of painful left hip dislocation and ultimate deformity of the femoral head. **B,** T1-weighted axial magnetic resonance image of the hips of the same patient. Extreme capsular laxity is evident in the left hip (arrows). The patient's pain resolved; he currently participates in most activities without discomfort or any new episodes of pain. His parents did not elect surgical treatment of his hip but understand that hip pain and degeneration of the hip may develop in the future.

inferior pubic ramus stress fracture, and wound hematoma in their eight adult patients treated with periacetabular osteotomy.

With long-term studies showing pain and decreased ambulation in adult Down syndrome patients with hip subluxation, diagnosis and treatment in childhood are important. Because hip instability in Down syndrome patients is a multiphase problem that can present at different ages as well as with variable levels of instability and acetabular deficiency, we individualize our treatment recommendations. In the absence of radiographic changes in a child whose hip dislocates, we recommend immobilization in a hip spica cast or hip abduction orthosis. When radiographic changes are evident in the acetabulum or when signs of subluxation of the hip are present, we recommend surgical intervention. Osteotomy of the acetabulum or the proximal femur may be needed, accompanied by capsular plication to address the underlying laxity. However, treatment must be individualized (Figure 4).

Slipped Capital Femoral Epiphysis

The incidence of slipped capital femoral epiphysis (SCFE) in patients with Down syndrome is estimated at 1.3%,[31] and patients frequently have poor results. One study identified two cases of SCFE in 161 patients, in both of whom osteonecrosis developed.[28] Dietz et al[37] found that patients with Down syndrome who had SCFE were more likely to present with unstable slips and high-grade slips (>50% displacement). They noted a high rate of osteonecrosis in both unstable and stable SCFE. In another study of SCFE in eight patients with Down syndrome, Bosch et al[38] noted that progression occurred more frequently (6/11 hips), osteonecrosis was common (2/8 patients), and the time to physeal fusion was longer (average, 3 years). Most studies have demonstrated very high complication rates in this patient group, with revision rates from 64% to 80%.[28,37,38]

The relationship between endocrine abnormalities and SCFE has been suggested as a possible explanation for SCFE in Down syndrome, but this has not been fully investigated. Bosch et al[38] reported hypothyroidism in six of eight patients. Although Merrick et al[15] reported that 20% of patients had hypothyroidism in their study of musculoskeletal problems in Down syndrome, there were no cases of SCFE in 475 community and institutionalized patients.

Communication difficulties may complicate the diagnosis of SCFE in patients with Down syndrome. Hip, thigh, and knee discomfort and refusal to bear weight are common presentations. Once identified, SCFE in children with Down syndrome should be treated with in situ screw fixation. In addition, thyroid function tests should be obtained to investigate the possibility of an underlying endocrine abnormality.

Patellar Instability

Patellofemoral instability is even more common than hip instability in Down syndrome and is estimated to occur in 10% to 20% of patients.[15,39] As with many other

musculoskeletal manifestations of Down syndrome, ligamentous laxity and hypotonia are thought to be the primary causes. Merrick et al[15] found a significant ($P < 0.01$) relationship between joint laxity and patellofemoral instability. Dugdale and Renshaw[40] divided patellofemoral instability in patients with Down syndrome into different stages by the degree of laxity—ability to subluxate, to dislocate, to dislocate but reduce, or inability to reduce. Frank patellar dislocation is found in 2% to 8% of patients with Down syndrome.[15,28,39,40]

Even in patients with an unstable patella or dislocated patellae, most continue walking.[40] Although most patients with patellar instability have no pain with full range of motion and no limitation in walking, deformity and arthritis that can result in disability develop in some patients' knees. Because patients with Down syndrome are living longer, the disability from instability of the patellofemoral joint may increase.

Treatment of patellofemoral instability begins with nonsurgical management. Mendez et al[39] found that nonsurgical treatment, including physical therapy and orthotics, was effective in maintaining or improving ambulation in most patients who were ambulatory before treatment. However, nonsurgical treatment did not improve ambulatory status in those with fair or poor ambulation before treatment. In knees without significant deformity, the authors recommended surgical treatment, with attention paid to soft-tissue balancing and repositioning of the insertion of the patellar tendon. They did caution that degenerative arthritis eventually developed in patients with underlying deformities, despite the method of treatment of patellofemoral instability. Dugdale and Renshaw[40] noted redislocation in three of five patients treated by a variety of surgical procedures. No published studies with large numbers of patients or long-term follow-up of surgical treatment of patellofemoral instability in Down syndrome are currently available.

We generally recommend nonsurgical treatment of patellar instability in patients with Down syndrome. This includes a knee sleeve, activity modifications, and, when pes planus is a component of the problem, arch supports. We do not recommend any specific restrictions for patients who are asymptomatic. When symptoms are persistent, we recommend surgery, including a lateral release, medial reefing, and, in skeletally mature patients, bony realignment of the patellar tendon.

Foot Disorders

Although foot disorders affect many patients with Down syndrome and with time can affect ambulation, very little has been published about the natural history and treatment. Pes planus is found in 2% to 6% of patients with Down syndrome[15,28] and is thought to develop as a result of ligamentous laxity. In those studies,[15,28] severe flatfoot with bony changes was uncommon; flatfoot was found more frequently in institutionalized patients with Down syndrome than in those living in the community. Moderate or mild flatfoot may respond to orthotic management. Diamond et al[28] suggested that good pain relief can be achieved with surgical intervention in severe, fixed pes planus, but no details were included on complications, patient function, or rate of recurrence.

Disorders of the first ray in Down syndrome include metatarsus primus varus and hallux valgus. These can result in difficulties with comfortable shoe wear and decreased ambulation. In severe hallux valgus with increased intermetatarsal angle, patients may benefit from first ray realignment and exostectomy.[28] The complications and recurrence rates with surgical treatment of these deformities have not been reported.

We recommend nonsurgical treatment for foot deformities in Down syndrome. We recommend shoes with appropriate width in the toe box to minimize problems with the first ray. Heel cord stretches and orthoses with arch supports have been helpful for symptomatic flexible flatfoot. In advanced or symptomatic foot deformities, modified shoes with increased width may help preserve ambulation. As with most other problems of ligamentous laxity in patients with Down syndrome, we reserve surgical intervention for intractable pain or deformity that prevents ambulation.

Summary

Approximately 20% of all patients with Down syndrome have some associated musculoskeletal problem. With advances in management of cardiac anomalies and other medical problems, patients with Down syndrome are now living longer. The need for better understanding of the natural history of the orthopaedic manifestations of Down syndrome is amplified as life expectancy increases. Upper cervical spine instability is potentially the most ominous orthopaedic manifestation of Down syndrome and can cause neurologic symptoms. When neurologic impairment occurs, surgical stabilization of occiput-C1 and C1-C2 instability is recommended. Scoliosis, patellar instability, and foot disorders can cause functional difficulties and may become more troublesome later in life. Hip instability occurs rarely but can lead to pain and disability in adulthood. Nonsurgical management of these problems, at least initially, is often best. If surgery is undertaken, the patient and family should be informed that surgical intervention is associated with higher complication rates, including infection and recurrence of deformity or instability. The role of the orthopaedic surgeon is to understand and anticipate the musculoskeletal manifestations in Down syndrome and to treat those that may cause functional impairment.

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Citation numbers printed in **bold type** indicate references published within the past 5 years.

Evidence-based Medicine: There are no prospective, randomized, level I or II studies referenced. The cited references are level III/IV case-control cohort studies or expert opinion.

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