

Orthopaedic Management in Down Syndrome

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Abstract:

Down syndrome is the most common chromosomal disorder and typically results from a maternal duplication of chromosome 21 yielding trisomy 21. General features include a flat facial profile, short stature, oblique palpebral fissures, epicanthal folds, and associated medical conditions such as congenital heart disease, vision problems, and hearing loss. Most musculoskeletal manifestations of Down syndrome are related to generalized ligamentous laxity, joint hypermobility, and hypotonia which can lead to atlantoaxial instability, atlanto-occipital instability, scoliosis, spondylolisthesis, hip dysplasia/instability, patellar instability, pes planus, and hallux valgus. Importantly, the orthopaedist may also be the first to discover systemic conditions such as hypothyroidism associated with slipped capital femoral epiphysis (SCFE) or leukemia or inflammatory arthritis leading to musculoskeletal pain. The purpose of this review is to summarize what the orthopaedist needs to remember when evaluating and treating their patients with Down Syndrome.

Key Concepts:

- Musculoskeletal manifestations in Down syndrome are related to generalized ligamentous laxity and can be variable in presentation at almost every anatomic level.
- Subsequent management of orthopaedic conditions such as patella instability, pes planus, and hallux valgus are driven by symptoms. A high index of suspicion should be maintained for other associated conditions such as cervical instability, scoliosis, hip instability or SCFE as a subtle presentation is common.
- Surgical management can be complicated by the pathologic laxity present in patients with Down syndrome and a multidisciplinary and multisystem approach should accompany any surgical treatment.
- The orthopaedist may be the first to evaluate a Down patient for systemic disorders such as hypothyroidism, leukemia, and inflammatory arthritis. These comorbidities can have profound negative effects if missed.

Introduction/Background

Down syndrome is the most common chromosomal disorder and is characterized by physical features such as a flat facial profile, short stature, oblique palpebral fissures, and epicanthal folds and associated medical conditions including congenital heart disease, hearing loss, and cataracts.¹ There are a variety of musculoskeletal conditions encountered in children with Down

syndrome thought to be related to the generalized ligamentous laxity, joint hypermobility, and hypotonia.² These musculoskeletal conditions include atlantoaxial instability, atlanto-occipital instability, scoliosis, spondylolisthesis, hip instability, slipped capital femoral epiphysis, patellar instability, pes planus, and hallux valgus.

Pathogenesis

Down syndrome is the most frequent chromosome abnormality occurring in humans. The current prevalence is estimated at 1 in 700 babies born in the United States, with increased prevalence associated with advancing maternal age: 1 in 1550 births in women under 20 years to 1 in 25 in mothers over 45 years.^{1,3} Specialized prenatal testing has resulted in increased awareness of Down syndrome in utero with the birth prevalence impacted by a rise in pregnancy terminations.^{4,5}

The predominant genetic makeup in 95% of Down syndrome patients is three copies of chromosome 21 that occurs secondary to meiotic nondisjunction.¹ A Robertsonian translocation occurs in 4% of cases where the long arm of chromosome 21 fuses to another acrocentric chromosome—most commonly chromosomes 13, 14, or 15. The final 1% of cases are related to genetic mosaicism.¹

The increased ligamentous laxity and hypotonia associated with Down syndrome may be related to an increased quantity of type VI collagen which is in part encoded by genes located on chromosome 21.^{6,7} The COL $\alpha 1$ (VI) and $\alpha 2$ (VI) chains are encoded by genes located on chromosome 21 and have a higher dosage in individuals with Down syndrome.⁶ In addition, there is a single nucleic peptide (SNP) on chromosome 2 that also seems to induce structural and functional changes in COL $\alpha 3$ (VI).⁶ Type VI collagen is crucial for cardiac and skeletal muscle function which is consistent with most of the hypotonia issues seen in Down syndrome.^{6,7}

Screening and Diagnosis

The American College of Obstetricians and Gynecologists recommend all pregnant females be offered screening for Down syndrome early in pregnancy regardless of maternal age or baseline risk.⁸ Screening options include first-trimester screening (nuchal translucency measurement and measurement of maternal serum human chorionic gonadotropin, pregnancy-associated plasma protein, and alpha-fetoprotein levels), second-trimester screening (measurement of human chorionic gonadotropin, alpha-fetoprotein, dimeric inhibin A, and

unconjugated estriol), and cell-free DNA screening.⁸ Cell-free DNA screening is the most sensitive and specific screening test available with a detection rate of 99%.⁸ Chorionic villi sampling and amniocentesis are diagnostic tests used for confirmation following a positive screening test.⁸ If the diagnosis has not occurred in utero, the initial preliminary diagnosis usually can be made postnatally via the classic clinical features of Down syndrome such as a flat facial profile, oblique palpebral fissures, and epicanthal folds.⁹ Confirmatory diagnosis can be performed with the help of a genetics consultation and a formal karyotype.⁹

Natural History of Down Syndrome

Due to medical advances, the life expectancy of patients with Down syndrome increased from 25 years of age in 1983 to 60 years of age in 2020.¹⁰ In particular, significant advancements have been made in the treatment and management of congenital heart disease. Despite these advancements, adults with Down syndrome have a higher risk of death from dementia, pulmonary disease, congenital heart disease, and choking compared with the general population.¹¹ While in the past children and adults with Down syndrome were often institutionalized, today many children and adults live at home with their families into adulthood. Bertoli et al. found that 88% of adults with Down syndrome lived at home with their parents.¹² Many adults with Down syndrome are able to obtain employment in both facility-based and community employment. Bush et al. found that 30.1% of adults with Down syndrome have paid facility-based employment and 15.6% have paid employment in the community.¹³ When adults with Down syndrome are integrated into the community rather than institutionalized, this can indirectly contribute to the increase in their lifespan.¹⁴

Important Social Considerations

Treatment of the child with Down syndrome requires attention and resources to address the complex medical and social dynamics that affect these patients. It is important to consider behavioral problems, cognitive impairment and parental stress, and to have an overall understanding that there are social implications when

caring for the child with Down syndrome. Parents of children with Down syndrome may face significant challenges. It has been shown that parents caring for a child with a developmental disability may be more likely to develop anxiety, low self-esteem, depression, hypertension, and poor neuroendocrine and immune function.^{15,16} The pediatric orthopaedic surgeon should be aware of these complex family and psychosocial dynamics that could affect decision-making and treatment.

Important Medical Considerations

While orthopaedic manifestations of Down syndrome will remain the focus of the orthopaedic exam, it is important for the pediatric orthopaedic surgeon to be aware of the various nonorthopaedic congenital malformations and medical conditions encountered when treating the child with Down syndrome. Phenotypic manifestations of Down syndrome include brachycephaly, brachydactyly, broad hands, epicanthal folds, upward-slanting palpebral fissures, fifth finger clinodactyly, single transverse palmar crease, flat nasal bridge, mental retardation, small mouth, short stature and hallux valgus¹⁷ (Table 1).

Systemic Diseases for the Orthopaedic Surgeon to Consider

There are specific systemic diseases associated with Down syndrome that have significant crossover with the musculoskeletal system and may impact the orthopaedist in their evaluation or management of a patient with Down syndrome (Table 2). Specifically, obesity, low bone mineral density, thyroid disease, leukemia, and arthropathy of Down syndrome can complicate treatment or may present primarily in the orthopaedic clinic.

Obesity

Children with Down syndrome have high rates of obesity, which can affect their overall physical and musculoskeletal function. Approximately 25% of children with Down syndrome and over 50% of adults with Down syndrome are obese.^{9,18} Abnormal leptin levels, lower physical activity levels, lower resting energy expenditure, dietary patterns, and comorbidities such as congenital

**Table 1.
Associated Medical
Conditions in Down syndrome¹⁷**

| |
|--|
| Congenital heart disease |
| Vision problems |
| Cataracts |
| Refractive errors |
| Hearing loss |
| Otitis media |
| Hypodontia and delayed dental eruption |
| Hypothyroidism |
| Seizure disorder |
| Gastrointestinal atresia |
| Leukemia |
| Transient myeloproliferative disorder |
| Iron deficiency |
| Obstructive sleep apnea |
| Periodontal disease |
| Celiac disease |
| Behavioral problems |
| Autism |
| Hirschsprung disease |

heart disorders and thyroid disease have all been attributed.¹⁹ Children with Down syndrome have

shorter limbs than those without the syndrome thus altering body mass relative to height.²⁰ Differing growth rates and body weight distribution has led to the development of specific growth charts for patients with Down syndrome for weight, height, and head circumference.^{20,21} These variations alter biomechanics, affect gait, and may contribute to certain orthopaedic pathologies such as pes planus.²²

Decreased Bone Mineral Density

At baseline, patients with Down syndrome have lower bone mineral density than age matched controls.²³ This disparity increases with age²³ and may be related to al-

tered cellular signaling pathways, low muscle tone, and endocrine abnormalities.^{24,25} Evaluation is complicated as the traditional DEXA scan may underestimate bone mineral density. Additionally, these patients may demonstrate a higher vitamin D requirement.²⁴ Reza and colleagues found that adding weight-bearing exercise and calcium supplementation improved bone mineral density in children with Down syndrome.²³

Thyroid Disease

Thyroid disease presents in a variety of ways in Down syndrome patients including subclinical hypothyroidism with pain, congenital hypothyroidism, Hashimoto's Disease, and Grave's Disease.²⁴ Signs and symptoms of hypothyroidism in children are highly variable but may include constipation, dry skin, poor growth, muscle pain, fatigue, and cold intolerance.²⁷ It is recommended by the American Academy of Pediatrics that children with Down syndrome have screening TSH levels at birth, 6 months, and then annually beginning at 1 year of age.¹⁷ Pierce and colleagues found that risk of developing thyroid disease in children with Down syndrome increases by 10% per year with increasing age and up to 50% of children with Down syndrome will have thyroid disease by adulthood.²⁶ Patients with Down syndrome who present with slipped capital femoral epiphysis should be evaluated for underlying thyroid disease and thyroid function should be medically optimized. Failure to detect hypothyroidism can be detrimental as thyroid hormone is essential for normal neurologic and cognitive development.²⁷

Leukemia

Children with Down syndrome are at a higher risk for leukemia, including the acute myeloid and acute lymphoblastic subtypes, compared to the general population.²⁸⁻³⁰ Transient myeloproliferative disorder also presents at a higher rate and is characterized by excessive immature megakaryoblasts.³⁰ Down syndrome patients have a 2.1% risk of leukemia at 5 years of age and a 2.7% risk at 30 years of age.²⁸ The risk of acute myeloid leukemia (AML) is increased 150 fold and the risk of acute lymphoblastic leukemia (ALL) is increased 20-30 fold compared to the

general population.^{29,30} AML in Down syndrome is typically curable; however, ALL in Down syndrome is characterized by poorer survival rates than in the general population due to a higher relapse rate.³⁰ Interestingly, children with Down syndrome have a lower risk of solid organ tumors, possibly due to tumor suppressor genes on chromosome 21.²⁸ Signs and symptoms of acute leukemia include bone/joint pain, easy bruising/bleeding, and lymphadenopathy.¹⁷ Infants and young children may initially present with limp or refusal to ambulate due to leukemic infiltration of the bones and periosteum.³¹ orthopaedic surgeons must be vigilant in looking for leukemia when evaluating a patient with Down syndrome who presents with musculoskeletal pain.

Arthropathy of Down syndrome

Arthropathy of Down syndrome (ADS) has an estimated prevalence of 8.7 per 1000 patients with Down syndrome.³² ADS is typically polyarticular and rheumatoid factor negative.³² The wrist and small joints of the hand are most commonly affected.^{2,32} Foley et al. demonstrated that patients with ADS have a significantly greater proportion of erosive changes noted on x-ray than children with juvenile idiopathic arthritis.³² Diagnosis is frequently delayed for multiple reasons including lack of awareness of ADS, differences in pain expression, limited verbal skills, and hypermobility which may make the musculoskeletal exam difficult to interpret.³² Treatment of ADS follows the same treatment guidelines as juvenile idiopathic arthritis.³²

Congenital Heart Disease

Congenital heart disease affects approximately 50% of patients with Down syndrome compared to 0.3% in the general population.³³ The most common congenital heart defects in Down syndrome are atrioventricular septal defect, ventricular septal defect, persistent ductus arteriosus, atrial septal defect, and Tetralogy of Fallot.³⁴ It is recommended that all newborns undergo echocardiogram even if a fetal echocardiogram was performed for the detection of congenital heart disease.¹⁷ Congenital heart disease is typically amenable to surgical correction in this patient population.³⁴ When proposing orthopaedic

Table 2. Preoperative Considerations for Patients with Down syndrome³⁵

| | |
|-----------------------|--|
| Cervical Spine | Check lateral cervical spine radiographs for evaluation of atlantoaxial instability to help correct positioning during surgery and/or need for fiberoptic intubation |
| Cardiac | Preoperative cardiology evaluation if any known heart issues or cardiac risk |
| Pulmonary | Assessment for pulmonary hypertension and upper airway obstruction |
| Hematologic | Check complete blood count for evaluation of underlying hematologic disturbance such as myeloproliferative disorders as well as thyroid panel |

intervention, congenital heart disease must be considered as part of the preoperative work-up evaluating the status of any congenital heart repair and the presence of any residual defect that may affect the patient's overall function.³⁵

Obstructive Sleep Apnea

Obstructive sleep apnea (OSA) affects approximately 50-79% of patients with Down syndrome.¹⁷ OSA may be related to obesity, midfacial hypoplasia, central apnea, low muscle tone, large tongue, and tonsillar/adenoid enlargement.^{5,36,37} Symptoms of OSA include heavy breathing, snoring, restless sleep, daytime sleepiness, and apneic pauses.¹⁷ If not identified, OSA can lead to growth abnormalities and pulmonary or cardiac complications.³⁶ Treatment includes weight loss, tonsillectomy/adenoidectomy, and CPAP or BIPAP.^{36,37} Similarly to congenital heart disease, airway obstruction is an important preoperative consideration for children with Down syndrome undergoing orthopaedic surgery procedures.³⁵ Table 2 summarizes important and specific preoperative considerations.

Orthopaedic Management of Down Syndrome

Atlantoaxial Instability

Atlantoaxial instability is encountered in 10-30% of patients with Down syndrome by adolescence.³⁸ This articulation is stabilized primarily by the transverse ligament

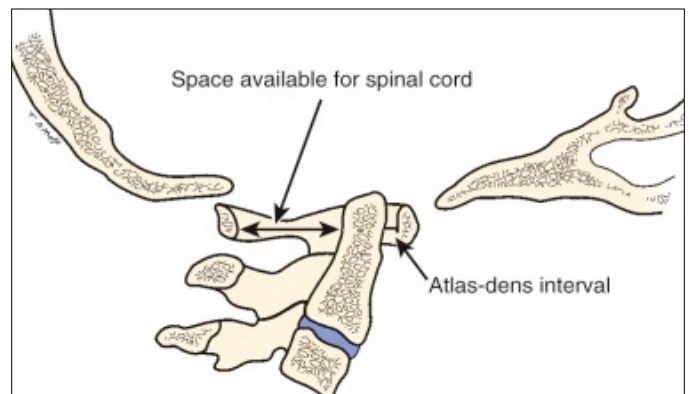


Figure 1. Murphy, Robert, Hedequist, Daniel, Glotzbecker, Michael. *Congenital Anomalies of the Cervical Spine. Rothman-Simeon and Herkowitz's The Spine. 7th edition. Philadelphia, PA. Elsevier. 2018. 609-640*

and secondarily by the alar ligaments.³⁸ Ligamentous laxity in conjunction with flattened facets is thought to predispose to instability in this patient population.^{38,39} American Academy of Pediatrics guidelines recommend cervical spine radiographic screening for atlantoaxial instability for any child with neck pain, radicular pain, weakness, spasticity or change in tone, gait difficulties, hyperreflexia, change or bowel or bladder function of signs and symptoms of cervical myelopathy.^{17,40} When a patient is symptomatic, neutral cervical spine radiographs are recommended, and if no abnormality is present, then flexion and extension radiographs may be obtained as indicated.¹⁷

Two key parameters evaluated on the lateral cervical spine radiograph are the atlas-dens interval (ADI) and



Figure 2. Radiographs of a 20-year-old male with Down syndrome with persistently symptomatic atlantoaxial instability who underwent a C1-C2 fusion.

the space available for spinal cord (SAC) also referred to as the posterior atlanto-dens interval (PADI).³⁸ The ADI is measured from the anterior aspect of the dens to the posterior aspect of the anterior ring of the atlas.³⁸ (Figure 1) Instability is considered with ADI over 5mm.³⁸ Symptoms typically occur with instability greater than 7-10 mm.^{38,41} The SAC is the distance between the posterior aspect of the odontoid or axis and the foramen magnum or posterior ring of the axis.³⁸ Patients with instability noted on plain radiograph should undergo MRI for further evaluation.⁴² Less than two percent of patients with evidence of atlantoaxial instability are symptomatic.^{43,44} Symptoms are more common in females and patients under the age of 10 and relate to spinal cord compression by the odontoid.⁴⁴ Symptoms include neck pain, gait abnormalities, hypertonicity of extremities, weakness, and bowel/bladder incontinence.⁴⁴

Patients wishing to participate in the Special Olympics are required to undergo screening with lateral cervical spine radiographs and must have an ADI of less than 4.5 mm prior to participation.⁴⁴ However, no guidelines exist for activity restriction for atlantoaxial instability and some reviews suggest patients with Down syndrome should not be screened or be barred from athletics.^{44,45}

Depending on institutional bias, children with Down syndrome undergoing surgery may be required to obtain lateral cervical x-rays prior to their procedure as part of a preoperative evaluation.⁴⁶ A study by Litman and colleagues found that 64% of anesthesiologists would obtain lateral cervical x-rays for a patient with symptoms concerning for atlantoaxial instability and 18% would obtain lateral cervical x-rays for asymptomatic patients prior to undergoing surgery.⁴⁶

Surgical intervention is considered in patients with ADI over 10 or SAC less than 14 mm.⁴⁷ Surgical options include rigid internal fixation with C1 lateral mass to C2 screws, C1-C2 transarticular screws, and transoral decompression with posterior plating.³⁸ It should be noted that approximately 50% of both cervical flexion and rotation occur at the C1-C2 articulation and are lost with fusion. (Figure 2).

Summary: Atlantoaxial instability is frequently encountered in patients with Down syndrome. The American Academy of Pediatrics no longer recommends routine screening in asymptomatic patients. Key radiographic parameters are atlanto-dens interval (ADI) and Space Available for the Cord (SAC), also known as posterior atlanto-dens interval (PADI). Surgical management includes cervical fusion. (Table 3)

Table 3. Atlantoaxial Instability Evaluation and Management^{40,43,47}

| | |
|--------------------------------------|--|
| When to get screening x-rays? | Consider in any child with neck pain, radicular pain, weakness, spasticity or change in tone, gait difficulties, hyperreflexia, change or bowel or bladder function or signs and symptoms of cervical myelopathy |
| When to get MRI? | Consider MRI when atlantoaxial instability noted on x-ray |
| When to surgically intervene? | ADI >10 |
| When to restrict sports? | Controversial—consider restriction with ADI > 4.5 mm |

Atlanto-Occipital Instability

While much of the literature pertaining to cervical spine pathology addresses atlantoaxial instability, more recent attention has been directed towards instability at the atlanto-occipital articulation. The prevalence of atlanto-occipital instability has been reported in up to 64% of patients with Down syndrome.⁴⁸ Normal atlanto-occipital stability is created by the cup-shaped atlanto-occipital articulation which allows flexion and extension while preventing lateral and rotational stress.^{39,49} Additional stability is created by the tectorial membrane, the alar ligaments, and the apical ligaments.^{39,49} Patients with Down syndrome may have dysplastic architecture of the occipital condyle resulting in a flat joint³⁸ and instability is further exacerbated by ligamentous laxity.⁴⁸ Radiographic evaluation of occipitocervical dislocation involves use of the Powers ratio which is determined by dividing the distance from the basion to the posterior arch by the distance from the anterior arch to the opisthion (Figure 3).⁵⁰ A Powers ratio of over 1.0 is concerning for anterior dislocation while a power ratio of less than 1.0 is concerning for posterior atlanto-occipital dislocation.⁵⁰ The Harris Rules of 12 uses the basion-dens interval (BDI), with a value over 12 mm considered positive for atlanto-occipital instability.⁵¹ Alternatively the Weisel-Rothman technique can be used, which uses three points with point 1 is located at most caudal point of posterior arch of the atlas, point 2 at the center of the anterior arch of the atlas, and point 3 at the basion (Figure 4).^{52,53} A perpendicular line is drawn at the posterior edge of the anterior arch of the atlas and the distance between this

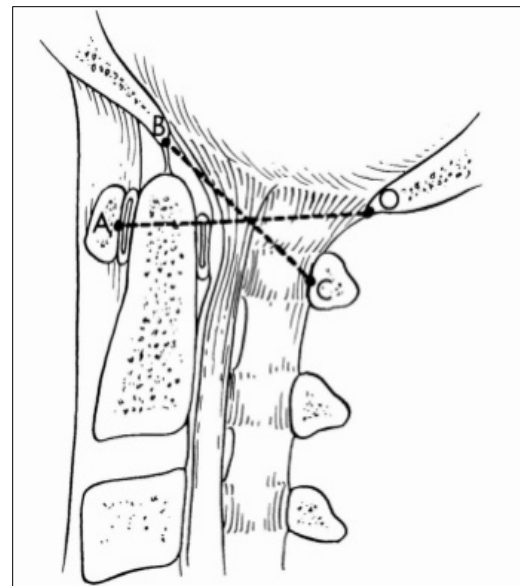


Figure 3. Powers ratio BC/OA . Powers B, Miller MD, Kramer RS, Martinez S, Gehweiler JA Jr. *Traumatic anterior atlanto-occipital dislocation. Neurosurgery.* 1979 Jan;4(1):12-7. doi: 10.1227/00006123-197901000-00004. PMID: 450210.

perpendicular line and point 3 is measured in flexion and extension with a normal value being under 1 mm.^{52,53} Indications for treatment of atlanto-occipital instability are poorly defined, but treatment typically involves occipitocervical fusion.

Summary: Atlanto-occipital instability occurs frequently in patients with Down syndrome. The Powers ratio and basion-dens interval are common radiographic parameters for evaluation of atlanto-occipital instability. Symptomatic instability can be addressed with occipital cervical fusion.

Scoliosis

Ten percent of noninstitutionalized and up to 50% of institutionalized patients with Down syndrome have scoliosis.^{54,55} Thoracogenic scoliosis as a result of prior thoracotomy for treatment of congenital heart disease increases the risk between 22 and 31%.^{56,57} Literature regarding the treatment of scoliosis in Down syndrome is limited but treatment typically mirrors that of adolescent idiopathic scoliosis with bracing and surgery based on Cobb angle progression. Their complication rate, however, is much higher than that of the idiopathic type. Milbrandt et al. found that children with Down syndrome undergoing spinal fusion for scoliosis had a 57% complication rate including pseudarthrosis, implant failure, superior junctional kyphosis, and infection.⁵⁵ In comparison, the Scoliosis Research Society found a 5.7% complication rate in children with adolescent idiopathic scoliosis.⁵⁸

Summary: Scoliosis is common in patients with Down syndrome. Treatment mirrors that of children with adolescent idiopathic scoliosis; however, there is a higher risk of complications.

Spondylolysis/Spondylolisthesis

The incidence of spondylolysis and spondylolisthesis in the general population is 3-6% and 2.7-8.4%, respectively.⁵⁹ A cross-sectional study by Hansdorfer et al. demonstrated the incidence of spondylolysis and spondylolisthesis in patients with Down syndrome to be 18.7% and 32.7%, respectively.⁵⁹ Interestingly low back pain and leg pain were more frequent in Down syndrome patients with spondylolisthesis than in patients with spondylolisthesis in the general population.⁵⁹ There have been no studies to date regarding treatment of spondylolysis or spondylolisthesis specifically in patients with Down syndrome.

Summary: Spondylolysis and spondylolisthesis may be more common in children with Down syndrome and they also tend to be more symptomatic, but there are no specific treatment recommendations specific to Down syndrome.

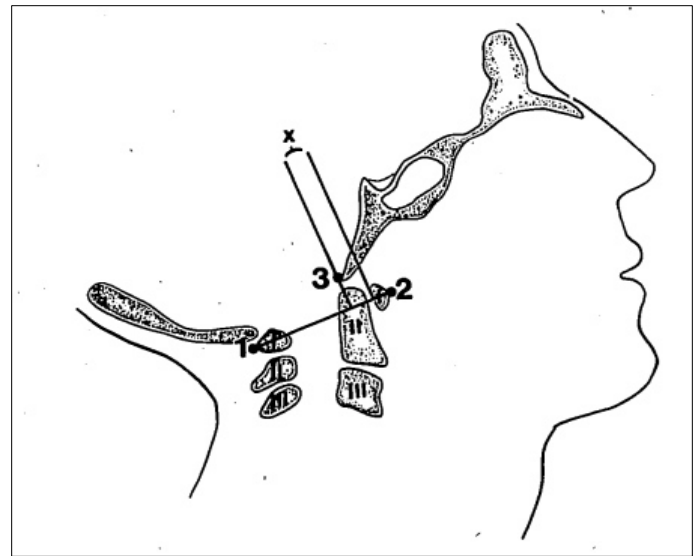


Figure 4. Weisel-Rothman Technique. Point 1 is located at most caudal point of the posterior arch of the atlas, point 2 is at the center of the anterior arch of the atlas, and point 3 is at the basion. The difference between point 3 and a perpendicular line is drawn at the posterior edge of the anterior arch of the atlas is calculated in flexion and extension. Gabriel KR. Occipito-atlanta translation in Down's syndrome. *Spine*. 1990;15(10):997-1002. doi:10.1097/00007632-199015100-00003

Hip Instability

The prevalence of hip instability in patients with Down syndrome is 1-7%⁶⁰ and can result in progressive loss of mobility and function. The etiology of hip instability is likely a combination of hypotonia, ligamentous laxity, and abnormal hip morphology.²⁹ Bennet et al. described the natural history of hip instability in patients with Down syndrome with regard to clinical and radiologic features in a series of 45 dislocations in 28 patients.⁶¹ The initial presentation seen in children less than 2 years old is delayed walking and generalized muscular hypotonia and ligamentous laxity leading to hypermobile hips. Next, from age 2 to 8 years hip instability may progress to dislocations that can be easily reduced. After age 8 years, hip subluxation progresses with loss of concentric reduction and acetabular dysplasia.⁶¹ The final phase occurs in late adolescence or early adulthood with a fixed dislocated hip (Figure 5).^{60,61}

Patients often have abnormal hip morphology with coxa valga and acetabular retroversion.^{60,62} Coxa valga has been reported in over two-thirds of adults with Down syndrome with a mean neck shaft angle of 167 degrees.^{62,63} Sankar et al. demonstrated that pediatric patients with Down syndrome had a mean acetabular version of 2.1 +/- 11 degrees compared to a control group that had an acetabular version of 12.4 +/- 5.5 degrees.⁶³ Secondary changes to the acetabulum include a reduced center edge angle, increased Tonnis angle, and widening of the acetabular teardrop.⁶⁴ Patients often have posterior instability due to poor posterior acetabular coverage secondary to insufficient posterior wall or acetabular retroversion.⁶⁰

Management of hip instability depends on both age and phase of hip instability.⁶⁴ For patients under age 2, non-operative treatment is typically preferred.⁶⁰ For older patients with habitual dislocation without secondary acetabular dysplasia, treatment should aim to stabilize the hip and prevent secondary acetabular dysplasia.^{60,64} Kelley et al. recommends that patients undergo femoral varus osteotomy with or without derotation.⁶⁴ On the other hand, Sankar et al. recommends an isolated anteverting triple osteotomy of the acetabulum for habitual dislocation.⁶⁵ For patients with fixed subluxation, treatment is similarly aimed at obtaining a concentrically reduced hip and correcting acetabular dysplasia.^{60,64} Kelley et al. recommends Bernese periacetabular osteotomy with a closed triradiate cartilage or a triple osteotomy with open triradiate cartilage in these patients.⁶⁴ Patients with fixed dislocation may require total hip arthroplasty, especially in older patients with osteoarthritis.⁶⁰ Yet, patients who undergo total hip arthroplasty (THA) are at a higher risk of perioperative medical problems (urinary tract infections, pneumonia, and increased length of stay) and surgical complications when undergoing THA.⁶⁶

Summary: Hip instability is common in patients with Down syndrome due to ligamentous laxity and abnormal hip morphology. Treatment depends on age at presentation, degree of instability. Various surgical

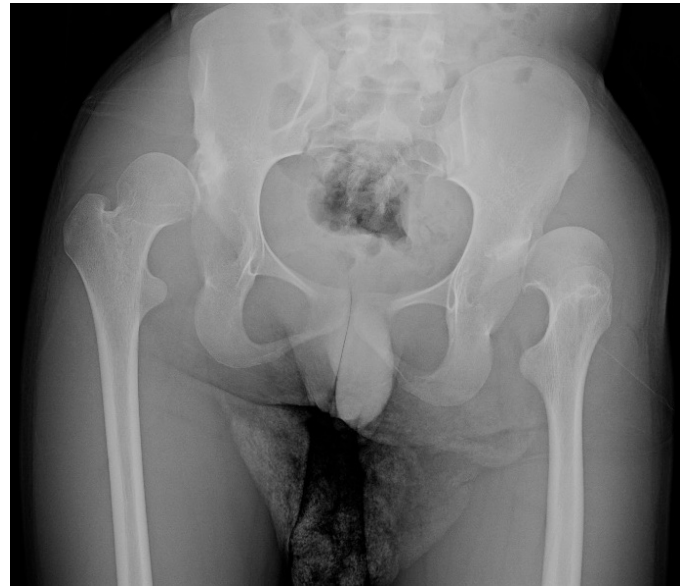


Figure 5. Radiograph of a 15-year old male with bilateral hip dislocations

techniques have been described, all which aim at containment of a concentric hip whenever possible.

Slipped Capital Femoral Epiphysis

The incidence of SCFE in children with Down syndrome is 1.3% compared to 0.01% children in the general population.^{67,68} Children with Down syndrome are more likely to present with unstable and high-grade slips.⁶⁹ Dietz et al. found six of eight children with Down syndrome and SCFE to have a grade III SCFE and five of the eight children developed avascular necrosis despite operative treatment.⁶⁹ Bosch et al. evaluated eight children with Down syndrome and SCFE and found that three patients had bilateral slips and that six of the eight children were found to have hypothyroidism.⁶⁷ This data suggests that children with Down syndrome may present with unstable and high-grade slips.⁶⁷ Furthermore they should be evaluated for hypothyroidism.⁶⁷

Summary: Down syndrome has associated with higher rates of SCFE and may present with unstable and high-grade slips. Down syndrome patients who present with SCFE should be evaluated for hypothyroidism. Pinning of the contralateral hip should be considered in cases of unilateral slipping.

Patellofemoral Instability

The prevalence of patellofemoral instability in children with Down syndrome has been reported to be 10-20% (Figure 6).^{70,71} Patellofemoral instability is rarely painful in children with Down syndrome; however, they can present with frequent falls, limping, and pain.⁷³ Patellofemoral instability in Down syndrome was classified by Dugdale et al. where grade 1 indicates a stable patellofemoral joint, grade 2 indicates the patella can be subluxated laterally more than one-half of patellar width, grade 3 indicates the patella is dislocatable, grade 4 indicates a dislocated patella that can be reduced and grade 5 is an irreducible patella.⁷²

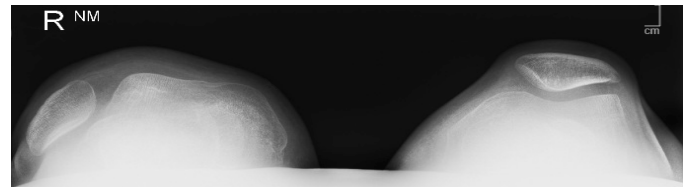
Nonoperative treatment is typically recommended as initial management.^{72,74} Evidence for surgical options for patellofemoral instability in patients with Down syndrome is limited and is primarily based on case reports or case series with few patients and differing techniques. Dugdale et al. evaluated the knees of 361 patients with Down syndrome and described the surgical treatment of eight knees in five patients with Down syndrome using differing techniques in each patient, noting unsatisfactory results in four of the eight knees treated.⁷² They concluded that patellar instability is well tolerated and that moderate to severe patellar instability was rarely disabling.⁷²

Several techniques can be utilized to address patellofemoral instability in patients with Down syndrome. The Roux-Goldthwait-Campbell procedure, Green's quadricepsplasty, with and without modified Galeazzi procedure, and a Camanho's modified MPFL reconstruction with lateral release and medial capsulectomy have had varying degrees of success and can be utilized to stabilize the patellofemoral joint in this population.⁷³⁻⁷⁶

Two excellent reviews in *JPOSNA* (Vol. 2, No. 2, August 2020) by Imbergamo et al. and Lin et al. describe a stepwise approach to PF instability in all patients with Down syndrome.



Figure 6. A 16-year-old child with Down syndrome with fixed dislocated right patella. In addition to likely ligamentous laxity, the child's genu valgum will make a soft tissue only reconstruction prone to failure.



Summary: Initial management of patellofemoral instability is nonoperative, even in moderate to severe cases. Symptoms should drive treatment rather than severity of dislocation. Many different techniques have been reported to address patellofemoral instability. Given the inherent laxity of Down syndrome patients, failure rates are relatively high.

Pes Planus and Hallux Valgus

Foot and ankle deformity are commonly encountered in patients with Down syndrome and are reported to account for 30% of reported orthopaedic referrals in children with Down syndrome.² Common foot and ankle disorders include pes planus and hallux valgus. As a result of these conditions, patients with Down syndrome may develop out-toeing and a wider base of support with "poor foot control."⁷⁷ Two studies have found that 76 to 91% of patients in with Down syndrome had pes planus that usually persists into adulthood.^{2,78,80} Numerous nonoperative

treatments have been described including heel wedges, heel cups, shoe inserts, and custom made shoe orthotics.⁸¹ However, there is limited evidence whether orthotic use can change the natural course of asymptomatic flat-foot.^{81,82} Multiple studies have shown benefit of orthotics in patients with painful pes planus.^{83,84}

Hallux valgus has been reported in 10 to 60% of patients with Down syndrome^{78,85} and may be related to structural factors including metatarsus varus, pes planus, ligamentous laxity, and a tight heel cords.^{86,87} In children with Down syndrome, hallux valgus can be associated with foot-specific disability during school and play activities and the use of narrow-fitting footwear has been associated with increased disability.⁷⁸

Summary: Management of hallux valgus and pes planus is mainly nonoperative. Orthotics such as SMOs have not been shown to change the natural history of painless flexible flatfoot in Down syndrome.

Summary

There are a variety of musculoskeletal and medical conditions the orthopaedic surgeon must be familiar with when treating the child with Down syndrome. These conditions are thought to be related to the generalized ligamentous laxity, joint hypermobility, and hypotonia. (Table 4). These musculoskeletal conditions include patellar instability, hip instability, atlantoaxial and atlanto-occipital instability, scoliosis, spondylolisthesis, SCFE, pes planus, and hallux valgus. Involvement of a multi-disciplinary team is essential when treating children with Down syndrome (Appendix: page 17). When the orthopaedist treats a musculoskeletal condition in a child with Down syndrome, the entire child must be taken into consideration. One must be thoughtful of possible other musculoskeletal ailments, other comorbidities, and the social implications. The goal of treatment is not only to reduce pain or to gain function but to make the child a functional and independent member of society.

Table 4. Summary of Orthopaedic Conditions in Down syndrome

| System | Pathology | Diagnosis | Treatment |
|--------|-------------------------------------|--------------------------------------|---|
| Spine | Atlantoaxial instability | Cervical spine x-rays (ADI, SAC) | Depends on degree of instability, can include observation, avoidance of activities and even fusion. |
| | Atlanto-occipital instability | Cervical spine x-rays (Powers ratio) | Depends on degree of instability, can include observation, avoidance of activities and even fusion. |
| | Spondylolysis and Spondylolisthesis | Lumbar spine x-rays | Mostly nonoperative |
| Hip | Hip instability | Hip/pelvis x-rays | Initially nonoperative, surgery depending on phase of instability |
| | SCFE | Hip/pelvis x-rays | In situ hip pinning, workup for possible hypothyroidism |
| Knee | Patellar instability | Knee x-rays (Dugdale classification) | Mostly nonoperative, surgical options as described |
| Foot | Pes planus | Foot x-rays | Mostly nonoperative, modified shoe wear and orthotics if symptomatic |
| | Hallux Valgus | Foot x-rays (HVA, IMA, DMAA) | Mostly nonoperative |

Additional Links

National Down Syndrome Society:

<https://www.ndss.org/>

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Appendix**Health supervision and screening (adopted from Pediatrics 2011;128:393–406)**

| Age | Health supervision/screening |
|----------------------|--|
| Birth to 1 month | Echocardiogram for evaluation of heart defects Feeding problems Cataracts Congenital hearing loss with auditory evoked response of otoacoustic emission Duodenal atresia Car seat safety evaluation Constipation Gastroesophageal reflux Complete blood count for hematologic abnormalities Thyroid-stimulating hormone concentration |
| 1 month to 1 year | Pediatric ophthalmology referral for evaluation of strabismus, cataracts, and nystagmus Repeat TSH at 6 months and 12 months Monitor for cardiac defects Obtain hemoglobin concentration at 1 year Monitor for neurologic dysfunction Discuss importance of cervical spine neutral position with anesthesia, surgical, or radiographic procedures |
| 1 year to 5 years | TSH annually Referral to pediatric sleep study for evaluation of obstructive sleep apnea Monitor for neurologic dysfunction Hemoglobin concentration annually CRP and ferritin for child at risk of iron deficiency Cervical spine radiologic evaluation in symptomatic patients after age 3 |
| 5 years to 13 years | Monitor growth patterns Annual ear-specific audiologic evaluation Ophthalmologic evaluation every 2 years TSH annually Hemoglobin concentration annually and serum ferritin and CRP or reticulocyte concentration annually if at risk for iron deficiency Monitor for neurologic dysfunction |
| 13 years to 21 years | Monitor growth patterns Annual ear-specific audiologic evaluation Ophthalmologic evaluation every 2 years TSH annually Hemoglobin concentration annually and serum ferritin and CRP or reticulocyte concentration annually if at risk for iron deficiency Monitor for neurologic dysfunction |