## ORTHOPAEDIA: PEDIATRICS

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## THE CODMAN GROUP

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Please visit *http://www.orthopaedia.com* for the most current version of this text. At the website, you will also find its sister publications covering Foot & Ankle disorders, Hand, Sports Medicine, Spine, Fractures, and others as they become available.

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Pediatrics is a unique specialty within the realm of orthopaedics and musculoskeletal medicine. Unlike those physicians who focus on one region of the body or one type of disease, pediatric musculoskeletal specialists are given the opportunity to care for the whole patient (head to toe, quite literally) across many types of pathology, including trauma, tumor, and congenital conditions.

The pediatric musculoskeletal specialist is also afforded the chance to develop a relationship with patients and their families, watching patients mature and grow.

Pediatric musculoskeletal medicine offers treatments that can (almost) cure. Relocating a dislocated hip in the neonate may allow a fairly normal hip joint to develop; a forearm fracture sustained at age 6 might have no residual consequence in the adult at all. (No knock on my adult-focused colleagues, but a knee joint with a reconstructed cruciate ligament is not a normal knee joint, and a total knee replacement is not a "knee" joint at all.)

Of course, on the other hand, the pediatric musculoskeletal specialist also takes care of some diseases that are, if not progressive, irreversible. (The six chapters in the section "Musculoskeletal Aspects of Pediatric Syndromes" provide plenty of examples.) Yet in those cases too, the physician can make a big difference, substantially improving the patients' quality of life.

This book – a collaborative effort by more than fifty leading physicians and researchers from dozens of children's hospitals, healthcare systems and medical schools – was conceived as a tool for students, a tool that will help them improve their future patients' quality of life. Its aim is to contribute toward a foundation of knowledge that will allow physicians to make a positive impact.

I decided to practice pediatric orthopaedics for that very reason: to engage in work that has the potential for positive, lifelong impact on not only patients, but on their families as well. I try to live up to the standard articulated by Nelson Mandela: "History will judge us by the difference we make in the everyday lives of children." I consider it the ultimate privilege to care for a child. It is my hope this text helps others acquire the tools that will let them have a similar positive impact as well.

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There is a great profusion of medical information available for free on the Internet, and a lot of it is good. Yet even good information may not be completely useful to the reader who may not know if it is trustworthy. By contrast, there is also a lot of information of medical information available for sale that is produced by well known authors and organizations, though not always for free.

This volume aims to be both free and accurate.

To ensure medical accuracy, all of the material was reviewed by the section editors, who are of course content experts of great renown. In addition, each chapter was reviewed by an expert who was not involved in the creation of the material. These reviewers were asked to read the chapter with one overriding goal in mind: to detect errors. The reviewers were then asked to "certify" the chapter as a reasonable presentation of the topic without any glaring mistakes in content. We are grateful to our reviewers, listed below:

- Fractures of the Growth Plate was reviewed by Jon Schoenecker MD PhD. Dr. Schoenecker is an Associate Professor of Orthopaedic Surgery at Vanderbilt University.

- Fractures of the Upper Extremities was reviewed by Anthony Riccio MD. Dr. Riccio is a Professor of Orthopaedic Surgery at UT Southwestern Medical Center.

- Fractures of the Lower Extremities was reviewed by Ying Li MD. Dr. Li is an Associate Professor of Orthopaedic Surgery at the University of Michigan.

- Non-Accidental Trauma / Child Abuse was reviewed by Marty Herman MD. Dr. Herman is a Professor Orthopaedic Surgery at Drexel University College of Medicine.

- Musculoskeletal Infections was reviewed by Julia Sanders MD. Dr. Sanders is an Assistant Professor of Orthopaedic Surgery at the University of Colorado.

- Lower Limb Deformity was reviewed by Ray Liu MD. Dr. Liu is a Professor of Orthopaedic Surgery at Case Western Reserve University of Medicine.

- Foot Deformities was reviewed by B. David Horn MD. Dr. Horn is an Associate Professor of Orthopaedic Surgery at the University of Pennsylvania School of Medicine.

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- Developmental Dysplasia of the Hip was reviewed by Salil Upasani MD. Dr. Upasani is an Associate Professor of Orthopaedic Surgery at the University of California San Diego School of Medicine.

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- Slipped Capital Femoral Epiphysis was reviewed by Michelle S. Caird MD. Dr. Caird is a Professor of Orthopaedic Surgery at the University of Michigan.

- Adolescent Idiopathic Scoliosis was reviewed by Michael Glotzbecker MD. Dr. Glotzbecker is an Associate Professor of Orthopaedic Surgery at Rainbow Babies and Children's Hospital.

- Early Onset Scoliosis was reviewed by Lindsay Andras MD. Dr. Andras is an Associate Professor of Orthopaedic Surgery at Keck School of Medicine of USC.

- Scheuermann's kyphosis was reviewed by Jeff Sawyer MD. Dr. Sawyer is a Professor of Orthopaedic Surgery at the University of Tennessee Health Science Center, Campbell Clinic.

- Down Syndrome was reviewed by Ken Illingworth MD. Dr. Illingworth is an Assistant Professor of Orthopaedic Surgery at Keck School of Medicine of USC.

- Osteogenesis Imperfecta was reviewed by Paul Sponseller MD MBA. Dr. Sponseller is a Professor of Orthopaedics at Johns Hopkins University School of Medicine.
- Muscular Dystrophy was reviewed by Judson Karlen MD. Dr. Karlen is an Assistant Professor at the University of Arizona College of Medicine.
- Spina Bifida was reviewed by Colyn Watkins MD. Dr. Watkins is an Instructor of Orthopaedic Surgery at Harvard Medical School.
- Cerebral Palsy was reviewed by Keith Baldwin MD MPH MPST. Dr. Baldwin is an Associate Professor of Orthopaedic Surgery at the University of Pennsylvania School of Medicine.
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The material was also reviewed by the following students from the Perelman School of Medicine at the University of Pennsylvania:

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*This material is not intended to represent the only, nor necessarily best, method or procedure appropriate for the medical situations discussed, but rather is intended to present an approach which may be helpful to others who face similar situations. We cannot can take any responsibility for the consequences following the application of any of the information presented here.*

***The information provided here cannot substitute for the advice of a medical professional.*** *Even if a given statement is completely true in the abstract, it may not apply to a given patient.*

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The final version of this volume was produced by editing, refining and merging the work of the contributing editors listed below. These experts generously produced the first drafts of the chapters, and even more generously allowed their work to be edited, refined and merged, according to the overall needs of the project.

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# ACUTE CONDITIONS

# FRACTURES OF THE GROWTH PLATE (PHYSIS)

## THE PHYSIS

The physis, or growth plate, is a complex cartilaginous structure that is responsible for longitudinal growth of the skeleton. In order to allow for bone growth, the physis is not fully ossified. That makes it an area of weakness and prone to fracture.

## ANATOMY AND STRUCTURE

Long bones in children have four distinct segments (Figure 1): The epiphysis is the region of bone adjacent to the joint surface. Below it lies the physis, the area where growth occurs. Distal to that is the metaphysis, a flared region of bone, and below that lies the narrower shaft of the bone, or diaphysis. (A nice way to remember the meaning of these terms is to consider the etymology: "physis" means "origin" (of bone); "epi" means "above"; "meta" means "beyond"; and "dia" means "through" or "towards".



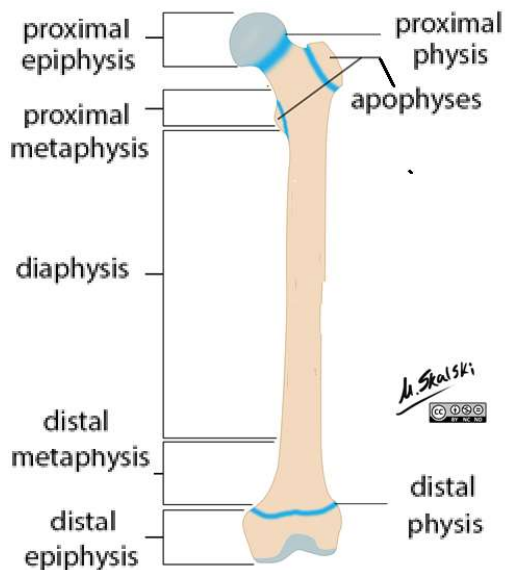*Figure 1: Bone segments of the femur. As the figure makes clear, in a long bone there is only one diaphysis, but there can be physes and associated regions at both ends. Also shown are the apophyses, the growth plates of the secondary growth centers where tendons attach [In the proximal femur, these will become the trochanters.] (Modified from Dr Matt Skalski, Radiopaedia.org. From the case https://radiopaedia.org/cases/29729)*

The physis is found between the epiphysis and the metaphysis. Here cartilage cells progress through a series of layers and mature to create new bone that is added to the metaphysis. The physis itself can be broken down into four zones, beginning at the epiphysis and ending with new bone formation at the metaphysis (Figure 2). The germinal or reserve zone contains chondrocytes, or cartilage cells, in a relatively quiet state that will serve as the source of cells for growth. In the proliferative zone, the cells begin to divide, flatten, and form columns. Next, in the hypertrophic zone, the cells grow, mature and spread out as they produce additional extracellular matrix. This is the weakest layer of the growth plate and the most common location of physeal fractures. Finally, in the zone of provisional calcification the cartilaginous matrix begins to calcify, and immature bone formation occurs.
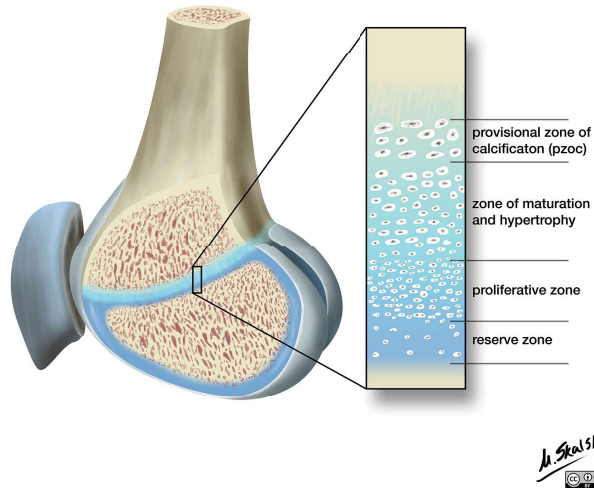
# Physeal anatomy



*Figure 2: Physeal anatomy showing the four zones. (Case courtesy of Dr Matt Skalski, Radiopaedia.org. From the case https://radiopaedia.org/cases/27354)*

Horizontal growth also occurs at the physis, through a circumferential collection of immature cells at the periphery known as the Node (or groove) of Ranvier. Overlying this is a layer of strong fibrous tissue that stabilizes the physis by connecting the epiphysis to the periosteum of the metaphysis, known as the perichondral ring of LaCroix. Blood supply to the growth plate comes from several sources. Epiphyseal vessels supply the resting and proliferative zones. Additionally, metaphyseal-sided blood supply comes from perforating metaphyseal and diaphyseal intramedullary nutrient vessels. There is also a perichondral artery that enters at the Node of Ranvier. Damage to these blood vessels can contribute to growth arrests that occur after fractures through the growth plate.

## EPIDEMIOLOGY

Physeal fractures make up 15-30% of all fractures in children. They are more common in boys than in girls (2:1), and occur more frequently in adolescents during periods of rapid growth when the physis is weaker. Growth plate fractures are typically the result of trauma and often occur due to falls engaging in sports and other recreational activities. Physeal fractures can, however, be caused by high energy mechanisms as well as physical abuse. The physis is an area of weakness and is less resistant to stress than the surrounding bone, ligaments, and other soft tissues. As such, mechanisms of injury that in an adult would cause sprain ligaments more likely will cause physeal fractures in skeletally immature patients.

## EVALUATION

Patients presenting after mechanisms of injury suggestive of physeal fractures report pain at the end of the bone (near the joint), an inability to use or put weight on the extremity, and possible visible deformity. The skin should be carefully examined for signs of soft tissue injury including bruising, swelling, bleeding, open wounds, and exposed bone. The joints above and below the area of suspected injury should be palpated and brought through a range of motion as pain allows. Particular attention should be paid to confirming the presence of distal pulses and capillary refill and whether they are symmetric to the contralateral side. As with all orthopaedic injuries, it is important to evaluate for other associated injuries by checking for tenderness, deformity, crepitus, and pain with range of motion in all four extremities, the pelvis, and spine. Biplanar radiographs, typically anterior-to-posterior and lateral views, should then be obtained.

## SALTER-HARRIS CLASSIFICATION

There are several classification systems describing physeal fractures, however Salter-Harris is the most commonly used (Figure 3). It describes the anatomic location of the fracture while also providing prognostic information for outcomes and complications. The Salter-Harris classification of a given fracture is determined by following the path of the fracture line through the physis and the surrounding metaphysis and epiphysis bone segments.
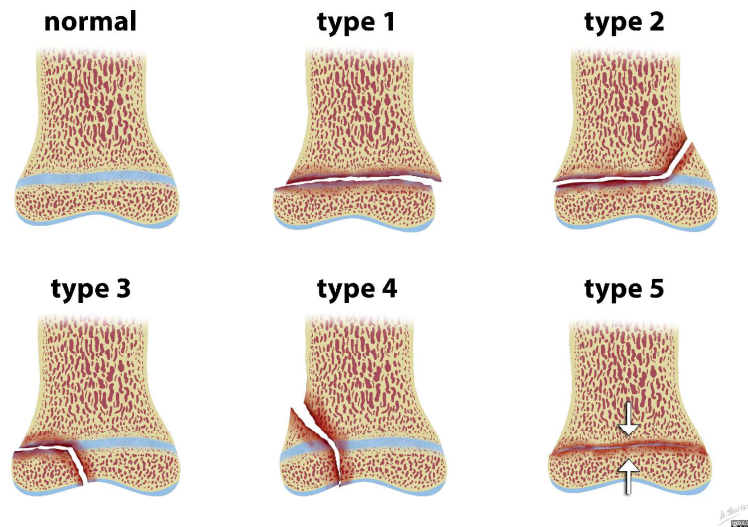


Figure 3: The Salter-Harris classification system for physeal fractures. (Case courtesy of Dr Matt Skalski, Radiopaedia.org. From the case https://radiopaedia.org/cases/27144)

Type I – The fracture travels transversely through the growth plate. Also known as a physeal separation, there is no extension into the metaphysis or epiphysis (Figure 4). If non-displaced, they can be difficult to diagnose on radiographs. In these cases, tenderness directly over the growth plate is the best indication of injury.

Type II – The fracture travels transversely through the growth plate and extends vertically into the metaphysis, typically resulting in a triangular fragment of metaphysis of variable size that remains attached to the physis and epiphysis (Figure 5). Among the definitively diagnosed fractures, these are the most common Salter-Harris fracture type (noting that non-displaced Type I fractures, which are likely the most common, may be under-diagnosed).



Figure 4: Salter-Harris I fracture of the distal femoral physis, treated with closed reduction and percutaneous pinning.

Figure 5: Salter-Harris II fracture of the distal femoral physis showing a large metaphysis fragment (Thurston-Holland fragment). Treated with open reduction and cannulated screw placement.

Type III – The fracture travels transversely through the growth plate and extends vertically into the epiphysis and typically to the articular surface of the joint (Figure 6). These are more prevalent in older children in whom the physis has begun to close.

Type IV – The fracture travels vertically through the metaphysis, across the physis, and through the epiphysis (Figure 7).

*Figure 6: Salter-Harris III fracture of the distal femoral physis showing the fracture line splitting the epiphysis and continuing to the knee joint. Treated with open reduction to ensure restoration of the articular surface and held with cannulated screws.*



*Figure 7: Salter-Harris IV fracture of the distal tibia fracture showing the fracture line traveling through the metaphysis, through the physis, and into the epiphysis exiting in the ankle joint. Treated with open reduction and cannulated screw fixation.*

Type V – Crush injury to the growth plate resulting from a compression force. This type might be difficult to appreciate on radiographs, but may be recognized by narrowing of the physis or compression of the nearby metaphysis.

## DIFFERENTIAL DIAGNOSIS

If a fracture is visible on an x-ray, there is no differential diagnosis per se: the injury is seen and diagnosed definitively. In those cases, the "differential diagnosis list" is really the "additional diagnosis list" – that is, the physician must exclude other injuries. It is possible, however, that a growth plate fracture is present yet the

fracture is not visible on an x-ray because it is not displaced – the x-ray will appear normal. Here, the differential diagnosis is that of a sprain. However, as noted, a growth plate fracture is more likely, owing to the relative strength of the ligament to that of the physis.

## RED FLAGS

Mechanisms of injury that in an adult would cause sprain ligaments more likely will cause physeal fractures in skeletally immature patients. Thus, a 12 year old who twisted his ankle and had normal appearing x-rays may nonetheless not have sprained his ankle (as would be assumed in the adult) but indeed have a growth plate fracture. This patient needs to have his ankle immobilized, and either treated empirically as if a fracture were present (see below) or re-assessed 10-14 days after injury.

## TREATMENT OPTIONS AND OUTCOMES

Treatment of physeal fractures is determined on a case-by-case basis depending on the anatomic location, amount of displacement, Salter-Harris type, and involvement of the articular surface. To limit the potential for growth arrest and to maintain function, the goals of treatment are to restore alignment of the growth plate and any involved joint surface.

In general, Type I and II fractures with minimal or no displacement are treated with closed reduction (if needed) and immobilization (Figure 8). Most patients have a good clinical outcome. However, fractures that are widely displaced and or unstable may require surgical stabilization (Figures 5 and 6). Bear in mind as well that widely displaced fractures often have soft tissue blocks to reduction (typically a piece of periosteum, muscle, or tendon). If a reduction is difficult, one should not force it.



*Figure 8: Displaced Salter-Harris II fracture of the distal radius, treated with closed reduction and casting.*

Salter-Harris Type III, IV and V require surgical intervention to realign the physis and ensure articular congruity (Figures 7 and 8). In all fracture types, the number of reduction attempts should be limited to prevent damage to the growth plate. If possible, it is best to avoid crossing the growth plate with implants. This is however necessary in some cases to maintain stability, and when needed smooth, small diameter implants should be used preferentially.

The most concerning potential complication from a fracture involving the growth plate is growth arrest. In any physeal fracture there is the potential for damage to the growth plate tissue with formation of a "bony bar" across the physis that can disrupt the normal bone growth. Partial growth arrests can lead to asymmetric growth and result in angular deformities (as say the lateral aspect of the bone continues to grow whereas the medial does not). Complete growth arrests can result in a clinically significant limb length discrepancy.

The risk growth disturbance increases with increasing Salter-Harris type number. In Type I and Type II injuries, the fracture typically travels through the hypertrophic zone and the germinal and proliferative zones stay in continuity. The likelihood of growth disturbance is therefore lower. In some regions of the body, such as the distal femur and proximal tibia, where the growth plate is undulated (that is, more curvilinear), Type I and II fractures are at higher risk of growth disturbance.

In Type III and IV fractures, the germinal and proliferative zones are interrupted leading to higher rates of physeal bar formation and growth disturbance. Type V crush injuries cause severe damage to all layers of the physis and have a very high rate of growth arrest.

Growth arrest and physeal bars can be treated in a variety of ways. If relatively small, the bone that crosses the growth plate can be removed and that space filled with bone wax, fat or other fillers. Typically, this is best reserved for bony bars that are less than 50% of the growth plate surface area and in patients who have at least two years of growth remaining. Larger bars are usually best treated by completing the growth arrest and addressing any resulting deformities. In the case of angular deformities, osteotomies can be used to restore mechanical alignment of the limbs. For unequal limb lengths, procedures to lengthen the short bone can be performed or the growth of the longer bone can be stopped at the appropriate time to allow the shorter limb to "catch up".

## MISCELLANY

The Salter-Harris classification was developed by two orthopaedic surgeons. Dr. Robert B. Salter was a Canadian pediatric orthopaedic surgeon who also created the Salter osteotomy for treatment of developmental dysplasia of the hip. Dr. William H. Harris is an American orthopaedic surgeon best known for developing the Harris Hip Score, an outcome studies measurement (and the focus of the most highly cited paper in orthopaedic surgery [as of 2020]).

## KEY TERMS

Growth plate, physis, epiphysis, metaphysis, diaphysis, reserve zone, proliferative zone, zone of hypertrophy, zone of provisional calcification, Salter-Harris classification, fracture, growth arrest, physeal bar, reduction.

## SKILLS

Identify the four basic segments of a long bone. Identify and describe the four cellular layers of the physis. Describe the Salter-Harris classification and correctly apply it to radiographs of bone injuries.

# PEDIATRIC FRACTURES OF THE UPPER EXTREMITIES

## STERNOCLAVICULAR DISLOCATION

Sternoclavicular (SC) injuries in skeletally immature patients can occur as a result of either a true sternoclavicular dislocation or a medial clavicular physeal separation. The medial clavicular physis is the last to ossify and can fuse as late as 25 years old, so physeal separations can occur in young adults as well. SC injuries are relatively uncommon and account for <1% of pediatric fractures. The clavicle more commonly displaces anteriorly to the sternum, but posterior displacement is more dangerous, as the posteriorly displaced clavicle can cause injury or compression to vital mediastinal structures (Figure 1).
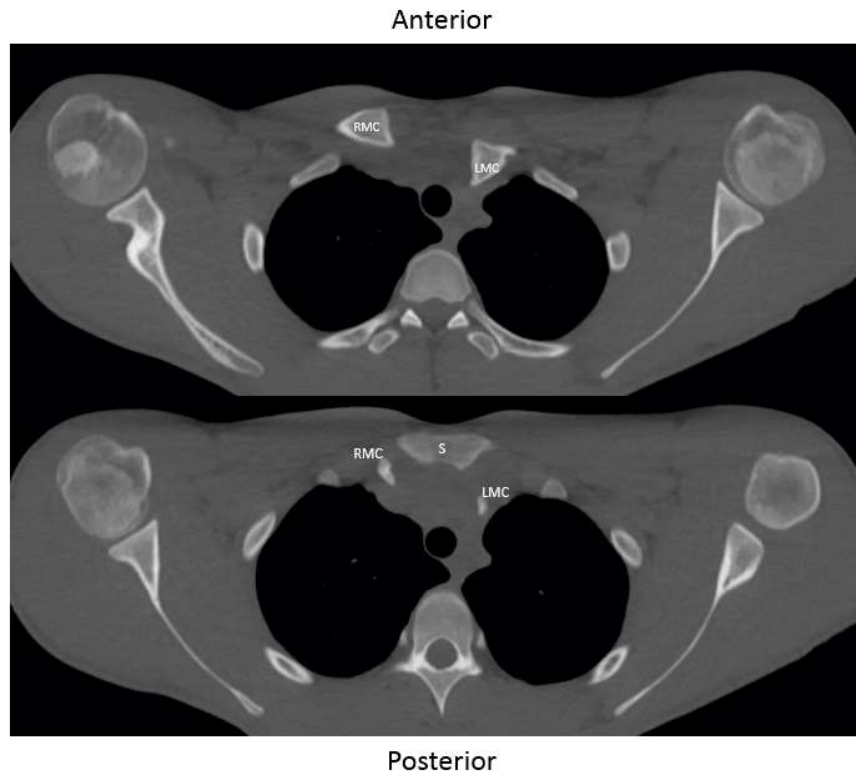


*Figure 1: Consecutive axial CT scan images of the chest demonstrating posterior displacement of the left medial clavicle relative to the sternum (S=Sternum, RMC=Right medial clavicle, LMC= Left medial clavicle).*

Anterior SC injuries can be felt as a bump near the SC joint with visible deformity. Posterior SC injuries are more subtle and can be easily missed unless clinical vigilance is maintained. Red flags that may help to identify patients requiring more urgent treatment for posterior dislocation include dyspnea, dysphagia, stridor, pulse changes from the contralateral arm, venous congestion of the arm, and paresthesias.

SC injuries can be identified with AP and serendipity (40 degree cephalic tilt) radiographs of the bilateral clavicles in order to detect subtle differences between the injured and un-injured SC joints. Often an axial CT scan will show the diagnosis more definitively, including the direction of dislocation and potential mediastinal compression.

Patients with asymptomatic anterior injuries may be treated non-operatively. Anterior SC injuries can be treated with an attempt at closed reduction, with open reduction and suture fixation reserved for patients with symptomatic instability. Patients with posterior injuries can be treated with open reduction and suture fixation of the SC joint or physeal injury.

## SUPRACONDYLAR HUMERUS FRACTURES

Supracondylar (distal humeral metaphysis) fractures of the humerus are the most common elbow injury in children. They usually occur in children between 4 and 8 years old after a fall on an outstretched hand.

Supracondylar fractures can be described as either a flexion or extension injury type, meaning that the distal humerus fragment is flexed or extended relative to the proximal humeral shaft on a lateral radiograph. Extension-type fractures are far more common.

Children with supracondylar fractures present with elbow pain and swelling; deformity and ecchymosis might be present with a more severe injury. The neurovascular exam at presentation is critical in children with supracondylar humerus injuries. The anterior interosseus nerve is most commonly injured in extension-type injuries, and the ulnar nerve is most commonly injured in flexion type injuries. The brachial artery can have decreased flow (owing to stretch or spasm) when it is tented over the distal humeral metaphysis. Examination of radial pulses and perfusion of the hand (including skin color and capillary refill) is required.

Plain radiographs are sufficient to diagnose supracondylar injuries. Occasionally, non-displaced fractures might not be easily visible. These so-called "occult fracture" can be identified by the presence of a "posterior fat pad sign." This sign is created when bleeding into the joint elevates the posterior fat pad off the bone (as seen in Figure 2). Radiographs will also demonstrate the direction and degree of displacement in the other injury types.

A line drawn down the anterior humerus can also help to determine the severity of these injuries. Typically the line will intersect the middle third of the capitellum distally, but will cross more anteriorly, or not at all, in the setting of extension type fractures (Figure 2). Fractures can be classified as non-displaced, hinged and completely displaced (separation of both the anterior and posterior cortices).
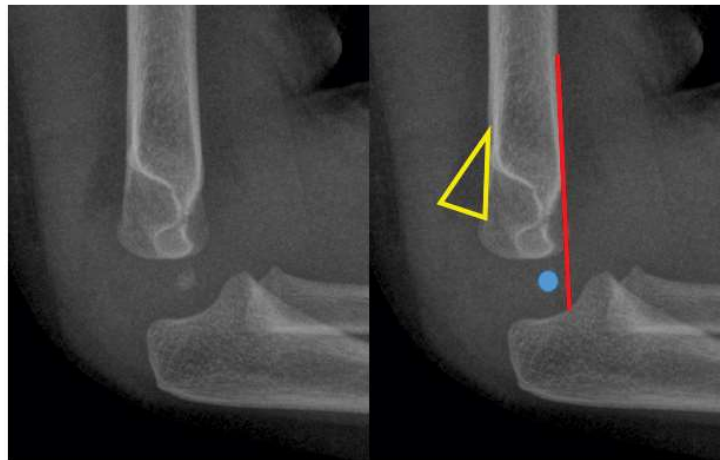


*Figure 2: Lateral radiograph of a Type 2 supracondylar humerus fracture with a sail sign (outlined in yellow on the right) indicating an elbow effusion. The anterior humeral line (in red on the right) does not intersect with the capitellum (in blue).*

The differential diagnosis that should be considered includes a distal humerus transphyseal separation, which is a variant of supracondylar fractures that occurs in very young children (neonate to 2 years of age) and is very suspicious for non-accidental trauma. Other diagnoses to consider include lateral condyle fractures, medial epicondyle fractures, and radial neck fractures (discussed below).

A red flag in children with supracondylar fractures is an ipsilateral forearm fracture, as this injury, termed a "floating elbow," may place patients at a higher risk of compartment syndrome. The three A's — anxiety, agitation, and increasing analgesia requirements — are clues of an impending compartment syndrome.

Pulseless supracondylar injuries with a warm, well perfused hand can often be treated with reduction and fixation of the fracture and close monitoring postoperatively, but patients who present with a white, pulseless hand require urgent treatment, often with operative exploration of the anterior neurovascular bundle.

Treatment of supracondylar injuries depends on the pattern of the fracture. Non-displaced injuries can be treated non-operatively with a long arm cast for three weeks. Most incomplete, hinged fractures are treated with closed reduction and percutaneous pinning, but casting might be tried. Completely displaced fractures should all be treated with closed reduction and pinning (Figure 3). Open, rather than percutaneous, pining is reserved for injuries that cannot be anatomically reduced or exploration of the neurovascular bundle is required for pulseless injuries.
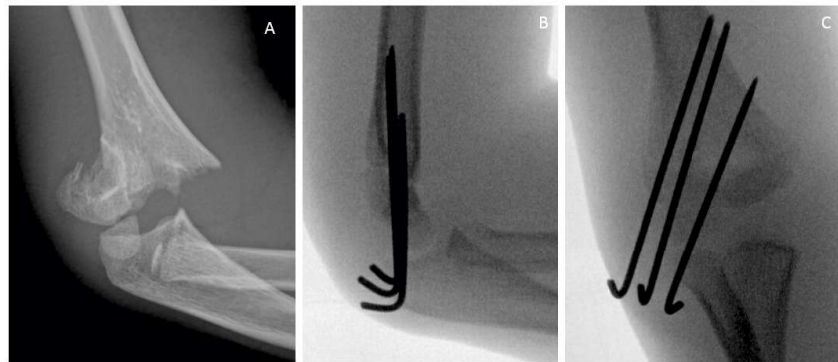


*Figure 3: Lateral radiograph at injury (A), lateral (B), and AP (C) radiographs after closed reduction and lateral percutaneous pinning of a displaced and rotated Gartland Type 3 fracture.*

Children typically regain full elbow range of motion after resuming their daily activities, and they usually do not require physical therapy. The most common complications of surgical treatment include pin migration and pin site infection. Compartment syndrome can occur as a result of the injury itself or with hyperflexion of the elbow during treatment. Compartment syndrome should be treated urgently with forearm fasciotomies in order to prevent long term ischemic contractures of the forearm and hand. Cubitus varus (an angular malalignment of the elbow, also known as a "Gunstock deformity") can occur with inadequate reduction of the fracture. It usually does not cause significant functional deficits, but can be treated with a distal humerus valgus osteotomy if severe.


## MEDIAL EPICONDYLE FRACTURES

Medial epicondyle fractures are common injuries of children typically between 9 and 14 years old. These injuries can occur in isolation but 60% are associated with elbow dislocations.

The medial epicondyle is an apophysis (a second growth center) of the distal humerus and begins to ossify between 5-7 years of age. It is the origin site of the flexor-pronator mass of the forearm, as well as the ulnar collateral ligament (the most important valgus stabilizer of the elbow). It is the last physis of the distal humerus to undergo fusion (around age 15) and is at risk of fracture during high valgus stresses to the elbow.

Patients with medial epicondyle fractures will present with medial elbow pain. It is important to obtain any history of instability or self-reduced dislocation at the time of injury, and range of motion and a complete neurovascular exam are important. AP, lateral, and oblique radiographs of the elbow are necessary to evaluate a medial epicondyle fracture, although they can often underestimate the true displacement of the fragment. In cases of elbow dislocation, it is possible for the epicondyle fragment to become trapped in the joint. The joint should be carefully evaluated on lateral radiographs to ensure that the fragment is not trapped and that the

joint is congruous (Figure 4). CT scan can help to better estimate the displacement and location of the fragment when treatment is controversial. Differential diagnoses include concurrent elbow dislocation, ulnar collateral ligament injury, and other fractures around the elbow.



*Figure 4: Lateral (A) and AP (B) radiographs and sagittal (C) CT scan images demonstrating a medial epicondyle fracture with the fragment incarcerated in the elbow joint. Intraoperative AP (D) and lateral (E) radiographs show reduction and fixation of the fragment with a screw, with restoration of elbow joint congruity.*

Chronic valgus stress to the elbow, e.g. with baseball pitching, can also lead to stress fractures or fibrous unions of the medial epicondyle physis and should be considered. Red flags include instability of the elbow and crepitus or a mechanical block during range of motion. These should alert the examiner to the possibility of a prior dislocation and/or an incarcerated fragment.

Treatment of medial epicondyle fractures has traditionally been non-operative in a long arm cast. Absolute indications for surgical treatment are open fractures and fractures with the medial epicondyle incarcerated in the joint. Citing the high valgus stresses that athletes like gymnasts and throwers place on their medial elbow, some surgeons have advocated operative treatment for this population. Surgical treatment typically consists of open reduction and screw fixation into the medial column, with careful protection of the ulnar nerve. Suture of the ulnar collateral ligament and flexor pronator insertion can augment the fixation of the medial epicondyle.

Loss of motion and bony non-union are two of the most common complications of both surgical and non-surgical treatment of medial epicondyle fractures. Patients tend to have more difficulty regaining full motion after these fractures compared to other pediatric fractures of the elbow, which is why stable fixation to allow early motion is important. While some studies have reported 15-30% non-union rates for medial epicondyle fractures, most patients obtain asymptomatic fibrous unions and do not require further treatment.

## LATERAL CONDYLE FRACTURES

Lateral condyle fractures of the elbow are intra-articular fractures that start in the lateral distal humeral metaphysis and extend into the joint. The forearm extensor muscles originate on the lateral epicondyle and tend to displace fractures posteriorly and laterally.

Lateral condyle fractures are the second most common pediatric elbow fracture. Because most of the lateral condyle fragment is cartilage and not easily visible on radiographs, these fractures are commonly missed on initial imaging. Therefore, a high index of suspicion is needed.

Patients with lateral condyle fractures of the elbow present with elbow pain after a fall, and physical exam typically reveals lateral elbow tenderness and pain with active wrist flexion and extension. AP and lateral radiographs can help to make the diagnosis, but an internal oblique radiograph of the elbow is the most critical view for both diagnosis and determining the degree of displacement. The differential diagnosis for lateral condyle fractures includes supracondylar humerus fractures and radial neck fractures, which can present similarly in pediatric patients and can be difficult to distinguish when they are non-displaced.

Treatment of lateral condyle fractures depends on the degree of displacement. Fractures that are truly non-displaced on all views can be treated non-operatively in a long arm cast for four to six weeks. Any lateral condyle with displacement is typically treated operatively because of their decreased healing potential. Minimally displaced fractures are treated with closed reduction and percutaneous lateral pinning, particularly if the articular cartilage appears to be hinged and remains intact. Closed reduction and percutaneous pinning can be supplemented with a post-reduction arthrogram in order to confirm the reduction and joint congruity (Figure 5).
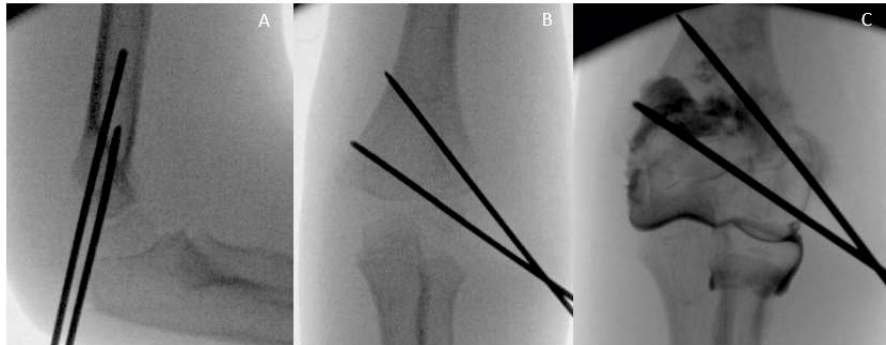


*Figure 5: Intraoperative fluoroscopy demonstrating lateral (A) and AP (B) views of a lateral condyle fracture after reduction and percutaneous pinning with two lateral pins. Figure C is the AP view after administration of intra-articular dye (arthrogram), that demonstrates the outline of the cartilaginous portion of the distal humerus.*

Significantly displaced fractures, or fractures in which an anatomic reduction cannot be obtained through closed means, are treated with open reduction through a lateral approach to the elbow and percutaneous pinning. Lateral condyle fractures are associated with complications to a greater extent than other pediatric elbow fractures. Possible complications include stiffness, delayed union and non-union, osteonecrosis and deformity. Stiffness is most common and usually resolves with time. Deformity can present as cubitus valgus with or without an ulnar nerve palsy.

## RADIAL NECK FRACTURES

Radial neck fractures tend to occur in children between 8-11 years old, slightly older than the typical supracondylar humerus fracture. Radial neck fractures can occur in isolation or in combination with other elbow injuries, such as olecranon fractures or elbow dislocations. The ossification center of the radial head begins to ossify between 3 and 5 years old, and fuses with the radial shaft between 16 and 18 years old.

Radial neck fractures often occur after a fall onto an outstretched hand, often when the elbow is extended and undergoes a valgus force. Patients present with elbow pain, tenderness over the radial head, and pain with elbow motion, particularly pronation and supination. Radial neck fractures are described by their degree of angulation relative to the radial shaft. Older children and adolescents who present with tenderness over the radial head but no fracture seen on radiographs can be assumed to have an occult radial head fracture.

The differential diagnosis includes supracondylar and lateral condyle elbow fractures, as well as a nursemaid's elbow (see below).

Red flags include signs and symptoms of compartment syndrome, which should be considered after higher energy injuries, especially those associated with other elbow injuries like elbow dislocation.

Non-displaced fractures, occult fractures and those with less than 30 degrees of angulation can also be treated non-operatively. Fractures with more than 30 degrees of angulation should be treated with an attempt at closed reduction either in the emergency department or in the operating room. When performed in the operating room, a percutaneous k-wire can also be used to help obtain and maintain the reduction. For more severely displaced fractures that cannot be reduced with closed techniques, an open reduction and percutaneous pinning can be used to fix the fractures.

Loss of pronation and/or supination is the most commonly reported complication of radial neck fractures.


## NURSEMAID'S ELBOW

Nursemaid's elbow is a common injury in very young children, and it occurs when the annular ligament subluxes and becomes trapped between the radial head and the capitellum. Most nursemaid's elbows occur in children between 2 and 3 years old; and it is very uncommon after the age of 5. Nursemaid's elbow occurs after a longitudinal force is applied to the forearm while it is pronated with the elbow in extension, like when a parent pulls on the child's hand or forearm. The child usually presents with pain and refusal to move the elbow, with the elbow held in flexion and forearm pronation.

Radiographs should be obtained in any children with elbow deformity or a presentation that is not completely consistent with nursemaid's elbow. (Radiographs are not required to make the diagnosis if the history and exam are classic.) Radiographs, if obtained, are often normal or will show a slight subluxation of the radial head.

The differential diagnosis includes supracondylar or transphyseal humerus fractures, radial neck fractures, and congenital conditions like radioulnar synostosis. Red flags include elbow deformity, unknown mechanism of injury, and children over 5 years old.

Nursemaid's elbow is easily treated with closed reduction maneuvers. The two most commonly used techniques are: 1- hyperpronation with the elbow in 90 degrees of flexion; 2- supination of the forearm followed by flexion at the elbow. A click is usually felt in the elbow when the annular ligament reduces, and children often feel immediate relief and will start to fully range the elbow again. No further immobilization or treatment is required after reduction, and children typically have good outcomes without any further sequelae related to the elbow. Recurrence of nursemaid's elbow is sometimes seen in younger children, but usually does not happen again after the age of 5.


## BOTH BONE FOREARM FRACTURES

The radius and ulna form a ring in the forearm that transmits forces from the wrist to the elbow during a fall. Both bone forearm fractures are typically described based on their location (i.e. middle third), direction of angulation, and whether the fractures are complete or incomplete (i.e. greenstick) fractures. The radius and ulna each have a proximal and distal physis, but the distal physis in both of the bones account for the vast majority of growth in the forearm. Thus, fractures closer to the distal physis have greater potential for remodeling after they heal.

Patients with a both bone forearm fracture present with forearm pain and deformity after a fall. The wrist and elbow must be examined for any findings that may signify anything more than a straightforward both bone forearm fracture. The differential diagnoses for this injury include isolated distal radius fracture, a so-called Galeazzi fracture (radial shaft fracture with associated distal radioulnar joint dislocation), or a Monteggia fracture (ulna fracture with associated radial head dislocation). The presence of elbow pain in a patient with

a forearm fracture may signify a concomitant supracondylar fracture, creating a "floating elbow" (as discussed above). Other red flags include persistently oozing wounds, which typically indicate an open fracture that requires irrigation and antibiotics.

AP and lateral radiographs of the forearm will demonstrate the fracture pattern. Additional radiographs of the elbow and wrist are necessary to rule out associated injuries of these joints.

Although both bone forearm fractures in adults are often treated surgically, both bone forearm fractures in children are often more stable because the thick periosteum usually remains intact and can usually be treated non-operatively with closed reduction and casting (Figure 6).



*Figure 6: Both bone forearm fracture AP and lateral radiographs before (A and B) and after (C and D) closed reduction and long arm cast placement.*

Surgical treatment is reserved for fractures with unacceptable alignment after reduction, as well as for both bone fractures in adolescents who have nearly finished growth.

Outcomes after treatment are all typically satisfactory, with at most a mild loss of motion (particularly pronation or supination) but no functional deficits.

# PEDIATRIC FRACTURES OF THE LOWER EXTREMITIES

## PELVIC AVULSION INJURIES

Pelvic avulsion fractures occur in children and adolescents because their muscles and tendons are stronger than the area of bone (the secondary ossification centers) where the tendons attach. These fractures occur after a strong and sudden muscle contraction, which pulls the bony insertion site off of the pelvis. This typically occurs in track athletes and soccer players, who frequently perform eccentric contractions. The most common pelvic avulsions are of the anterior superior iliac spine (ASIS) by the sartorius; of the anterior inferior iliac spine (AIIS) by the rectus femoris; and of the ischial tuberosity by the hamstrings.

Athletes with pelvic avulsion fractures will present with pain at the avulsed insertion site and pain with concentric contraction of the involved muscle or muscles. The differential diagnosis includes muscle strain and other patterns of pelvic and/or acetabular fracture. High energy trauma and intra-abdominal injury are red flags that may indicate a more complex pelvic fracture involving the pelvic ring or acetabulum. AP pelvis radiographs are usually sufficient to diagnose pelvic avulsion fractures, but additional radiographs or a CT scan can be obtained when the diagnosis is uncertain or higher energy pelvic fractures are suspected. The radiographs will reveal a separation of the insertion site (i.e. ASIS or AIIS) from the rest of the pelvis. Treatment of these injuries is almost always non-operative because the pelvis remains stable. NSAIDs and crutches for four to six weeks can be prescribed for symptomatic treatment until the fracture heals, followed by a slow return to sports. These fractures usually heal and do not result in significant long-term sequelae.

## FEMORAL SHAFT FRACTURES

Femoral shaft fractures are described based on their location and fracture pattern. Patients will present with leg pain, refusal to ambulate, or a deformity after a fall or other trauma. Physical exam should include a complete evaluation for other associated injuries, particularly in the setting of high energy trauma. Differential diagnoses include proximal femur and femoral neck fractures, hip dislocation, and distal femoral physeal fractures.

An important red flag is a young child with a femoral shaft fracture who is not yet of walking age. The risk of non-accidental trauma in these children is very high and should be considered during the workup.

Femoral shaft fractures can be diagnosed with an AP and lateral femur radiograph. Advanced imaging is usually not necessary unless the fracture extends proximally or distally into a physis.

Treatment is based primarily on age and size of the child as well as stability of the fracture pattern. Children younger than 6 months old can be treated in a soft harness. In this age group, the fracture is apt to heal without significant sequelae because of the bone's remodeling potential. Children 6 months to ~6 years old can usually be treated successfully with a hip spica cast. Many pediatric femoral shaft fractures are low energy injuries with a spiral pattern. These fractures are not at significant risk for shortening. Furthermore, because the injured bone typically overgrows the contralateral side by up to 2 cm, the mild shortening that will occur during spica cast treatment is well-tolerated (Figure 1).
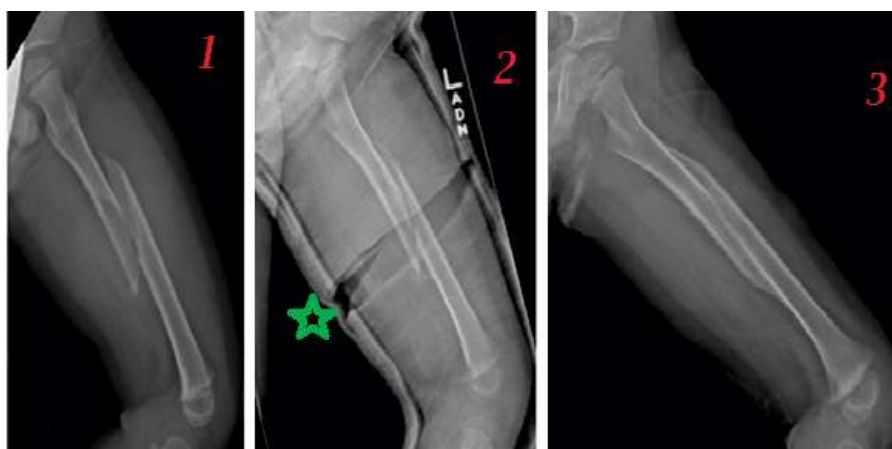
*Figure 1: Radiographs demonstrating a femoral shaft fracture at injury (panel 1), one week after initial casting (panel 2), and two months after casting with complete healing (panel 3). The green star and lines in panel 2 denote the area where the cast was wedged to better align the bone.*

Older children between 6 and 12 years old are treated based on their weight and fracture pattern. Children in this age group weighing less than 50 kg with stable fracture patterns are treated with flexible intramedullary nails. Fractures in heavier children or those with unstable fracture patterns are treated with surgical plating. Children older than 12 years of age can be treated with rigid intramedullary nails using a lateral trochanteric entry point. This surgical approach minimizes the risks of damaging the blood supply to the femoral head and of resultant avascular necrosis. Most femoral shaft fractures typically heal without future functional deficits.

## TIBIAL TUBERCLE FRACTURES

The tibial tubercle is the insertion site of the patellar tendon, an apophysis just distal to the proximal tibial epiphysis during growth. Eccentric contraction of the quadriceps, as seen during the landing from a jump, can avulse the tibial tubercle.

The clinical presentation of tibial tubercle fractures includes pain, ecchymosis, and deformity over the fracture site. Patients will often be unable to perform a straight leg raise. Fractures that extend into the knee joint will be associated with a hemarthrosis. The differential diagnosis includes proximal tibial shaft fractures, tibial spine fractures, and patellar sleeve fractures. Red flags are any signs or symptoms of lower extremity compartment syndrome.

Radiographs of the knee are used to diagnose tibial tubercle fractures; a CT scan of the knee can also be obtained for diagnosis and surgical planning when the fracture pattern is unclear.

The treatment of tibial tubercle fractures is dictated by the fracture pattern and displacement. Fractures of the tip of the tibial tubercle can be immobilized in knee extension if they are non-displaced and if the extensor mechanism remains intact. Fractures that extend between the tubercle apophysis and the proximal tibial epiphysis are usually treated surgically with open reduction and internal fixation of the tibial tubercle in order to restore/preserve the extensor mechanism. Arthrotomy or arthroscopy might be needed in some cases to ensure that the meniscus is not trapped in the fracture site (Figure 2).
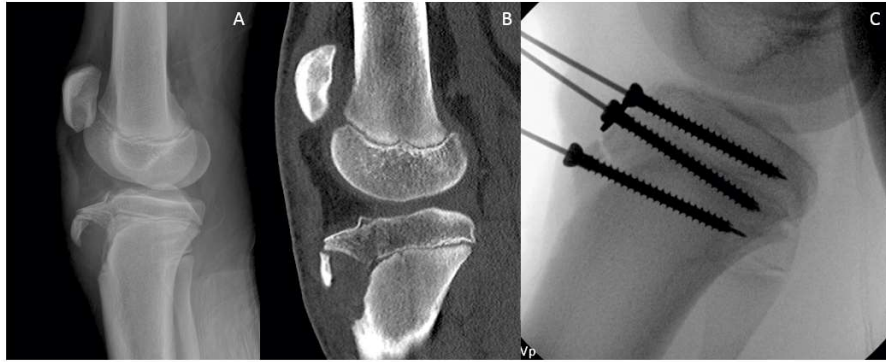
*Figure 2: Lateral radiograph (A) and sagittal CT scan (B) demonstrating a displaced tibial tubercle fracture extending through the tubercle apophysis and into the anterior epiphysis, as well as an intraoperative lateral radiograph after reduction and screw fixation of the fracture (C).*

Compartment syndrome is the most concerning short-term complication of tibial tubercle fractures; and patients should be monitored after both surgical and nonsurgical treatment of their injury. Injury and growth arrest of the tibial tubercle apophysis can also lead to a recurvatum deformity of the knee over time. This is a greater concern in younger patients, as they have more growth remaining. Younger patients should therefore be monitored with periodic radiographs. The most common complication of surgical treatment is symptomatic hardware over the tibial tubercle. This is seen because there is minimal soft tissue coverage in this area. Many patients thus elect to have their hardware removed after their fractures have healed.

## TIBIAL SPINE FRACTURES

The tibial spine is the intra-articular insertion point of the anterior cruciate ligament (ACL). Fractures of the tibial spine have long been described as an "ACL tear equivalent" condition because the mechanism of injury and resultant instability are similar. Fractures of the tibial spine are far less common than ACL tears, even in the pediatric population.

Patients with tibial spine fractures often present with knee pain and hemarthrosis after a contact or non-contact sports injury. Their exam can be limited by pain and swelling, and the differential diagnosis of a knee hemarthrosis in children is an ACL tear, osteochondral or intra-articular fracture, meniscal tear, or patellar dislocation. Red flags include extremely limited motion, which may indicate an associated bucket handle meniscus tear or large intra-articular osteochondral fragment, as well as inability to perform a straight leg raise, which may indicate a patellar sleeve or tibial tubercle fracture. The diagnosis can be made with AP and lateral radiographs of the knee, which will show displacement of the tibial spine fragment. An MRI is often performed to evaluate for additional intra-articular injuries, including meniscus tears and osteochondral fractures, which are commonly associated with tibial spine fractures. The MRI can also help to more accurately define fracture displacement, which is an important consideration in treatment decision-making.

Tibial spine fractures that are non-displaced or minimally displaced can be treated in a long leg cast with the knee in extension. If there is more than 5 mm of displacement, surgical reduction would be needed to restore tension to the ACL. Surgical treatment can be either open or arthroscopic, with either suture or screw fixation.

The most common complication of tibial spine fractures, particularly those with more severe fracture displacement or a longer operative time, is arthrofibrosis. Arthrofibrosis produces knee stiffness in both flexion and extension. Immobilization of the knee for more than four weeks is a known risk factor for arthrofibrosis and is accordingly avoided whenever possible.

## PATELLAR SLEEVE FRACTURES

A patellar sleeve fracture is an avulsion of the inferior pole of the patella. The mechanism of injury is a forceful eccentric contraction of the quadriceps. Instead of rupturing the patellar tendon, as would be seen in adults, this contraction avulses a fragment of distal patellar bone and cartilage (i.e., the "patellar sleeve").

Patients present with anterior knee pain, hemarthrosis, and inability to perform a straight leg raise (i.e., the extensor mechanism is not intact). In some cases, with wide displacement of the fragments, a defect can also be felt over the distal patella at the location of the injury.

The differential diagnosis for this injury includes tibial tubercle fractures, distal femur physeal fractures, and intra-articular injuries like osteochondral fractures and tibial spine fractures. The primary red flags for this injury are signs and symptoms of vascular injury or compartment syndrome, which may indicate the presence of another injury, such as a distal femur fracture or tibial tubercle fracture.

Patellar sleeve fractures are diagnosed with AP and lateral radiographs of the knee. These images will usually reveal at most a small bony fragment. Nonetheless, there is often a large piece of patellar cartilage attached to this bone and not apparent on radiographs as well. Patella alta and a knee effusion will also indicate the disruption of the extensor mechanism on radiographs and confirm the diagnosis (Figure 3).



*Figure 3: A lateral radiograph of the knee, demonstrating an avulsed distal patellar bone fragment is shown at left. The image at right is the same x-ray, with the cartilage and bone fragment highlighted in yellow and red, respectively. Note also the proximal migration of the rest of the patella.*

Treatment of patellar sleeve fractures is surgical, as the extensor mechanism must be restored in order to regain active knee extension. In addition, fixation of the fracture will restore the articular surface of the patella when a large chondral fragment is attached to the distal pole fragment. Potential complications of patellar sleeve fractures are residual extensor lag, quadriceps weakness, and loss of reduction and malunion or non-union of the fracture site, although these complications are uncommon.

## TRANSITIONAL ANKLE FRACTURES

When children are nearing the end of their growth, the distal tibial physis does not close all at once. Rather, the central region of the physis begins to close first. The medial region closes next, and the lateral region is last. Because the open physis is weaker than the nearby bone, common patterns of "transitional" ankle fractures occur.

The triplane fracture is Salter-Harris Type IV injury involving the epiphysis, physis, and metaphysis. In this injury, there are, as the name implies, three distinct fracture lines: one through the epiphysis in the sagittal plane, another through the open physis in the axial plane, and a third through the metaphysis in the coronal plane.

The Tillaux fracture is a fracture of the anterolateral epiphysis, or a Salter-Harris Type III injury.

Triplane and Tillaux fractures are two of the most common transitional ankle injuries, but other variants can occur.

Most injuries occur after an ankle twisting mechanism, and patients present with ankle pain, swelling, and inability or decreased ability to bear weight. Examination of the skin and a distal neurovascular exam should be performed. The differential diagnosis for transitional ankle fractures includes ankle sprain, extra-physeal fractures of the distal tibia or fibula, and syndesmotic ankle injury. Red flags include an open fracture and other severe soft tissue injury that may indicate a higher energy injury.

Transitional ankle fractures can be diagnosed with radiographs of the ankle, as well as radiographs of the knee, tibia and fibula, and foot, to rule out associated injuries. Because transitional fractures are intra-articular, a CT scan is often needed to help define the injury pattern and determine the degree of displacement (Figure 4).
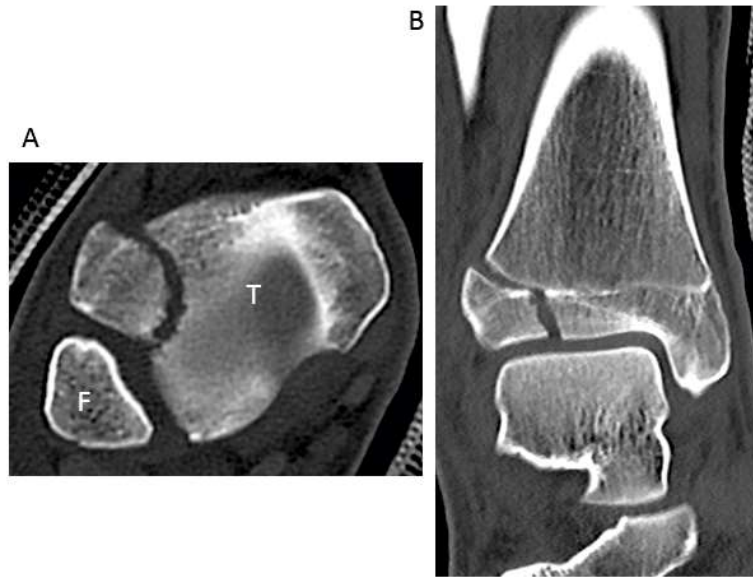


*Figure 4: Axial (A) and coronal (B) CT scan images of an ankle demonstrating a tillaux fracture injury pattern (T=tibia, F=fibula).*

Treatment is typically chosen based on the amount of fracture displacement inside of the joint itself, and the goal of treatment is to restore a congruent joint line. Non-displaced fractures can be treated with non-weightbearing in a long leg cast and transition to a short leg cast for a total of four to six weeks. Fractures with >2 mm of displacement inside of the joint are treated with open reduction and usually one to two percutaneous screws, depending on the fracture pattern, followed by a period of casting and non-weightbearing.

Physeal arrest causing a clinically significant leg length discrepancy or angular deformity is usually not a concern with these fractures. That is simply because patients who sustain these fractures are older and have minimal growth remaining.

**SKILLS**

Understand the growth center relationship to unique pediatric fractures. Be able to perform a neurovascular exam of a child with a supracondylar humerus fracture or triplane ankle fracture. Understand the differential diagnosis for a hemarthrosis of the knee in children and adolescents.

# NON-ACCIDENTAL TRAUMA / CHILD ABUSE

Non-accidental trauma or child abuse is the leading cause of childhood traumatic injury and death in the United States. It is essential that medical providers recognize non-accidental trauma in order to prevent further morbidity or even mortality. Child abuse can present in a wide spectrum of forms: physical, sexual, psychological and neglect.

According to The Child Abuse Prevention and Treatment Act and the Keeping Children and Families Safe Act, the legal definition of child abuse is defined as "any recent act or failure to act on the part of a parent or caretaker which results in death, serious physical or emotional harm, sexual abuse or exploitation, or an act or failure to act that presents an imminent risk of serious harm."

## PATIENT PRESENTATION

The clinical manifestations of child abuse can present in a wide range of pathology. Careful history, physical examination and an appropriate radiographic exam should always be performed.

The history should be detailed and thorough and note any prior injury or diseases that might be associated with bone fragility. The caregiver's description of when and how the injury occurred should be recorded. Any delays to seeking treatment must be noted. All witnesses to an injury should give their account. A patient's medical and developmental stage is important when considering the possibility of non-accidental trauma.

A full physical exam must be conducted; all clothes should be removed. The child's general appearance, weight, nutritional status, affect, and demeanor should be noted. The child's general appearance, weight, nutritional status, attained or unattained developmental milestones, affect and demeanor should be noted. The affect and demeanor of siblings are also important when considering the mechanism of injury.

The skin exam is used to identify any bruising. Bruises are the most common physical finding of abuse and if found, the size, location, shape and pattern should be recorded. Multiple bruises in various stages of healing are worrisome. Similarly, burns may be a sign of abuse. Burns and bruises in certain shapes or patterns may raise suspicion of abuse (e.g. fork, cigarette, hand, belt). Photo documentation of any bruises or burns should be obtained if possible.

The head of the patient should be carefully evaluated. Skull fractures may present as swelling, crepitus, depression, or bulging of the fontanelles. Head trauma should also be suspected if the child is obtunded or lethargic. A head CT scan and ophthalmologic exam should be performed if head trauma is suspected.

Fractures are common and are a present in about one-third of abused children. On physical exam, fractures may occult or be readily apparent. Gross deformity or angulation is rare. Swelling and bruising may be visible. On palpation, the child may grimace or cry. The limb may be held in a fixed position for comfort.

About three quarters of abused children have multiple fractures. A skeletal survey – total body x-rays – should be ordered for all children suspected of traumatic abuse.

Metaphyseal corner or chip fractures are very suspicious for child abuse and are the result of longitudinal traction and torsion of the affected limb (Figure 1). Posterior rib fractures (Figure 2) and fractures of the hand or foot in non-ambulatory infants are concerning for abuse.

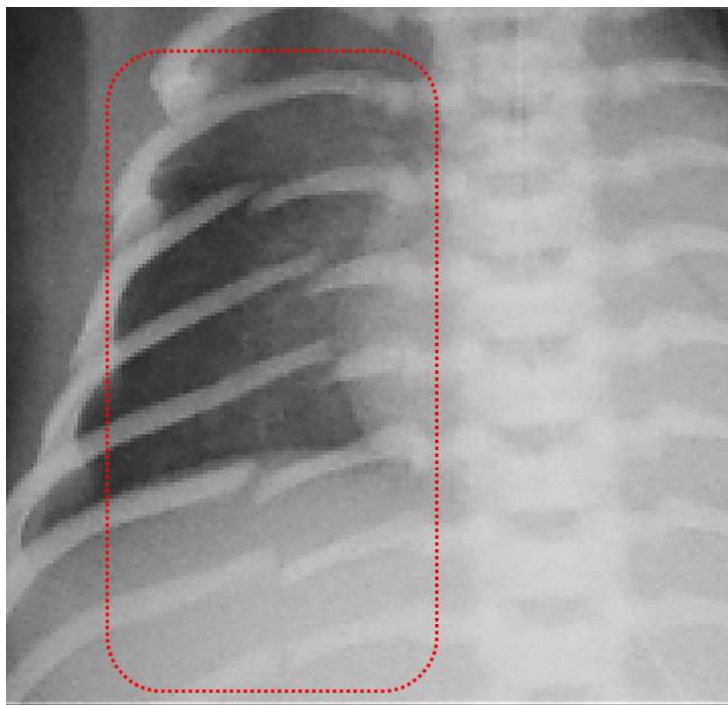*Figure 1: Metaphyseal Corner Fracture of the Tibia.*



*Figure 2: Posterior Rib Fractures.*

Femur fractures in a non-ambulatory child (Figure 3) should mandate an abuse evaluation. Fractures in various states of healing are not uncommon in children suffering from abuse.

*Figure 3: Femur fracture in a non-ambulatory infant. (Case courtesy of Dr Andrew Dixon, Radiopaedia.org, rID: 10321)*

## EPIDEMIOLOGY

Child abuse is both serious and under-recognized despite getting an abundance of attention in medical education, literature, and the news. Reports have estimated that non-accidental trauma is the second leading cause of infant death. According to the Children's Bureau in 2017, 9.1 per 1,000 children were the victims of child abuse. Often the pediatric orthopaedic surgeon may be the first physician to recognize a child suffering from non-accidental trauma.

Infants are at the greatest risk; incidence of child abuse in the first year of life is 25.3 per 1,000 children. Neglect is the most common form of abuse and physical abuse is the second, accounting for 74.9% and 18.3% of reports, respectively. In 2017, 1,720 children died of abuse/neglect at a rate of 2.32 deaths per 100,000 children.

## DIFFERENTIAL DIAGNOSIS

Other conditions that may produce "unusual" musculoskeletal findings include osteogenesis imperfecta, congenital pain insensitivity, scurvy, infantile cortical hyperostosis, birth trauma, osteopenia of prematurity, rickets, congenital syphilis, coagulation disorders (von Willebrand, hemophilia, thrombocytopenia), and leukemia.

## RED FLAGS

In all realms of diagnosis, there is usually a sensitive/specificity trade-off: that is, false negatives (missing a case) can be decreased only by increasing false positives (over-diagnosis). In the case of child abuse, the need to protect children demands that the sensitive/specificity thresholds are tilted towards higher sensitivity. Yet recognizing that high sensitivity might lead to more false positives, a two-pronged approach is needed, whereby any worrisome finding will trigger a close and detailed investigation. This investigation can then exclude the false positives.

This two-pronged approach requires a certain equanimity on the part of caregivers and physicians alike: an investigation of suspected child abuse is not to be considered as an accusation.

Elements of the history and physical that should prompt a closer look include the following:

- Vague or inconsistent history from the caregivers.
- Delayed presentation of the injury.
- Abnormal interactions between the caregiver and patient.
- A history of other injuries.
- A mechanism of injury that is insufficient to explain an injury or inconsistent with the developmental level of the child.
- A history of domestic abuse.
- Premature infants or those with low birth weight are at higher risk (as are those with chronic medical conditions).
- Metaphyseal fractures.
- Scapula fractures.
- Spinous Process fractures.
- Sternal fractures.
- Rib fractures.
- Any fracture or bruise in a non-ambulating child.
- Detection of a previously undiagnosed healing fracture.
- Bruising on the torso, ear, or neck for a child younger than four years of age.

## TREATMENT OPTIONS AND OUTCOMES

Treatment for non-accidental trauma is focused on the presenting problem, includes a thorough evaluation to avoid missed injuries, and prioritizes preventing another injury or problem. The safety of the child and children within the household is a priority.

A multi-disciplinary approach is best used. Sub-specialists should be consulted as the evaluation dictates. Social workers should be notified to help with care and the transition of care to home.

All US states have mandatory reporter laws that dictate that medical providers must report child abuse if it is suspected. The rules established by these laws differ by state. Providers should be familiar with their responsibilities under the law.

The patient should remain under medical care and only be discharged once a plan has been determined that ensures the patient's safety.

Outcomes from non-accidental trauma are largely determined by the extent of injury.

Figure 4 demonstrates the routine healing response of a femur fracture. Acceptable alignment has been maintained and abundant callus is present at 16 days after initial diagnosis.

*Figure 4: An x-ray of the femur taken approximately 16 days after a fracture. Abundant callus is present.*

It is important to mention that unrecognized child abuse can be disastrous. In one series of 54 children with head injuries caused by non-accidental trauma but not initially recognized as such, 15 were reinjured after the missed diagnosis and five children ultimately died of further abuse.

### Holistic Medicine

Care of the abused child and family should be thorough and thoughtful. A non-accusatory discussion with the patient's caregivers is essential. The history and physical should be focused on the care of the patient. Objective findings should be well documented in the Electronic Medical Record and conjecture or accusations should be avoided.

Caring for a victim of child abuse requires a multi-disciplinary team. Social workers and child protection teams should be contacted once suspicion has been raised to coordinate care. Sub-specialists in neurosurgery and ophthalmology should be consulted if there is evidence of head trauma. Good communication among team members helps to ensure quality care for the patient and family.

## RISK FACTORS AND PREVENTION

Child abuse affects all races, religions, and classes of society, but epidemiology studies have elicited that some populations are a greater risk. It is important to remember that each case must be taken on an individual basis and the absence of risk factors does not preclude abuse.

Risk factors can be categorized into three groups.

1. Risk factors intrinsic to the child.
2. Risk factors intrinsic to the perpetrator.
3. Risk factors intrinsic to family structure and society.

*(I)*

The risk of non-accidental trauma is inversely related to age with the majority of victims younger than two years of age. First born and step children are at an increased risk of abuse. In addition, any condition that makes the child more difficult to care for predisposes them to maltreatment. Examples include children with behavioral issues, chronic illness, and premature delivery.

*(II)*

Factors which are intrinsic to the perpetrator of abuse are many. Parents with limited knowledge of child development, low sense of parental competence, or inconsistent parenting may predispose a caregiver to abuse children. Having been a victim of child abuse or in an abusive relationship are predisposing factors as well.

Drug or alcohol abuse, mental illness, low income, unemployed parents, and a lack of social support increase the risk of Non-accidental trauma within the household.

*(III)*

Looking within the family and societal structure reveals many risk factors. Abuse is more common when families perceive a lack of community support and lack of connection to the community. Abuse is more common in communities of lower socioeconomic status, as well. Within the US, the rate of abuse varies amongst race and culture. According to the 2017 Child Maltreatment Report, African American children are at an increased risk of abuse. The rate of African American child fatalities due to non-accidental trauma is more than double the rate of Caucasian children and three times greater than the rate of Hispanic children.

## MISCELLANY

Here are some helpful memory aids:

- If you don't cruise, you don't bruise.
- TEN-4 (Bruises on Torso, Ears, Neck before the age of 4 raise concern).
- 3 S's: Scapula, Spinous Process & Sternal fractures.

## KEY TERMS

Non-accidental trauma, Child abuse

## SKILLS

Perform a thorough history and physical exam on infants suspected of experiencing abuse. Recognize the history and physical exam "Red Flags" of non-accidental trauma. Be able to clearly and objectively document findings in writing. Communicate clearly with family in a non-accusatory manner, focusing on care and safety of the child.

# PEDIATRIC MUSCULOSKELETAL INFECTIONS

## OSTEOMYELITIS

Osteomyelitis represents an infection of bone. In children, it occurs most commonly in the metaphysis of long bones. About two-thirds of cases occur in the lower extremity, with the femur and tibia most often affected. Osteomyelitis is subdivided based on chronicity into acute osteomyelitis (symptom duration less than two weeks), subacute osteomyelitis (symptom duration from two to six weeks), and chronic osteomyelitis (symptom duration longer than six weeks).

Osteomyelitis in children most commonly occurs due to hematogenous seeding, that is, circulating bacteria in the bloodstream land in the bone. Osteomyelitis can also occur as a result of local spread of bacteria from an adjacent infection or from direct inoculation due to an open fracture or puncture wound.

In children, the metaphyseal capillaries make a sharp hairpin turn as they approach the physis. This turn decelerates the blood flow, which can allow bacterial seeding of bone to take place.

When bacterial seeding occurs, a local immune response leads to increased vascular permeability, edema, increased vascularity, and recruitment of polymorphonuclear leukocytes (PMNs). This purulence increases pressure within the medullary canal and can further obstruct blood flow. It can also cause extrusion of purulent fluid through the bone's surface to the periosteum, resulting in a subperiosteal abscess. Increased pressure can cause venous stasis and thrombosis, leading to necrosis of bone. The necrotic and infected bone might become sequestered by new bone formation, making the eradication of bacteria difficult or impossible without surgical excision. (The new bone tissue is called an "involucrum" and the infected bone it surrounds is called the "sequestrum.") At times, an involucrum may spontaneously break down and drain purulent fluid through a sinus tract to the skin.

In children under 18 months, the metaphyseal capillaries extend across the physis to the epiphysis. This allows infection to possibly damage the physis and seed the joint causing septic arthritis (see below).

Osteomyelitis in pediatric patients is most commonly caused by Staphylococcus aureus, followed by group A beta-hemolytic Streptococcus. Haemophilus influenzae infections were previously common, however the prevalence has decreased due to widespread immunization. Group B streptococcus and Enterobacter infections occur more commonly in infants and very young children. Salmonella infections may occur in children with Sickle Cell disease. Immunocompromised patients can develop infections from atypical pathogens.

The annual incidence of acute and subacute osteomyelitis in children is about 13 per 100,000, and there is no significant difference in occurrence rates between males and females. Children with immunodeficiency, diabetes, hemoglobinopathy, and systemic inflammatory conditions are at increased risk.

In acute osteomyelitis, patients typically present with fever and progressive pain. If the lower extremity, pelvis, or spine is involved, it is common for the patient to have a limp or refuse to walk. If the upper extremity is involved, it is common for the patient to refuse to use the extremity.

On physical exam, the patient usually does not look well. The affected region is typically swollen, warm to the touch, and tender to palpation. The patient may also have limited motion of the joint adjacent to the region of pain.

In subacute and chronic osteomyelitis, patients typically present with vague discomfort, but no fever or constitutional symptoms. Their primary complaint is typically well localized pain in the metaphysis of the long

bone; however, this can also occur in the epiphysis or diaphysis. Patients typically report pain that is worse with activity and temporarily improved by rest. On physical exam, the patient typically does not look sick. The affected region is typically tender to palpation with mild swelling and possible limitations in range of motion of the joint adjacent to the region of pain. The patient may also have an antalgic gait.

Other diagnoses to consider include other musculoskeletal or soft tissue infections (e.g. septic arthritis, pyomyositis, cellulitis), inflammatory diseases (e.g. acute rheumatic fever), trauma, benign tumors (e.g. eosinophilic granuloma), and malignancies (e.g. leukemia, Ewing sarcoma, osteosarcoma).

If a patient presents with findings concerning for osteomyelitis, the following should be obtained as part of an initial work-up: a complete blood count (CBC) with differential, erythrocyte sedimentation rate (ESR or "sed rate") and C-reactive protein (CRP), blood cultures, and x-rays of the affected region.

The CRP will typically become elevated within eight hours of onset of the infection. The ESR may not become elevated until 24 to 48 hours after infection onset. Although ESR and CRP are nonspecific, they are helpful in establishing a diagnosis. CRP is also useful for monitoring response to treatment. The white blood cell (WBC) count may also be elevated; however, a normal WBC does not exclude a diagnosis of osteomyelitis.

Blood cultures are helpful as they are able to identify the infecting organism in 40 to 50% of patients and allow targeted antibiotic therapy.

X-rays tend to be normal or only demonstrate soft tissue swelling in the acute phase, however periosteal reaction, osteolysis, joint space widening, and soft tissue changes may develop. X-rays are also useful to rule out other disorders.

MRI represents the best available imaging modality for diagnosing osteomyelitis. Typical findings include bone marrow edema manifested as increased marrow intensity of T2 sequences and decreased intensity on T1 sequences. MRI may also reveal intra-osseous or subperiosteal abscesses (Figure 1).



Figure 1: MRI of a patient with distal tibia osteomyelitis and an associated subperiosteal abscess, outlined in red. (Image courtesy of Dan Miller, MD.)

Features of chronic osteomyelitis such as cloaca, sinus tracts, or sequestra are also well demonstrated on MRI. MRI is particularly useful in that it has superior soft tissue contrast and can reveal other musculoskeletal

pathology that may mimic osteomyelitis (e.g. cellulitis, pyomyositis, fracture etc.). Other modalities such as radionucleotide bone scan, CT, or ultrasound may be useful adjuvants, particularly with patients for whom MRI is contraindicated or not feasible.

If the diagnosis of acute osteomyelitis is suspected, the patient should undergo an MRI of the involved area. The presence of an intraosseous or subperiosteal abscess warrants surgical debridement.

Patients who do not require surgical debridement initially can be treated empirically with a trial of empiric intravenous antibiotics which are later tailored to the results of blood cultures. If patients fail to improve from a clinical and laboratory standpoint after 48-72 hours of empiric IV antibiotic therapy, repeat imaging and surgical debridement are indicated (Figure 2).



*Figure 2: Intraoperative fluoroscopic images demonstrating corticotomy and irrigation of the infected intraosseous space for distal tibia osteomyelitis. (Image courtesy of Dan Miller, MD.)*

Antibiotics are typically continued for four to six weeks in total assuming clinical improvement and resolution of inflammatory markers. The timing of transition from IV to oral antibiotics is controversial, however it is often feasible to make this transition after several days, provided the patient is clinically improved.

If acute osteomyelitis is not treated, it can develop into chronic osteomyelitis and cause destruction of bone as well as extension of the infection to surrounding tissues.

With appropriate treatment, the patient's clinical exam should normalize over six to twelve weeks. Skeletally immature patients with a history of osteomyelitis near the ends of long bones should be monitored long term for signs of physeal arrest. Other potential complications of osteomyelitis include venous thromboembolic disease, pathologic fracture, and avascular necrosis.

## SEPTIC ARTHRITIS

Septic arthritis is an infection of a joint. It occurs most commonly in the large joints of the lower extremities, such as the knees and hips, however it can occur in other locations as well.

Septic arthritis most commonly occurs due to hematogenous seeding of the synovium from an infection elsewhere in the body such as pneumonia, impetigo, or other skin infections. It can also occur as a result of local spread of bacteria from an adjacent osteomyelitis. This tends to happen in the setting of metaphyseal osteomyelitis when the metaphysis is intracapsular (hip, shoulder, elbow, ankle). Although much less common, penetrating wounds into the joint can also cause septic arthritis.

When spread hematogenously, bacteria travel via the bloodstream to the synovial capillaries, at which point they form microabscesses that rupture into the joint. When spread locally, bacteria in the epiphysis perforate the articular cartilage to enter the joint. When this occurs, the patient typically presents with an acute episode of septic arthritis and the osteomyelitis does not tend to become apparent for several days.

Once bacteria find their way into the joint, the acute synovial reaction results in the formation of a seropurulent exudate, ultimately leading to a painful joint effusion. During the inflammatory reaction, leukocytes release proteolytic enzymes, which can cause progressive and irreversible erosion of the articular cartilage, as well as the largely cartilaginous epiphysis. The increased intra-articular pressure can also reduce perfusion of the epiphysis, leading to avascular necrosis if left untreated. If the infection is not treated, loss of articular cartilage, joint fibrosis, bony alkalosis, bone destruction, and joint deformity can all occur.

The bacteria responsible for septic arthritis vary by the age of the patient.

In children under one month of age, the common causes are Staphylococcus aureus, Group B strep, Gram negative organisms, and Streptococcus pneumoniae.

In children between one month and three years of age, Staphylococcus aureus and Streptococcus pneumoniae are also common, but Streptococcus pyogenes, Kingella kingae, and Haemophilus influenza Type B are seen (the latter in unimmunized children in particular). These bacteria, with the exception of Kingella kingae, are seen in children older than three years of age as well.

In adolescents, the common causes are Staphylococcus aureus, Nisseria gonorrhoeae, Streptococcus pneumoniae, and Streptococcus pyogenes.

The annual incidence of septic arthritis in children in developed countries is about 4-5 per 100,000. It is more common in boys than girls, with a ratio of 2:1. Septic arthritis is not uncommon in healthy children, however children with immunodeficiency are at an increased risk.

In septic arthritis, patients typically present with acute onset guarding of a joint. Initially, pain is often poorly localized. A history of mild trauma is common (and might be coincidental); patients often have a history of a viral illness in the days to weeks prior to symptom onset.

If the lower extremity is involved, patients often have a limp or will completely refuse to bear weight on the extremity. If the upper extremity is involved, patient will often refuse to use the extremity. Patients typically also have systemic symptoms, such as malaise, fever, and poor appetite.

On physical exam, patients will often appear ill. They tend to hold the affected joint in a position to accommodate joint distention. Patients with septic arthritis of the hip tend to hold the hip in a flexed, abducted, and externally rotated position, whereas if the knee is involved, the joint is held in a slightly flexed position. Children are typically apprehensive, and resist attempts to examine the affected extremity. Any movement of the joint is typically painful. The joint is often tender to palpation. Joint effusions can be seen in subcutaneous joints such as the knee, elbow, and ankle. Effusions are often difficult to appreciate in less subcutaneous joints such as the hip, shoulder, and SI joint.

Other diagnoses to consider include transient synovitis, hemarthrosis, other infectious etiologies, inflammatory diseases, Legg-Calve-Perthes disease, and neoplastic processes. Hemarthrosis can occur secondary to

hemophilia or trauma. Infectious etiologies to consider include osteomyelitis, pyomyositis, and Lyme disease. Inflammatory diseases to consider include juvenile idiopathic arthritis, reactive arthritis, and rheumatic fever. Neoplastic processes to consider include leukemia and pigmented villonodular synovitis (PVNS).

If a patient presents with findings concerning for septic arthritis, the following should be obtained as part of an initial work-up: CBC with differential, ESR, CRP, and x-rays of the affected region. The CRP will typically become elevated within six to eight hours of the onset of symptoms. The ESR may not be elevated until 24 to 48 hours after the onset of symptoms. Although ESR and CRP are nonspecific, they are helpful in establishing a diagnosis. CRP is also useful for monitoring response to treatment. WBC may be elevated; however, it is often not elevated early on. Initial x-rays are often normal; however, they may reveal joint space widening. X-rays are also useful to rule out other disorders. Ultrasound may be helpful to confirm the presence of a joint effusion (Figure 3). Blood cultures can also be helpful and are able to identify the infecting organism in 40 to 50% of patients.
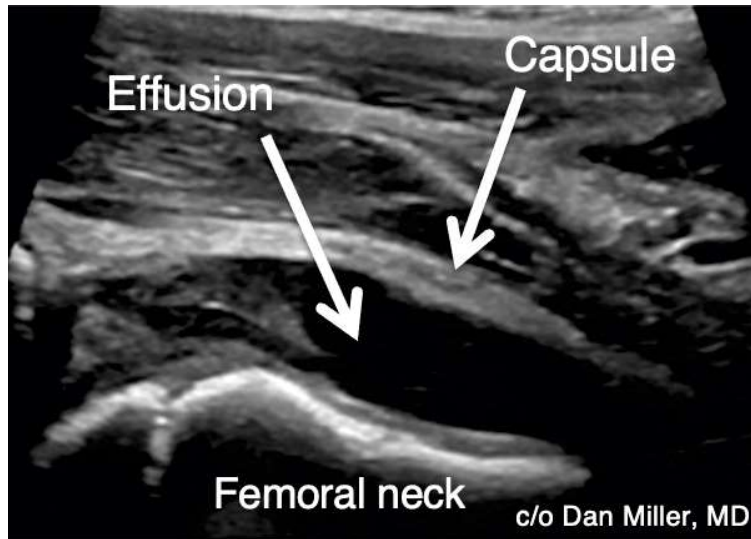


*Figure 3: Ultrasound of a patient with a large hip effusion. Note the elevation of the thickened capsule off the anterior femoral neck. (Image courtesy of Dan Miller, MD.)*

The Kocher criteria can be used to assist with diagnosis of septic arthritis of the hip. The four criteria are as follows: a history of fever over 38.5 degrees Celsius, an inability to bear weight on the affected extremity, an ESR greater than 40 mm/hr, and WBC greater than 12,000 cells/microliter. If the patient has three or four of these predictors, their predicted probability of having septic arthritis is 93.1% and 99.6% respectively. A CRP > 2.0 mg/dL is also a strong independent predictor for septic arthritis.

If the diagnosis of septic arthritis is suspected, the patient should undergo urgent aspiration of the joint and analysis of the joint fluid consisting of at minimum a cell count, gram stain, and cultures. A cell count of over 50,000 WBC/mm^3, a positive gram stain, and positive cultures are all suggestive of septic arthritis, however the absence of those findings does not exclude this diagnosis.

Once the joint is aspirated (Figure 4) and the synovial fluid is sent for cultures, the patient should be started on empiric IV antibiotic therapy. Antibiotic coverage should then be narrowed based upon the culture results. The patient should then undergo urgent surgical debridement of the joint via either open arthrotomy or arthroscopic washout. IV antibiotics are continued until the patient improves clinically, followed by oral antibiotics for another two to four weeks.

*Figure 4: Purulent material obtained from joint aspiration in a patient with septic arthritis of the knee. Due to the appearance of the aspirate, the patient underwent immediate arthrotomy with irrigation and debridement. The cell count later revealed 96,000 WBC with 90% PMNs and cultures grew Methicillin Sensitive Staph Aureus (MSSA). (Image courtesy of Dan Miller, MD.)*

If septic arthritis is not treated, the release of enzymes into the joint by leukocytes to destroy the bacteria will also destroy the articular cartilage (hence the urgent need to remove the fluid, not only for diagnosis).

The increased intra-articular pressure during the inflammatory reaction can also decrease perfusion of the epiphysis, leading to avascular necrosis. Long-term, untreated septic arthritis can lead to joint fibrosis, bony alkalosis, bone destruction, and joint deformity.

With appropriate treatment, patients generally have good outcomes. Possible complications of septic arthritis include contracture, growth disturbance, and avascular necrosis. Patients who have recovered from septic arthritis should be monitored radiographically for sequelae of infection, particularly in cases of septic arthritis of the hip.

## TRANSIENT SYNOVITIS

Transient synovitis is self-limited inflammation of the synovium. It can occur in any large joint, however it most commonly occurs in the hip.

The pathophysiology of transient synovitis is poorly understood, however there is thought to be a link between transient synovitis and antecedent viral infections. Patients often have a history of a recent upper respiratory infection.

The annual incidence of transient synovitis in children ages one to thirteen is 0.2%. It is more common in boys than girls, with a ratio of 2:1. The average age at presentation is six. Transient synovitis is the most common cause of hip pain in children.

In transient synovitis, patients typically present with joint pain and a limp. If the lower extremity is involved, patients may refuse to bear weight on the extremity. On physical exam, patients are usually afebrile and generally do not appear ill. On examination of range of motion of the affected joint, pain tends to be most severe at the extremes of motion and minimal in the middle of the range of motion arc.

Transient synovitis is a diagnosis of exclusion. If the diagnosis of transient synovitis is suspected, it is important to ensure the patient does not have septic arthritis, in which case the patient would need IV antibiotics and an urgent debridement of the joint in the operating room to prevent rapid joint destruction.

The Kocher criteria are useful in helping to differentiate between transient synovitis and septic arthritis of the hip. If the patient meets one or two of the criteria, their predicted probability of having septic arthritis drops to 3% and 40% respectively, and transient synovitis rises on the differential.

Other differential diagnoses to consider include traumatic injury, Legg-Calvé-Perthes disease, juvenile inflammatory arthritis, Lyme arthritis, osteomyelitis, and slipped capital femoral epiphysis.

If the patient presents with findings concerning for transient synovitis, the following should be obtained as part of an initial work-up: CBC with differential, ESR, CRP, and x-rays of the affected region. In transient synovitis, WBC, ESR, and CRP are usually normal, however they may be slightly elevated. X-rays tend to be normal, however they are useful to rule out other disorders. If there is any suspicion for septic arthritis, the patient should undergo joint aspiration and the synovial fluid should be sent for testing. MRI may also be useful to evaluate for osteomyelitis.

Transient synovitis is usually self-limited and spontaneously resolves in one to two weeks. Patients may be treated symptomatically with rest and anti-inflammatories. Patients should be observed closely by a primary care physician or pediatric orthopedic surgeon. If symptoms persist beyond a few weeks, a diagnosis of transient synovitis is less likely and the patient should undergo work-up for other etiologies.

Symptoms of transient synovitis generally begin to improve within 24 to 48 hours; however, it may take a few weeks for joint irritation to completely resolve. The recurrence rate of transient synovitis is as high as 20%. There are no known long-term sequelae of transient synovitis.

## LYME DISEASE

Lyme disease is an illness that affects multiple body systems. It is caused by the spirochete Borrelia burgdorferi, which is transmitted by the Ixodes tick.

Ticks are generally located on low-lying vegetation. Once they become transmitted to the host, they attach themselves to the host's skin to feed on the blood. The ticks may attach to the skin on any part of the body; however, they often attach in areas that are difficult to see such as the groin, armpits, and scalp. They must generally be attached to the skin for 36 to 48 hours before bacteria can be transmitted.

Ixodes ticks have a two-year life cycle consisting of four distinct developmental stages: egg, larva, nymph, and adult. The lifecycle begins when an adult tick lays eggs in the spring. In the summer, the eggs emerge as larva, which feed on small invertebrates such as mice and squirrels. The larvae emerge as nymphs the following spring, and also feed on small invertebrates such as mice and squirrels. The nymphs then mold into adult ticks in the fall and feed on larger animals, such as deer. Ixodes ticks acquire the Borrelia burgdorferi spirochete by feeding on infested animals during the larva, nymph, and adult stages. Mice and deer are able to carry the spirochete, however they do not become infected. Only ticks in the nymph and adult stages are able to transmit Borrelia burgdorferi. Most humans become infected through nymph bites, as the nymphs are small (less than 2 mm) and often hard to see. Adult ticks tend to be seen and removed before they can transmit the bacteria.

During an Ixodes tick bite, the tick saliva disrupts the local immune system, which creates a protective environment for spirochete replication. Replication of the spirochetes within the dermis leads to a localized post inflammatory response, which causes a bull's-eye rash termed erythema chronicum migrans. Over a period of days, the spirochetes spread via the bloodstream to the joints, nervous system, and cardiac tissue. Once present in the joint, the spirochete leads to an inflammatory response, which ultimately resulted in synovial hypertrophy and accumulation of immune complexes in the synovial fluid.

The annual incidence of Lyme Disease is 7.9 per 100,000. The incidence is higher in the northeast (Maryland to northern Massachusetts), the upper Midwest (Minnesota and Wisconsin), and the west (northern California and Oregon). Lyme disease is most common in children aged five to nine, and infection most often occurs during the summer.

Lyme disease is characterized by three distinct phases: the early localized phase, the early disseminated phase, and the late phase.

The early localized phase tends to occur within one month of the tick bite. This phase consists of the erythema chronicum migrans skin lesion (Figure 5). The skin lesion expands over a period of days to weeks, reaching a diameter of up to 20 cm. At this point, patients may have systemic symptoms similar to those of a viral syndrome, including: fatigue, fever, anorexia, headache, neck stiffness, myalgias, and arthralgias.



Figure 5: Example of the erythema migrans skin lesion associated with Lyme disease. (Case courtesy of Dr Mark Thurston, Radiopaedia.org, from the case rID: 55288)

The early disseminated phase tends to occur weeks to months after the tick bite. During this phase, pediatric patients often have multiple erythema migrans skin lesions. Conjunctivitis is also common and occurs in up to 10% of patients. Although rarer, patients may have cardiac or neurologic involvement. The most common neurologic manifestation is a Bell's palsy, or paralysis of the facial nerve. Other neurologic abnormalities that can occur include radiculopathy, cranial neuropathy, and meningitis. Cardiac manifestations that can occur include pericarditis and atrioventricular heart block.

The late phase tends to occur several months to years after the tick bite. During this phase, patients often have intermittent or persistent arthralgias of one or a few large joints, with the knee being most commonly affected. Radicular pain, distal paresthesias, and Lyme encephalopathy which leads to mild cognitive difficulties can also occur.

The differential diagnosis for Lyme disease includes acute rheumatic fever, idiopathic Bell's palsy, multiple sclerosis, peripheral neuritis, and reactive arthritis (formerly known as Reiter's syndrome).

If a patient presents with the classic erythema chronicum migrans rash, no further diagnostic testing is needed and the patient can be assumed to have Lyme disease. If a patient does not have the classic rash at the time of evaluation, however, there is concern that a patient may be in the early localized phase of Lyme disease. The following labs should be obtained as part of an initial workup: CBC with differential, ESR, CRP, LFTs, and a Lyme enzyme immunoassay or immunofluorescence assay. WBC may be elevated; however, a normal WBC does not exclude a diagnosis of Lyme disease. ESR and CRP are often elevated, albeit to a lower level when compared to values reached in the setting of septic arthritis. LFTs may demonstrate liver function abnormalities. If initial serologic tests are equivocal or positive, a Western immunoblot test should be obtained to confirm the diagnosis.

If a patient is experiencing joint pain, x-rays of the affected region should be obtained. These will often be normal; however, they are useful to rule out other disorders. Distinguishing between septic arthritis and Lyme arthritis is quite difficult but Lyme arthritis tends to have less significant reduction in joint passive range of motion and the child will often be willing to bear weight or use the extremity. If septic arthritis is on the differential, the joint should be aspirated and the synovial fluid should be sent for cell count, gram stain, and cultures. In Lyme arthritis, the synovial fluid WBC is typically elevated, but to a less significant level than in other forms of septic arthritis. It is generally very difficult to culture Borrelia burgdorferi from the synovial fluid, and serologic tests are generally sufficient to support a diagnosis of Lyme arthritis. An ECG should be obtained if Lyme carditis is suspected. A lumbar puncture should be obtained if Lyme meningitis is suspected.

In the early stages of infection, Lyme disease is treated with oral antibiotics (typically doxycycline, amoxicillin, or cefuroxime) for two to four weeks. Children under eight should not receive doxycycline. If there is no resolution in symptoms after an initial course of antibiotics, a second course may be needed. If patients have neurologic or cardiac involvement, they may need IV antibiotics (typically ceftriaxone, cefotaxime, or penicillin G). Patients with chronic Lyme arthritis that does not respond to IV antibiotics may need to undergo surgical removal of the joint synovium.

If Lyme disease is not treated in the early stage, it can progress and the infection can spread to the joints, heart, and nervous system. If Lyme disease is treated in the early stages, patients tend to recover quickly and completely without long term sequelae. Even after treatment, patients can sometimes have Post-Treatment Lyme Disease Syndrome, which can include persistent joint pain, fatigue, and cognitive impairment.

Currently, there is no available vaccine against Borrelia burgdorferi, however there are steps that can be taken to decrease the risk of Ixodes tick bites. People in high risk areas can wear long sleeved shirts tucked into pants and pants tucked into socks to decrease exposed skin. Skin and clothing can be checked for ticks once inside. Insect repellant can also be used to decrease the risk of tick bites. If ticks are found on the skin, they should be removed using tweezers. The tweezers should be used to pull traction on the tick until it releases the skin. Alcohol should then be applied to the skin.

# DEVELOPMENTAL AND SUB-ACUTE CONDITIONS

# CHAPTER 6.

# LOWER LIMB DEFORMITY

Pediatric lower limb deformity is a broad category of conditions that may be idiopathic or associated with an underlying congenital disorder. Clubfoot and other deformities of the feet are among the most common lower limb deformities and are further elaborated in separate chapters. Here, leg length discrepancy, in-toeing, and bowed legs will be reviewed as examples of longitudinal, rotational, or angular limb deformities.

## STRUCTURE AND FUNCTION

To identify abnormal, one first needs to identify normal. Although it might be reasonably expected that a person's legs should be straight and of the same length, a small leg length discrepancy or angulation can be seen in more than half of the US population.

Normal limb alignment is dependent on age. Alignment deformities can either be angular or rotational. Prior to age 2 children, have a relatively varus (bow leg) alignment in the frontal plane and this gradually becomes valgus (knock knee) as they grow. Typically, by age 7, children have a final femoral-tibial alignment of 5-6 degrees of valgus. Most rotational deformities are symmetric and considered variations of normal that typically improve with age.

Femoral rotation is defined as the angle of the femoral neck in the axial plane relative to the femoral shaft. The best way to think about femoral rotation is to consider a denuded femur resting on a flat surface. If the posterior femoral condyles are lying flat, the femoral neck will form an angle with the surface, with the head rising off the surface (Figure 1).
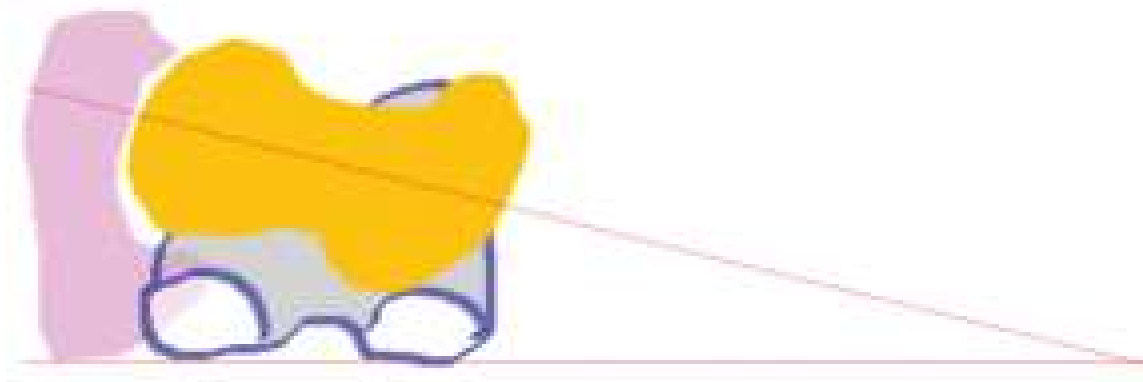


Figure 1: The normal femoral version of ~20 degree is shown. In this figure (and in figures 2 and 3) the perspective is "down the pike" of the femur of an individual lying supine on a surface. (That is, the view is given from a point perched on the ilium, looking distally, in line with the femur, toward the knee). The femoral condyles (outlined in blue) are resting on the surface; the femoral head and neck (shown in yellow) rise up off the surface to point to the acetabulum (pink).

The femur can be either anteverted (Figure 2) or retroverted.



*Figure 2: Here, excessive femoral anteversion is shown. This would tend to orient the head to the edge of the acetabulum when the feet point directly forward.*
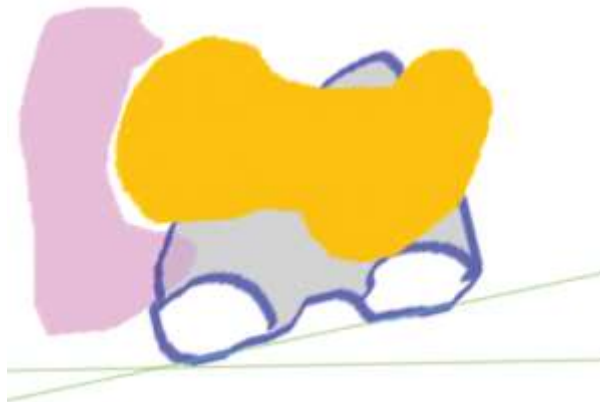


*Figure 3: Excessive femoral anteversion can be compensated by internally rotating the foot. As shown, this would lift the posterior femoral condyles off the resting plane. This is why children with femoral anteversion often walk with in-toeing.*

Femoral anteversion is more common in females, greatest in infancy with an average of 40°, and gradually decreases with age to an average of 16° at maturity. When the femur is anteverted, internal rotation of the lower extremity will tend to orient the femoral head to the center of the acetabulum (Figure 3).

Tibial rotation is defined as the inward or outward rotation of the foot as it relates to the knee. Normal intrauterine packing typically leads to internal tibial torsion that steadily progresses to slight external tibial torsion at maturity.

Although bowing at the knees is considered normal up until age 2, the angulation of the leg may be due to a deformity of the tibia itself, a condition known as "infantile tibia vara" or Blount's disease. (The term "varus" comes from a Latin word meaning "bent" but refers specifically to a bend or angulation of a bone or joint in which the distal part is more medial than it should be. In tibia vara, the foot is more medial than it would be had the tibia been straight. [When the distal part is more lateral, the deformity is denoted "valgus".]) Blount's disease can more readily be identified between the ages of 2-5 years old both clinically and radiographically. It is commonly seen in obese children, early walkers, as well as Hispanics and African Americans. Frequently it can be associated with in-toeing as well. Although much less common, children can present older than 10 years of age. These cases are denoted as adolescent Blount's disease. Adolescent Blount's disease is typically a less severe form. In addition, it is frequently associated with femoral deformity as well, and is more likely to be unilateral.

## PATIENT PRESENTATION

All patient's presenting with a concern of a lower limb deformity warrant a thorough history and physical that at minimum must include a birth history, family history, developmental milestones, gait analysis, neurological exam, limb measurement, and rotational profile.

### Leg Length Discrepancy

While the effects of leg length discrepancy can vary, most patients are asymptomatic. Parents are usually the first to report a discrepancy when they relay concerns about the way their child walks.

Children may present with a limp. Some report tiring easily with ambulation due to increased exertion required because of the discrepancy, and some complain of lower back pain.

A thorough gait analysis should be performed on every patient with suspected leg length discrepancy as well as clinical measurement of each limb.

Measurement can be performed using a tape measure from the anterior superior iliac spine to the medial malleolus in either supine or standing position or with block testing.

Block testing is performed with the patient standing using a series of blocks under the short leg until the iliac crests are level and then measuring the size of the block to determine the discrepancy.

In children who present with a leg length discrepancy at a very young age and are too young to ambulate, the physician must evaluate for a potential hip dislocation as the cause of the leg length discrepancy.

### In-Toeing

Parents complaining of their child walking or running with in-toeing (more commonly referred to as being "pigeon-toed") is an extremely common presentation in both the primary care and orthopedic setting. Most children initially present shortly after they begin walking. While most children are asymptomatic, parents often report that their child falls or trips frequently.

### Bowed Legs

Because of the possibility of Blount's disease causing the angular deformity, any toddler with bowed legs should be followed closely with serial examination. However, genu varum is not likely to suggest Blount's disease in children under the age of 18 months, unless the overall deformity is severe or there is a sharp angulation of the proximal tibia.

## OBJECTIVE EVIDENCE

### Leg Length Discrepancy

The most accurate method of assessing leg length discrepancy is full length (hip to ankle) radiographs, radiographic scanography or computerized tomography (CT).

Radiographic scanography (Figure 4) is a series of images of both legs with a ruler that are stitched together to allow measurement of the limb. CT scanography can be used similarly but additionally can assess for limb alignment and rotation. Newer imaging technologies also exist which utilize a low dose biplanar radiograph which can give accurate limb length measurements with less radiation than conventional XR (EOS Imaging). Bone age should additionally be assessed to determine the estimation of progression.

*Figure 4: A lower extremity radiographic scanogram is a series of x-rays obtained with a radio-opaque ruler in place and with the beam moved distally for each image to keep it perpendicular to the limb. The scanogram would typically include both limbs on the film. (Case courtesy of Dr Bruno Di Muzio, Radiopaedia.org, rID: 32058)*

### In-Toeing

When evaluating patients with in-toeing, radiographic evaluation is not usually required.

The exam should begin with a gait analysis. The "foot progression angle" (FPA) should be observed: this is the angle made by the long axis of the foot and the line of progression of gait ("straight ahead" as the patient walks). A negative FPA indicates in-toeing and a positive FPA out-toeing. A normal range is typically -5 degrees to +20 degrees, but can be variable with age. Once the FPA is determined to be abnormal, the next step is to localize the source of the deformity, i.e. hip, tibia, or foot.

Assessment of femoral rotation, or hip version, is performed with the patient prone on the exam table, knees bent to 90 degrees, and the legs maximally internally and externally rotated (Figure 5 Left and Center). Significantly greater internal rotation (typically greater than 70 degrees) with a decreased amount of external rotation (typically less than 20 degrees) is often indicative of increased femoral anteversion or internal rotation of the femur.

From the prone position, the thigh-foot angle (Figure 5, right) and transmalleolar axis can be examined to assess for tibial torsion. The thigh-foot angle is the angle formed by the axis bisecting the foot and the axis bisecting the thigh. The transmalleolar axis is the angle formed by axis of the malleoli of the ankle with the coronal plane of the tibia. Similar to FPA, inward rotation is designated a negative value and external rotation a positive value. Infants typically present with a negative thigh-foot angle and transmalleolar axis and progress to a more neutral or positive angle as they get older.



*Figure 5: Left) Prone hip external rotation; Center) Prone internal rotation of the hips; Right) The thigh foot angle (shown in white) is the angle defining the intersection between lines drawn down the center of the thigh (yellow) and foot (red).*

Lastly, a thorough foot exam should be performed. Patients who in-toe may have an apex or curve to their lateral foot rather than a straight lateral border, a sign indicative of possible metatarsus adductus. Additionally, this will often present with a medial plantar crease. The heel bisector line can be examined, which is the axis of the heel

exiting at the level of the toes. If the line exits through the 2nd and 3rd toe, this is considered normal, as the line exits more laterally from the toes this is a demonstration of forefoot adductus. Examiners should determine if such a deformity is rigid or flexible (passively correctable to neutral or beyond).

### Bowed Legs

True infantile and adolescent Blount's disease bowing will generally be grossly evident on exam or with ambulation.

For patients over the age of 18 months, varus alignment of the tibia on exam should prompt an x-ray. (It is generally not recommended to perform radiographs prior to this age regardless of the exam.)

The metaphyseal-diaphyseal angle of the tibia should be measured on all radiographs ordered to rule out tibia vara (Figure 6). An angle less than 10 degrees is considered normal and likely to resolve with time. Any angle greater than 10 degrees is considered abnormal and should be followed closely with serial exams and radiographs.
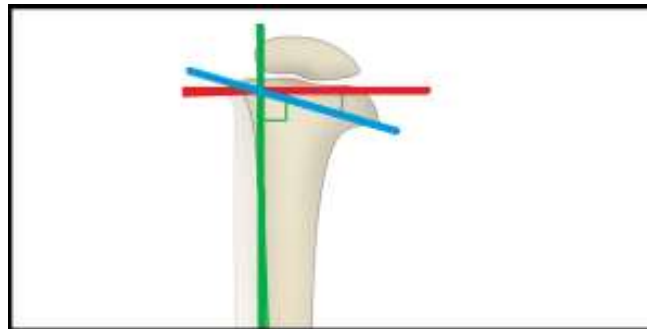


Figure 6: The metaphyseal-diaphyseal angle is a bit of a misnomer, as it is the angle between the slope of the metaphysis and a line perpendicular to the diaphysis, not the diaphysis itself. To find it, a line is drawn along the long axis of the tibia (green) and a second line drawn along the slope of the metaphysis (blue). Another line (red) is then drawn perpendicular to the diaphyseal line. The angle is defined by the intersection of this third (red) line and that of the (blue) line along the metaphyseal slope. (Diagram courtesy of Dr Matt Skalski, Radiopaedia.org, rID: 23625)

In more severe cases, metaphyseal beaking of the tibia as well as inferior and posterior sloping of the proximal epiphysis can be seen (Figure 7). Additionally, radiographic standing alignment films can be used to assess for angular deformity and determine the mechanical axis of the lower extremity.

*Figure 7: Infantile tibia vara with proximal metaphyseal beaking. (Case courtesy of Dr Alborz Jahangiri, case 47510)*

## DIFFERENTIAL DIAGNOSIS

### Leg Length Discrepancy

Leg length discrepancy can present as a result or combination of the following:

- Trauma: Long bone fractures are one of the most common causes of leg length discrepancy. A shaft fracture in a growing child can often times lead to accelerated growth in that extremity causing it to become longer than the contralateral leg. Alternatively, a fracture that heals in a shortened position or a fracture through the growth plate of a long bone can lead to a shortened extremity.

- Infection: Bone infections in pediatric patients, especially when involving the growth plate.

- Congenital disorders or dysplasias: Includes hemihypertrophy, proximal femoral focal deficiency, developmental dysplasia of the hip, fibular and tibial hemimelia, unilateral clubfoot, neurofibromatosis, multiple hereditary exostoses, idiopathic or neuromuscular scoliosis.

- Paralytic disorders: Includes cerebral palsy (due to spasticity and contractures) and polio.

- Tumor

### In-Toeing

In-toeing conditions are considered packing disorders caused by intrauterine positioning but may carry some hereditary component as they frequently run in families. They can present as a result or combination of the following:

- Metatarsus adductus: Most commonly presents in infants. Must additionally rule out skewfoot, congenital hallux varus, and clubfoot.

- Internal tibial torsion: Most commonly presents from 1-3 years of age.

- Femoral anteversion: Most commonly presents in children greater than 2 years of age.

*Bowed Legs*

Bowed legs most commonly present as physiologic tibia vara that resolves by 2 years of age. Persistent bowed legs beyond the age of 18 months are considered pathologic and can be a result of or associated with any of the following:

- Infantile tibia vara (Blount's): pathologic genu varum in children 2-5 years old

- Adolescent tibia vara (Blount's): pathologic genu varum in children greater than 10 years old. More often unilateral than infantile tibia varum which is most commonly bilateral (Figure 8).

- Congenital disorders or dysplasias: Multiple epiphyseal dysplasia, spinal epiphyseal dysplasia, focal fibrocartilaginous dysplasia, osteogenesis imperfecta, multiple hereditary exostosis, Maffucci syndrome, Ollier disease, congenital pseudarthrosis of the tibia (anterolateral bowing).

- Metabolic bone disease: Rickets (Figure 9), renal osteodystrophy.

- Acquired: Trauma, radiation, infection.



Figure 8: Adolescent tibia vara. (Case courtesy of Dr Jeremy Jones, Radiopaedia.org, From the case rID: 23622)



Figure 9: Severe bowing of the tibia (and femur) due to rickets. Rickets usually occur because of a lack of vitamin D or calcium, and results in weak bones in children. Adults can experience a similar condition, which is known as osteomalacia. (Case courtesy of Dr Angela Byrne, Radiopaedia.org, from the rID: 8116)

## TREATMENT OPTIONS AND OUTCOMES

If a congenital disorder/dysplasia or metabolic bone disease is suspected, a referral to a geneticist for formal testing might be helpful (though for some condition congenital disorders, e.g. fibula hemimelia, a genetics workup is fairly uncommon).

*Leg Length Discrepancy*

Treatment of leg length discrepancy in the pediatric patient is dependent on the cause and the projected discrepancy at skeletal maturity. If an underlying condition such as scoliosis, infection, or hip dysplasia is the cause of the leg length discrepancy, the condition should be addressed first and then the leg length discrepancy if still present.

In general, if the projected leg length discrepancy (based on future growth) is 2 cm or less, the best treatment approach is observation with shoe lifts if needed.

A projected leg length discrepancy of 2 to 5 cm has usually been treated with shortening procedures of the longer limb, such as an epiphysiodesis (i.e., surgical ablation of a physis to arrest growth).

A leg length discrepancy expected to be 5 cm to 20 cm is treated with lengthening procedures such as distraction osteogenesis. A lengthening procedure can sometimes be combined with a shortening procedure on the contralateral "long" leg.

Note that the precise cutoff at which a lengthening procedure should be used is evolving, as surgical techniques have improved. After a shared decision making process in which the family considers the risks and benefits, it may be reasonable to employ a lengthening procedure for a discrepancy of 4 cm or less.

If the projected leg length discrepancy is 20 cm or more, amputation is highly recommended.

### Bowed Legs

A majority of children with bowed legs can be treated with observation alone with spontaneous resolution expected between the ages of 2-3 years old. Mild bowing that persists beyond this age can be treated with a Knee-Ankle-Foot-Orthosis (KAFO), though this has had mixed results. More severe bowing or persistent bowing beyond the age of 4 should warrant a consideration of surgery. Surgical options include proximal tibia-fibula osteotomies, guided growth with lateral hemiepiphysiodesis, or medial epiphysiolysis (bar resection). Oftentimes these surgeries are performed in combination.

(Additional information about this rare condition is provided in this excellent review: Congenital Pseudarthrosis of the Tibia, *Journal of the American Academy of Orthopaedic Surgeons*: April 2008 – Volume 16 – Issue 4 – p 228-236)

### In-Toeing

In the vast majority of cases, in-toeing is treated non-operatively, coupled with parental reassurance and education. Most children will grow out of in-toeing as they age. These children should not have restrictions and should participate in activities similar to their peers.

In cases of rigid metatarsus adductus (that is, if the foot does not passively correct to neutral) serial casting can be performed. In even rarer cases of severe femoral anteversion or internal tibial torsion that do not improve by the age of 8 years old leading to significant cosmetic and functional disability, correctional osteotomy can be considered.

## KEY TERMS

anteversion, in-toeing, valgus, varus, retroversion

## SKILLS

Create a differential diagnosis for in-toeing. Understand clinical test to assess rotational profile of lower extremities. Know how to image for a leg length assessment. Understand the natural history of lower extremity alignment with age.

# PEDIATRIC FOOT DEFORMITIES

Pediatric foot deformities encompass a range of conditions affecting the bones, tendons, and muscles of the foot. Clubfoot, a prevalent and significant pediatric foot deformity, is discussed in its own chapter. Here, a brief review is presented of some of the other more commonly seen conditions: metatarsus adductus, tarsal coalitions, accessory navicular, planovalgus foot deformity (planovalgus), and cavas foot deformity (cavovarus).

## METATARSUS ADDUCTUS

Metatarsus adductus describes a congenital deformity in which the forefoot is turned inward relative to the hindfoot (Figure 1). Metatarsus adductus may be "flexible" (the foot can be straightened by the examiner's hand) or "nonflexible" (in which the foot cannot be straightened by hand).



*Figure 1: A drawing showing a "C"-shaped or bean-shaped foot characteristic of metatarsus adductus.*

The forefoot comprises the 5 metatarsal bones and 14 phalangeal bones. In metatarsus adductus, these bones are deviated medially. Thus, the inside border of the foot is concave, the outside or lateral border of the foot is convex, while the hindfoot remains in a relatively neutral position. The angulation is at the tarsometatarsal joint.

Metatarsus adductus is usually noted soon after birth, but can present at any age. Commonly, the parents or pediatrician of a child with metatarsus adductus will notice in-toeing incidentally. Parents may also comment regarding a wider gap between the first and second toes. This often suggests a mild flexible deformity that is observed to actively correct when the lateral border of the foot is tickled.

The exact cause of metatarsus adductus is not known, but it is thought to be a "packaging" disorder, or a result of positioning inside the uterus.

Metatarsus adductus is the most common congenital foot deformity occurring in approximately 1 of every 1000 live births, with equal frequency in males and females. Metatarsus adductus is bilateral in approximately 50% of cases.

Metatarsus adductus is defined on clinical examination by the heel bisector line. To determine this, a line is drawn on the plantar foot starting from the center of the heel directly vertical past the toes. In normal alignment, this line will exit the forefoot through the second webbed space, between the 2nd and 3rd toes (Figure 2). In metatarsus adductus, the line will exit more laterally in the forefoot. The greater the number of toes on the same side of the line as the great toe, the more severe the metatarsus adductus.

For patients presenting with in-toeing, one must also examine for and consider femoral anteversion and internal tibial torsion (see Pediatric Lower Limb Deformities). Another foot deformity called skew foot also presents with adduction of the forefoot, but unlike metatarsus adductus there are additional deformities of the midfoot in abduction and hindfoot valgus. Congenital hallux varus differs from metatarsus adductus in that the medial deviation is isolated to the great toe.

There is always full range of motion of the ankle and subtalar joint with metatarsus adductus. Stiffness in these areas should lead the examiner to consider other diagnoses.
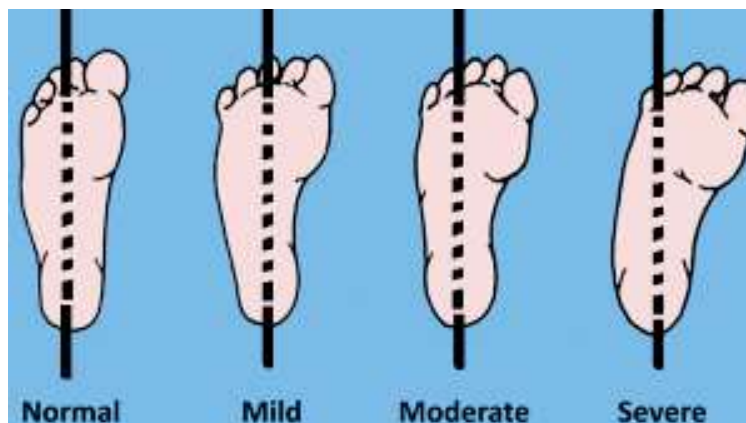


*Figure 2: The heel bisector line identifies the severity of metatarsus adductus.*

Although X-rays are not usually necessary to evaluate metatarsus adductus, they are recommended in the case of nonflexible metatarsus adductus, and in older children.

In the majority of cases, metatarsus adductus is mild and flexible. This will improve over time, up to about 4 years of age. Parents can perform stretching and stimulation of the foot.

If the metatarsus adductus is only partly flexible or rigid, serial stretching casts are sometimes necessary to achieve correction. Parents can be reassured that good results are expected with mild or moderate deformity and no functional limitations. It is rare that surgery is needed for correction of metatarsus adductus, reserved for the most severe rigid cases or cases resistant to serial casting. Soft tissue releases of the abductor hallucis and the first tarsometatarsal joint capsule followed by more casting may be indicated. If a child is older with rigid adductus osteotomies may be considered.

Metatarsus adductus is associated with other presumed packing deformities like torticollis and developmental dysplasia of the hip. It is important to examine the entire child when they present with a congenital foot deformity. Risk factors include twin pregnancy or oligohydramnios.

## TARSAL COALITION

Tarsal coalition describes the abnormal connection between two or more tarsal bones that limits range of motion and causes a rigid flat foot.

There are seven tarsal bones. These include the talus, calcaneus, navicular, cuboid, and the three cuneiforms, the medial, middle (or intermediate), lateral (see Figure 3).



*Figure 3: The tarsal bones, as seen from above.*

The abnormal connection between these bones may be composed of bone, fibrous tissue or cartilaginous tissue. This occurs embryologically due to failure of segmentation. Any abnormal connection between the tarsal bones can result in decreased range of motion of the subtalar joint, thereby limiting inversion and eversion of the heel.

Clinically, a tarsal coalition may present as a rigid flat foot. Patients usually present with a chief complaint of pain over the sinus tarsi, an aching pain along their arch, or pain under their medial malleolus that is worse with activity and alleviated with rest. The most common age for presentation of tarsal coalition is around 8 to 16 years, typically becoming symptomatic in adolescence. Some children may present with the complaint of recurrent ankle sprains or progressive flat foot.

On physical exam, patients will often have a rigid subtalar joint. This is best assessed with the foot held in neutral dorsiflexion and then applying an inversion and eversion force to evaluate subtalar motion. With standing toe rise there is no inversion of the hindfoot due to limited subtalar motion. Those with calcaneonavicular coalitions often have more preserved motion since the two connected bones do not cross the subtalar joint.

Imaging studies are required for diagnosis of a tarsal coalition. AP, lateral, oblique, and Harris axial views should be obtained. The Harris view is obtained by having the patient stand on the cassette with the x-ray beam angled between 35 and 45 degrees and will detect any coalition between the talus and calcaneus.

A calcaneonavicular coalition is best seen on an oblique x-ray of the foot (Figure 4). It may also be noticed on a standing lateral x-ray of the foot, seen as an elongated anterior process of the calcaneus. This is known as the "ant-eater sign." A talocalcaneal coalition may also be appreciated on the lateral radiograph by a finding known as the "C-sign." This is a "C" shaped line that is formed by the dome of the talus and the sustentaculum tali of the calcaneus (Figure 4). However, it is important to note that these findings have a low sensitivity, and their absence does not rule out the presence of a tarsal coalition.

The best imaging technique to evaluate for tarsal coalition is a CT scan. The CT can also aide in surgical planning and is used to look for other coalitions if surgery is being considered. If the history and physical exam are consistent with a coalition but the CT scan is non-diagnostic an MRI may reveal a fibrous coalition.



Figure 4: Calcaneonavicular coalition "ant-eater sign" seen on oblique foot x-ray (arrow).



Figure 5: Subtalar tarsal coalition "C-Sign."

Most commonly, tarsal coalitions are seen between the talus and calcaneus, and between the calcaneus and the navicular bones. These two combinations account for about 90 percent of all tarsal coalitions. Other described locations are between the talus and navicular, the calcaneus and cuboid, the navicular and cuneiform, and between a cuneiform and metatarsal, however these are uncommon. In about half of cases a tarsal coalition will be present bilaterally. In the case of rigid flat foot, it is also important to consider other causes such as juvenile idiopathic arthritis, septic arthritis, osteomyelitis, and other bone lesions for example an osteoid osteoma.

Any constitutional symptoms should prompt investigation into one of the differential causes of a rigid flatfoot. Pain worse in the morning rather than after activities may indicate a rheumatologic cause. There are syndromes associated with tarsal coalitions such as Apert and Crouzon syndrome and any dysmorphic features should be noted.

Up to 25 percent of tarsal coalitions are thought to become symptomatic, and only those that cause symptoms should be treated. First line treatment includes symptom management with activity modification, orthoses, and anti-inflammatories as needed. Second line treatment is to try cast immobilization for a period of about 4-6 weeks.

If non-operative treatment fails to provide long-term pain relief, surgery can be considered. Prior to surgery, a CT scan should be obtained to evaluate for the presence of a second coalition. Surgery begins with resection of the coalition with placement of interposed tissue (muscle or fat) to help prevent its reformation. If there is a significant foot deformity associated with the coalition, consideration can also be given to osteotomies for deformity correction. In cases where there is degeneration of the joint involved in the coalition, or if a subtalar coalition is particularly large, a fusion of the joint is considered; resection in these cases may lead to poorer outcomes.

There are no preventative measures for tarsal coalition. It is associated with certain other congenital abnormalities like fibular hemimelia, and syndromic associations.

## ACCESSORY NAVICULAR

An accessory navicular is a prominence of the navicular on the plantar medial surface and is considered a normal variant (Figure 6).



*Figure 6: A normal and an accessory navicular (NB: x-ray of an adult foot). (Image courtesy of FootEducation.com)*

The navicular bone sits along the medial aspect of the foot and articulates with the talus, the cuneiforms, the cuboid, and the calcaneus. An accessory navicular is clinically defined as an enlargement of the navicular bone along its plantar medial surface. This enlargement can be a separate piece of bone connected to the native navicular body by fibrous or cartilaginous tissue, or it can simply be an enlargement of the native bone beyond its normal size (as the "extra" part of the navicular can be completely ossified to the true navicular.)

Patients with a symptomatic accessory navicular often present with pain and tenderness over the medial palpable bony prominence. They may also have a flat foot, however no cause and effect relationship between an accessory navicular and a flat foot has been proven. Often the pain is aggravated by tight shoes that press against the medial prominence.

Plain radiographs are useful in diagnosing an accessory navicular as they can often be seen on standing AP and lateral views. A lateral oblique x-ray of the foot may also be useful to visualize an accessory navicular (Figure 7).

*Figure 7: Accessory navicular visualized on an oblique x-ray.*

This is the most common accessory tarsal bone seen in the foot with a prevalence of approximately 10%. When present, accessory navicular bones are often bilateral.

A pain and tenderness at the navicular insertion may be from tendinitis of the tibialis posterior tendon (with or without an accessory navicular). A bipartite navicular can also be seen as a separate osseous navicular structure; it is distinguished from an accessory navicular by the dorsal displacement of the separate fragment and maintained articulation with the talus. Less commonly a fracture or stress fracture of the navicular may be on the differential. An avulsion fracture of the accessory navicular can also occur with an eversion mechanism; the clinical exam and imaging should be scrutinized for acute changes in the cases of trauma with focal findings. Plantar fasciitis can also present with pain along the medial foot.

If the patient has a rigid flatfoot consider alternative diagnoses such as the presence of a concomitant tarsal coalition or underlying inflammatory arthropathy.

Patients who present with a painful accessory navicular are first treated with non-operative measures. First line measures include activity modification and shoe wear modification. If there is a planovalgus foot deformity concomitantly, a valgus correcting orthotic may help relieve pressure over the area. If this fails to relieve symptoms, rigid immobilization in a short leg cast can be done for a period of 4-6 weeks followed by gradual return to activities.

If extensive non-operative measures fail to provide long-term relief, surgical excision may be considered. Surgical options consist of removal of any ossicle that may be present along with the bony prominence of the main body of the navicular bone. The ossicle can sometimes exist within the substance of the posterior tibialis tendon, which must be taken into consideration when performing surgical excision. Others have described removal of the ossicle and advancement or re-routing of the tibialis posterior. While good results are reported with either approach, it should be noted that this is based on small, low level of evidence studies.

## PLANOVALGUS FOOT DEFORMITY

Pes planovalgus is defined as excessive valgus alignment of the heel (hindfoot) with loss or flattening of the medial longitudinal arch of the foot. It is commonly known as "flatfoot," (see Figure 8). In this condition, the forefoot is abducted outward but often rotated inward, or supinated, in relation to the hindfoot. A planovalgus foot is also called a flatfoot. This may be a physiologic variant or due to underlying pathology. The focus here will be on flexible pes planovalgus flatfoot.



*Figure 8: Clinical photo of flexible flatfoot. (Image courtesy of FootEducation.com)*

A flat foot is a combination of foot deformities that includes a valgus hindfoot and an abducted and supinated forefoot. In pes planovalgus there is flattening of the medial longitudinal arch of the foot along with the excessive hindfoot valgus. The normal alignment of the hindfoot can be up to 5 degrees of valgus. In pes planovalgus the forefoot is abducted and supinated in relation to the hindfoot. Young children develop a medial longitudinal arch over time and this flattening can improve.

Patients will present with loss of the medial longitudinal arch and a valgus hindfoot. In order to assess the degree of forefoot supination, the hindfoot must be corrected to a neutral position. If both deformities can be corrected to a neutral position, the flat foot is said to be flexible.

Another way to assess for flexibility is to have the child stand on their toes. If when standing on their toes the medial arch re-constitutes and the heel inverts, the deformity is said to be flexible. A flexible flat foot is caused by an equinus contracture due to a gastrocnemius muscle contracture and/or a tight Achilles tendon. To assess for the degree of gastrocnemius or Achilles tightness, the hindfoot must again be corrected to a neutral position. One may then perform a Silfverskiöld test to further assess where the tightness is originating. This test is performed by comparing the amount of maximal ankle dorsiflexion with the knee flexed versus extended. If more ankle dorsiflexion was achieved with the knee flexed than with the knee extended, the primary tightness is from the gastrocnemius muscle. If dorsiflexion is limited in both knee extension and knee flexion, the contracture is of the Achilles tendon itself also called the heel cord. This is important when considering surgical intervention.

Imaging studies are usually not needed for flexible flat foot diagnosis, but may be required if other etiology is suspected. All x-rays should be obtained while the patient is bearing weight. The lateral radiograph can be helpful to quantify the deformity using the talus-first metatarsal angle, also known as the Meary angle (Figure 9). This is measured by placing a line along the longitudinal axis of the talus and a second line along the longitudinal axis of the first metatarsal. The angle formed by the intersection of these two lines should be near zero in a normal foot.
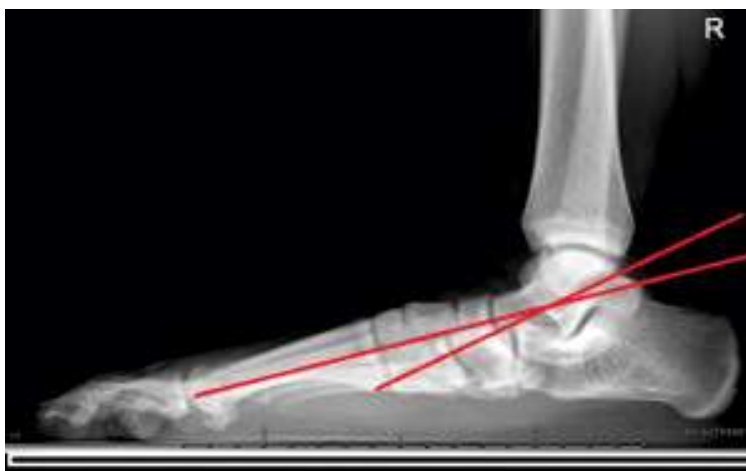
*Figure 9: Pes Planus on lateral standing x-ray. Meary's angle is the angle between a line drawn from the centers of longitudinal axes of the talus and the first metatarsal. In the normal foot, Meary's angle is roughly zero: the two lines are parallel.*

The incidence of flexible flat foot is not known, likely due to the fact that this is considered within natural physiologic variation. It is common in infants and children, while up to 20% of adults have a flexible flatfoot. A physiologic flat arch in a young child that is asymptomatic is likely to improve over time without intervention.

Underlying disorders that can be associated with a flatfoot include: accessory navicular, ligamentous laxity, neurologic or neuromuscular disorders, and obesity.

If a patient presents with a rigid flatfoot deformity the possibility of other diagnoses must be considered. These can include, but are not limited to: tarsal coalition, spastic peroneal flatfoot, inflammatory arthropathy, previous trauma, and congenital vertical talus.

If the patient has a rigid flatfoot you should consider alternate diagnoses such as a tarsal coalition or underlying inflammatory arthropathy.

Flexible flat foot may also be thought of as a normal anatomic variation and not a disabling deformity. No treatment is needed for asymptomatic flat feet. If children or adolescents are symptomatic, over the counter shoe inserts may help relieve symptoms but will not change the shape of the foot. If the child has a tight Achilles or gastroc-soleus-complex, an Achilles stretching program should be initiated. Surgery is rarely indicated in the treatment of flexible flat foot. When performed it may consist of tightening of the soft tissues medially, tendon lengthening and transfer, and osteotomies to lengthen the lateral column of the foot.

In the setting of primary tightness of the gastrocnemius muscle (as found with a positive Silfverskiöld test), a gastrocnemius recession can be performed to selectively lengthen the gastrocnemius muscle. If the Silfverskiöld test is negative, but passive dorsiflexion is limited, a tendoachilles lengthening is performed.


## CAVUS FOOT DEFORMITY

A cavus foot is one with a high arch in the midfoot due to pronation of the forefoot on the hindfoot. The hindfoot can be in neutral (isolated cavus) but is often in varus (cavovarus). In most cases, a cavovarus foot is secondary to an underlying neurologic disorder which causes a muscle imbalance. Cavus may also be a result of residual clubfoot deformity. Cavus may rarely result from trauma such as compartment syndrome, sciatic nerve injury, or tendon lacerations.

A cavus foot is the result of muscle imbalance with weak intrinsic muscles that become contracted. Cavovarus foot deformity is combination of the high arch with forefoot pronation and inversion or varus of the hindfoot.

The midfoot may be adducted or neutral. The ankle may be plantarflexed, neutral or in dorsiflexion. There is often concomitant external tibial torsion. Another type of cavus foot deformity is calcaneovarus where the entire arch is elevated off the ground from medial to lateral.

Often, patients will present with a complaint of recurrent ankle sprains or ankle instability. This instability can be the result of actual muscle weakness, loss of sensation over the foot, or from the deformity itself. It is also common for a callus to form along the lateral base of the fifth metatarsal due to the patient walking on the lateral border of their foot. It is vital to obtain a detailed neurologic exam as well as to obtain a detailed family history in these patients to evaluate for an underlying cause. Patients may have asymmetric muscle bulk or a leg length discrepancy. An examination of the spine is important to look for signs of spinal dysraphism.

To evaluate for the deformity itself, providers can look for what is called the "Peek-a-Boo" sign. This sign occurs when the medial aspect of the heel can be seen while looking from straight on, as the patient is standing (Figure 10).



*Figure 10: A clinical photo of the "Peek-a-boo sign." (Image courtesy of FootEducation.com)*

An assessment of both the hindfoot and the forefoot are important to ascertain the flexibility of each of these segments. The Coleman block test can be used to assess the flexibility of the hindfoot by placing a block under the lateral foot and metatarsals and allowing the medial forefoot to hang free (see Figure 11). If the varus position of the hindfoot corrects it is flexible.



*Figure 11: The Coleman block test is used to determine if the hindfoot varus deformity is flexible or fixed/rigid. As shown in image 1a, there is hindfoot varus of the right foot. By having the patient stand on the block such that only the lateral aspect of the foot is supported, 1b, hindfoot corrects to neutral, indicating a flexible deformity. The Coleman block test counteracts the plantar flexed first ray allowing the hindfoot to return to a normal alignment – if the hindfoot is still flexible. (Image courtesy of http://www.labome.org/research/ The-dynamic-coleman-block-test-a-novel-examination-technique-for-cavo-varus-feet. html)*

Upon presentation, plain radiographs are often obtained. A cavus foot will have an increased calcaneal pitch (>30 degrees), which is determined by the angle between the long axis of the calcaneus and a line parallel to the bottom of the foot. The Meary angle can also indicate a cavus foot if it angles upward with a magnitude of more than 4 degrees (Figure 12).
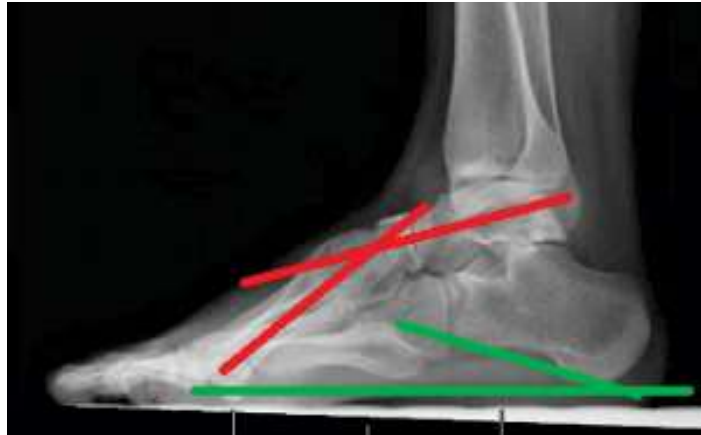


Figure 12: Pes Cavus on lateral standing x-ray. Meary's angle (red) and the calcaneal pitch angle, between the long axis of the calcaneus and the bottom of the foot (green), are shown.

An AP Pelvis radiograph should be obtained to fully evaluate the patient with a cavovarus foot, as one cause of the foot deformity, Charcot-Marie-Tooth, is associated with hip dysplasia. Likewise, spinal radiographs should be obtained to exclude scoliosis in patients with a cavovarus foot.

Cavus foot deformities are associated with neuromuscular disorders. The most common neuromuscular disorder causing a cavus foot deformity is Charcot-Marie-Tooth (CMT) disease. CMT is the most common hereditary motor sensory neuropathy. In CMT, the peroneus longus is the last muscle to atrophy and weaken. Unopposed pull of this muscle (ie, while it is relatively strong and other muscles are weak) will cause hindfoot varus and a cavus deformity to the arch. Rarely no underlying etiology can be identified for a cavus foot.

There are several causes of cavus foot deformities with some being more common in bilateral deformities. These can include: CMT disease, Friedrich's ataxia, spinal muscular atrophy (SMA), myelomeningocele, cerebral palsy (CP), and recurrent clubfoot. A unilateral cavus foot deformity can have the same causes, but should also consider previous trauma, peripheral nerve injury, poliomyelitis, tendon injury, or prior compartment syndrome.

A detailed evaluation of strength, sensation, reflexes, and vascularity is required. A new or progressive cavovarus foot deformity should raise concern for underlying neurologic abnormalities and a referral to neurology and advanced imaging of the spine and brain with MRI is indicated. A cavovarus foot should be considered a manifestation of an underlying neuromuscular disorder until proven otherwise.

The first step in treatment of a cavus foot deformity is to diagnose and treat any potential underlying cause (i.e. neuromuscular disorder). Non-operative treatment options for a cavus foot include arch supports and shoe modifications. However, many severe cavus foot deformities in children do require surgery in the long term as the deformity is often progressive.

Indications for surgery are progressive deformity, pain, pressure injuries, and gait instability. When deciding on surgical management, it is important to understand how rigid or flexible the deformity is and where. The hindfoot and forefoot are the important considerations and one may be flexible while the other stiff. This can be accomplished by using the Coleman block test as described. This test helps the examiner determine whether the deformity is forefoot or hindfoot driven, and what aspects of the foot need to be addressed during surgical reconstruction. There is not one surgical procedure for the management of a cavovarus foot, but rather there are many procedures that may be performed to address whatever aspects of the patient's foot are contributing to the deformity.

Surgical treatment can be divided into these broad categories: soft tissue, osteotomy, arthrodesis. Contractures should be released and the soft tissues should be rebalanced in all cases. In flexible feet, this may be all the surgery that is required. In more advanced cases with stiffness or rigidity, osteotomies to correct the adaptive boney changes are required. Finally, in cases of advanced rigid deformities, older patients may consider fusion as a last resort.

The biggest risk factor for a cavus foot deformity is an underlying neuromuscular disorder.

## MISCELLANY

Metatarsus adductus has also been called "bean foot" due to the shape.

The navicular is known as the scaphoid of the foot. Both names refer to the "boat shape" of the bone (sharing roots with the word "navy" and "skiff").

## KEY TERMS

Cavus, cavovarus, metatarsus adductus, flatfoot, hindfoot, forefoot, Silfverskiöld test, tarsal coalition, rigid flatfoot, subtalar joint, calcaneonavicular joint, navicular, tarsal bones, tibialis posterior

## SKILLS

Recognize the normal shape of the foot. Distinguish relationships between the forefoot, midfoot, hindfoot. Distinguish a flexible from a rigid flat foot. Be able to identify a cavus foot deformity. Understanding the "peek-a-boo" sign. Understand how to perform and interpret the Coleman block test. Recognize that there may be many normal anatomical variants in the foot with bony structure.

# CLUBFOOT

Talipes equinovarus, commonly known as "clubfoot," is a congenital deformity of the foot (Figure 1). The condition is characterized by an exaggerated arch (cavus), a convex curved outer border of the foot (adductus), inversion of the heel (varus) and plantar flexion (equinus) that may involve one or both feet. Taken together, these deformities cause the foot to resemble a club, hence the name. Clubfoot is often idiopathic and seen as an isolated birth defect, but it can also be caused by an underlying congenital disorder in approximately 20% of cases.



*Figure 1: Bilateral clubfoot in a newborn with characteristic equinus and varus deformities. (Courtesy of Steve Richards MD, Texas Scottish Rite Hospital)*

## STRUCTURE AND FUNCTION

The foot can be divided into three regions: the hindfoot, the midfoot, and the forefoot.

The hindfoot consists of the talus and the calcaneus. These bones are joined at the subtalar joint to allow for inversion and eversion. The talus also articulates with the tibia and fibula at the ankle, to allow for dorsiflexion and plantarflexion.

The bones of the midfoot are the navicular, the cuboid, and the three cuneiform bones. The midfoot forms the arch of the foot and serves as a shock absorber.

The forefoot consists of the metatarsals and phalanges.

Clubfoot deformity primarily affects the hindfoot and midfoot. Pathological features include an abnormally small calcaneus, talus, and navicular – though all of the bones in a clubfoot are slightly smaller than normal – and contracted ligaments between the hindfoot and midfoot. There is, accordingly, a plantarflexion deformity of the ankle (talocrural) joint, medial subluxation of the talonavicular and calcaneocuboid joints and inversion and adduction of the calcaneus, navicular, and cuboid.

The deformity can extend distally to the forefoot where there can be plantar flexion of the 1st metatarsal (cavus) and adduction of all metatarsals. The deformity can also extend proximally to the calf, with atrophy, fibrosis and shortening of the muscle-tendon units of the posteromedial leg muscles seen.

There are many theories about the etiology of clubfoot, but the definitive cause is still unknown. In the past, experts believed the positioning of the foot in the womb caused the deformity; today it is known that clubfoot is associated with multiple genetic abnormalities that influence the muscle contractile complex and bone development. For example, Gurnett et al (PMID: 18950742) found that abnormalities of the PITX gene, responsible for early limb development, has been associated with familial clubfoot.

## PATIENT PRESENTATION

Clubfoot is a congenital deformity that is immediately apparent at birth. Fetal ultrasound can diagnose clubfoot as early as the second trimester.

The affected foot is characteristically adducted ("varus"), plantarflexed ("equinus"), and possesses an exaggerated arch ("cavus"). Depending on the severity, the foot may be more or less rigid. Half of all cases will involve both feet. In unilateral clubfoot, the involved foot, calf, and leg will be smaller and shorter than the unaffected side. Even after correction of the deformity, the foot, calf, and leg may have some residual differences including atrophied calf muscles, a shorter leg, and a smaller foot. These residuals are not necessarily clinically significant.

Despite its dramatic deformities, clubfoot is not necessarily painful. However, if children begin to walk prior to successful correction of the deformity, they will bear weight on the dorsolateral aspect of the foot. This abnormal gait can cause focal loading on a small area and can be painful.

## OBJECTIVE EVIDENCE

Clubfoot is detectable via prenatal ultrasound in the second trimester. Early detection is important because it can prompt discussion with parents about treatment options and early screening for underlying neuromuscular diseases.

X-rays are not particularly useful in clubfoot evaluation because the neonatal bones are immature and poorly ossified. Radiographs are more useful for measuring progress in clubfoot treatment and long-term follow-up.

## EPIDEMIOLOGY

Clubfoot occurs in 1 in 1000 births and affects males twice as frequently as females. Approximately 50% of all cases are bilateral and 25% have a positive family history of clubfoot. In the US, incidence ranges across ethnic groups from 0.4 in 1000 in the Chinese population to 7 in 1000 in the Polynesian population.

Most cases are idiopathic but about 20% are due to a genetic or chromosomal abnormality. The most common of these are disorders of the nervous system including myelomeningocele and arthrogryposis. These cases tend to be stiffer and more resistant to standard treatment than idiopathic cases.

## DIFFERENTIAL DIAGNOSIS

Metatarsus adductus is a congenital foot deformity that is superficially similar to clubfoot. Metatarsus adductus is characterized by the forefoot (metatarsus) pointing inward (adductus) with normal positioning and mobility

of the hindfoot, forming a "C" shape. The incidence of metatarsus adductus is approximately the same as clubfoot. Examination of the hindfoot can distinguish metatarsus from clubfoot, since the hindfoot has normal mobilitiy in metatarsus adductus but cannot dorsiflex or evert normally in clubfoot.

Other congenital foot deformities include talipes calcaneovalgus (dorsiflexed and abducted foot) and vertical talus (rigid foot deformity similar to talipes calcaneovalgus). These deformities are structurally and visually distinct from clubfoot.

Clubfoot may be seen in association with spina bifida, arthrogryposis, and amniotic band syndrome. The evaluator should therefore pay close attention to the spine and motor function of the extremities.

Positional clubfoot is similar to clubfoot in that the foot is in equinus and varus. It is caused by a restrictive uterine environment that forces the baby's feet into an abnormal position. However, the foot and bony anatomy are completely normal in positional clubfoot, and the deformities usually correct without treatment.

## RED FLAGS

Although clubfoot is most often an idiopathic birth defect, some cases are secondary to underlying neuromuscular conditions such as spina bifida. Thus, the presence of clubfoot should prompt a close diagnostic evaluation to exclude these conditions.

## TREATMENT OPTIONS AND OUTCOMES

Treatment options for clubfoot include serial casting, bracing, physical therapy, and surgery.

The standard of care for uncomplicated clubfoot deformities is the Ponseti Method, which involves repeated manipulation and casting (Figure 2) to guide the growth of the foot toward normal alignment. The child's foot is manually stretched toward the correct position, and a cast is then applied to maintain the correction. This process is repeated weekly over the course of 4-6 weeks.



Figure 2: Clubfoot Cast (Courtesy of Steve Richards MD, Texas Scottish Rite Hospital)

At the end of the serial casting phase of treatment, most children will require a minor operation (percutaneous Achilles tendon tenotomy) to lengthen the Achilles tendon and release the foot from residual plantar

flexion. Rarely, patients may also require a transfer of the tibialis anterior from its normal insertion on the first metatarsal to a new insertion on the third. This transfer will reduce supination of the foot with dorsiflexion. The operation is usually performed after age 3 years. If the Ponseti Method of casting, bracing, and tenotomy is applied correctly, it will be successful in more than 95% of cases.

After casting, the child must wear a foot abduction brace (Figure 3) at night for four or five years to maintain the correction. Without bracing, the deformity will likely recur because the muscles of the foot will pull it back into an abnormal position.



*Figure 3: Abduction Bracing (Courtesy of Steve Richards MD, Texas Scottish Rite Hospital)*

Recurrence (despite bracing) occurs in approximately 10% of patients, and most will respond to a repeated course of manipulation and casting; a few will require additional surgery to prevent further relapse.

Although the Ponseti Method is highly effective in idiopathic clubfeet, some patients (especially those who present late or whose deformities are secondary to neuromuscular disease) will not respond to non-operative treatment and will require surgical intervention.

Surgery is normally performed at 9-12 months of age. The goal of surgery is to correct all the deformities in one operation. The surgical procedure varies from patient to patient but generally involves releasing all joint capsule contractures, lengthening any shortened muscle-tendon units, and realigning the bones of the foot.

Compared to the results of non-surgical methods, the long-term outcomes from surgery are associated with more pain, stiffness, deformity, and muscle weakness. However, it is important to note that surgery is reserved for patients with more severe disease. Accordingly, this is a not a true head-to-head comparison.

Even with optimal treatment, the corrected clubfoot will be functionally normal but structurally different than the unaffected foot. The affected foot is often smaller (requiring a different shoe size) and less mobile than the other foot. Additionally, the calf muscles in the affected leg may also be smaller. In some cases of clubfoot, the affected leg will stop developing prematurely. Even if this were to occur, the affected leg will usually be no more than ½ inch shorter than the other leg – a clinically insignificant discrepancy. Rarely, there will be a difference in limb length sufficiently large that a surgery will be needed to lengthen the affected leg.


## RISK FACTORS AND PREVENTION

Honien et al (PMID: 11032161) found that family history of clubfoot is a major risk factor for developing clubfoot (OR = 6.52). Smoking exposure in utero is also associated with increased risk of clubfoot (OR = 1.34). This risk is dramatically increased in babies that already have a past family history of clubfoot (OR = 20.30). As a result, mothers with a positive family history of clubfoot should be strongly urged not to smoke while pregnant to minimize the risk of their child having clubfoot.

Clubfoot can be identified in the second trimester using fetal ultrasound. This can provide an opportunity for counseling about genetic testing for associated conditions as well as education about the treatment and prognosis of the condition.

## MISCELLANY

Figure-skater Kristi Yamaguchi, soccer star Mia Hamm, and NFL quarterback Troy Aikman were all born with clubfeet but still attained great success as professional athletes.

Clubfoot is uniformly treated at a very early age in the US, but in low-income countries it is not uncommon to see older children with neglected clubfoot (a deformity that was never treated), residual clubfoot (a deformity that was incompletely treated and retains aspects of the deformity years later), and recurrent clubfoot (a deformity that was completely treated but reverted back due to insufficient bracing). These forms of clubfoot can severely decrease quality of life by subjecting patients to physical disability and alienation.

Horses walk on their toes; hence, a deformity of plantar flexion that places the toes in lone contact with the ground is called "equinus."

## KEY TERMS

Clubfoot, Talipes equinovarus, Ponseti Method

## SKILLS

Recognize the gross manifestations of clubfoot and differentiate these from other congenital foot deformities. Explain treatment options for clubfoot, including the Ponseti Method of serial casting, bracing, and tenotomy. Describe the possible outcomes of clubfoot treatment, including restoration of normal form and function, residual deformity, and recurrent deformity.

# DEVELOPMENTAL DYSPLASIA OF THE HIP

Developmental dysplasia of the hip (DDH) is a condition characterized by varying degrees of hip instability in a developing hip joint (Figure 1). The condition has a wide spectrum of severity ranging from a mildly shallow hip socket (acetabulum) to a completely dislocated hip (Figure 2). DDH is often idiopathic but hip dislocations may also be caused by an underlying congenital disorder.
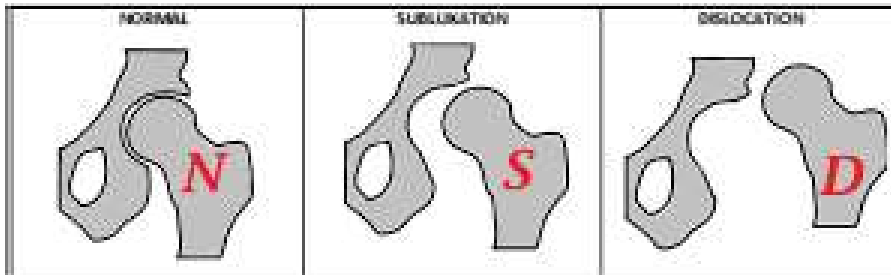


*Figure 1: Radiograph showing a dislocation of the left hip.*



*Figure 2: The dysplastic hip may be partially out of the socket (subluxated) or completely out (dislocated). Note that the acetabulum will not develop normally if the head is not located. (modified from Wikipedia)*

## STRUCTURE AND FUNCTION

The hip joint is a ball and socket joint: The femoral head (the ball) fits into the acetabulum (the socket). The joint is further stabilized by the surrounding joint capsule and ligaments.

The development of the femoral head and acetabulum is reciprocal: the concavity of the acetabulum develops in response to the presence of the spherical femoral head and vice versa. Thus, if the head is not located, neither side of the joint developed normally. This has profound implications for treatment of cases in which the diagnosis is delayed: it very well may be that the joint cannot be simply re-located, as the misshapen head will simply not fit in the shallow, under-developed acetabulum.

There are many theories about the etiology of DDH, but the definitive cause is still unknown. There are, however, known risk factors. Untreated DDH is a leading cause of degenerative joint disease in the hip.

## PATIENT PRESENTATION

DDH has a variable presentation depending upon patient age. Hip dysplasia in very young children can be detected only if one looks for it – the hip does not hurt, so the child does not complain, and because the child is not expected to walk, the parents don't notice it either.

In the first few months of life, infants are screened using the Ortolani and Barlow tests.

The Ortolani test reduces a dislocated hip, whereas the Barlow test attempts to dislocate an unstable hip. Both tests are performed on a supine infant.

The Ortolani test (Figure 3) is performed by an examiner flexing the infant's hips and knees and applying pressure on the greater trochanters while abducting the infant's legs. The test is positive if a 'clunk' can be heard or felt as the femoral head is relocated into the acetabulum.
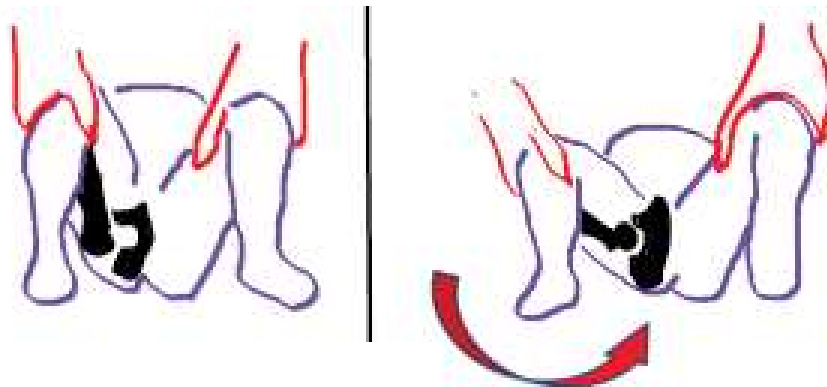


Figure 3: Schematic of the Ortolani test. In the panel at left, the hip is dislocated. The examiner's force, shown the arrows in the panel to the right, relocates the hip.

For the Barlow test (Figure 4), the examiner applies posterior pressure to the adducted leg in an attempt to subluxate or dislocate the femoral head from the acetabulum. If the femoral head can come out of the joint, a "clunk" may be felt as it comes out.
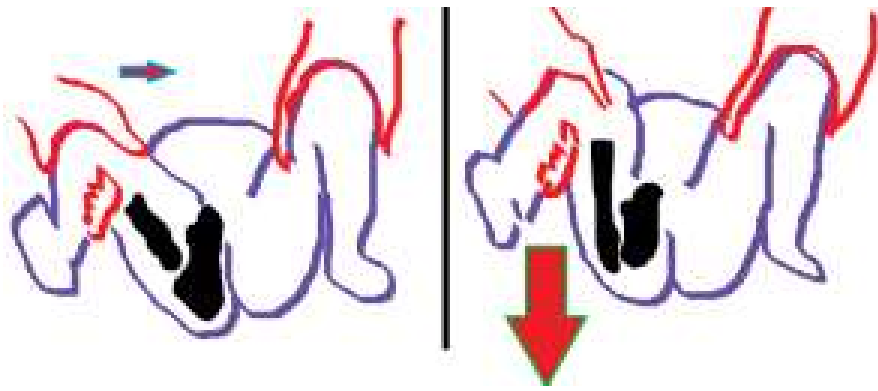
*Figure 4: Schematic of the Barlow test. In the panel at left, the femoral head is located in the acetabulum. The examiner's posterior-directed force, shown by the arrow in the panel to the right, dislocates the hip.*

By three to four months of age, soft tissues tighten and the Barlow and Ortolani maneuvers become less useful.

The soft tissue contractures may cause limited hip abduction on the affected side and the leg may appear shorter (Galeazzi sign, Figure 5) in a patient with a unilateral hip dislocation. In the case of a bilateral hip dislocation, abduction may be symmetric but limited.



*Figure 5: The Galeazzi sign. This photo of a prone child, taken from the foot of the bed, shows an apparent limb length discrepancy due to left hip dislocation. Indeed, the bones are the same length, but the left (as shown here) appears shorter because the left hip is not located.*

Children of walking age may develop an apparent leg length discrepancy or limited range of motion. In the setting of bilateral dislocated hips, the patient may walk with a waddling gait.

## OBJECTIVE EVIDENCE

For infants younger than 6 months, ultrasonography is the diagnostic imaging modality of choice. Ultrasound evaluates the morphology of the femoral head and acetabulum.

The alpha angle is a measurement that is the most useful to help guide treatment. It is the angle between the ilium and the acetabulum and should be greater than 60 degrees. In addition, ultrasound measures the femoral head coverage. In general, more than 50% of the femoral head (ball) should remain within the acetabulum (socket) (Figure 2 and 3).

In children older than 6 months, the femoral head begins to ossify, and thus plain radiographs can be used to evaluate for DDH.
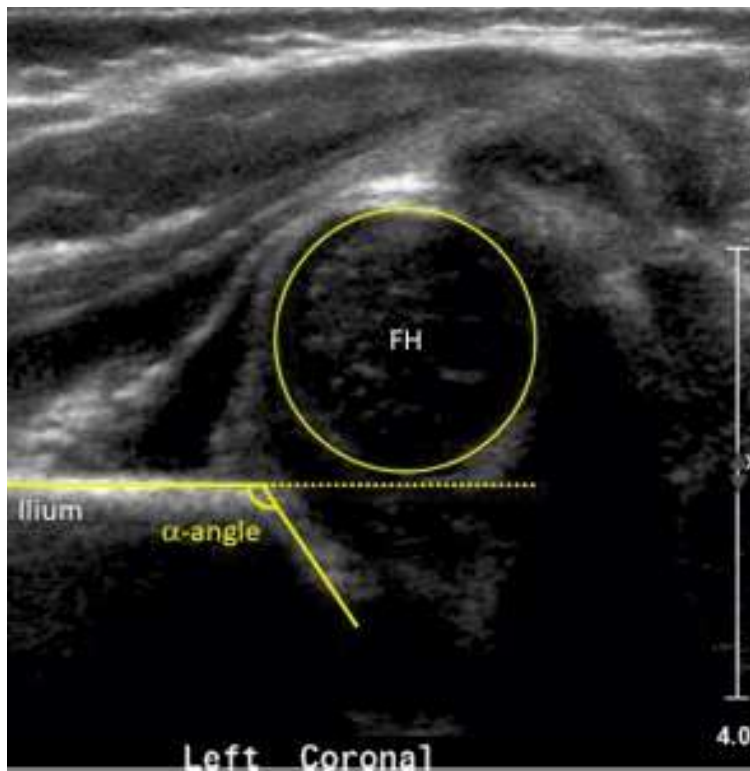
*Figure 6: Ultrasound image demonstrating a complete hip dislocation. Note that the femoral head is normally bisected by a line drawn from the ilium (dotted line), but in this case, the femoral head (FH) is completely uncovered.*
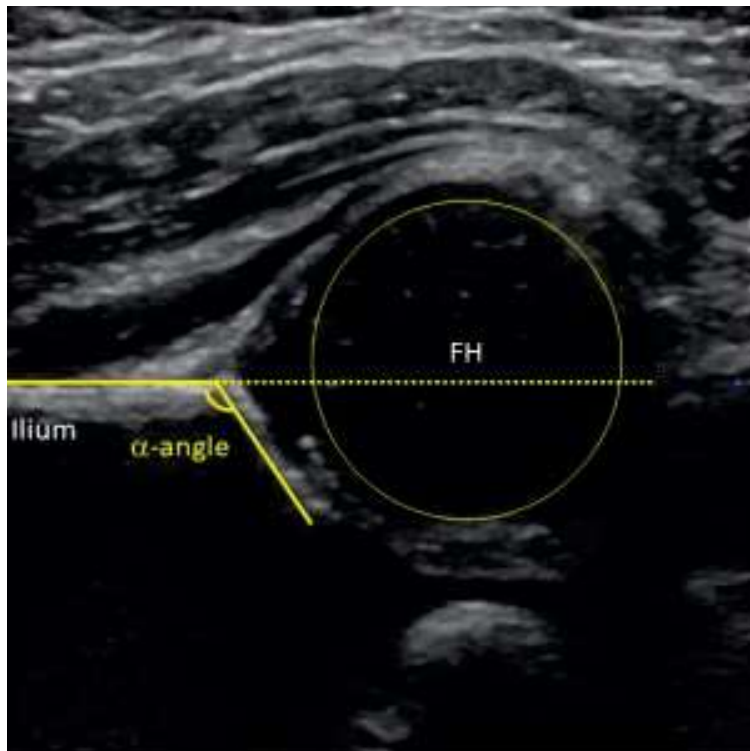


*Figure 7: Ultrasound image demonstrating a normally developing hip. Note that the femoral head is bisected by a line drawn from the ilium (dotted line).*

DDH is a common abnormality of skeletal development. Some form of DDH occur in about 1.3 out of 1000 births. It more commonly affects females as compared to males, and is more common in Native American, Eastern European, and Sami populations.

## EPIDEMIOLOGY

Developmental dysplasia in the form of a subluxated (or subluxatable) hip is very common, seen approximately in 1% of all births. Frank dislocation is much rarer, seen in approximately 1/1000 births. The condition is six times more common in females. Interestingly, the right hip is affected in isolation only approximately 20% of cases. In 60% of cases, the left hip is affected, and in the remaining 20%, the condition is bilateral. The risk of developmental dysplasia is five times higher if there is an affected sibling and 10 times higher if there is an affected parent.

## DIFFERENTIAL DIAGNOSIS

DDH is commonly idiopathic and seen in isolation. Some associated conditions include metatarsus adductus and torticollis. Metatarsus adductus is a condition in which the bones of the forefoot are turned inward creating a "pigeon-toed" appearance of the foot. Torticollis is a condition in which an infant holds his or her head tilted to one side and is caused by a shortened/tight muscle in the neck. Both of these conditions, along with DDH, are considered "packaging disorders" (so named because it appears that an otherwise normal fetus is "packaged" too tightly within the uterus, leading to deforming forces on the limbs).

Occasionally, hip dysplasia may be associated to an underlying neuromuscular condition, such as myelomeningocele or arthrogryposis. These cases are treated differently than idiopathic cases.

## RED FLAGS

Although DDH is most often an isolated condition, some cases are secondary to underlying neuromuscular conditions such as spina bifida. Thus, the presence of hip instability should be deemed a red flag, prompting a close diagnostic evaluation to exclude these conditions.

## TREATMENT OPTIONS AND OUTCOMES

The goal of treatment is to reduce and maintain the femoral head in the acetabulum as early as possible to allow the hip joint to develop normally.

The standard of care for DDH diagnosed before 6 months of age is treatment in a dynamic hip brace (eg, Pavlik harness) that maintains the hips in a flexed and abducted position (Figure 8). These devices gently nudge the femoral head into the correct position and prevent extension and adduction of the hips — the maneuvers in the Barlow test that would dislocate the hip.

These devices are worn for 23 hours/day for at least 6 weeks. It is critical that treatment is discontinued if the hip is not reduced by 4 weeks, as erosions of the pelvis may be caused. The overall success rate is 90%.

Children who are diagnosed between 6-18 months old or who failed brace treatment are typically treated with closed reduction and spica casting. A closed reduction procedure involves manually reducing the hip joint while the patient is sedated. A hip arthrogram (dye injected into the joint) is obtained to evaluate for a concentric reduction. Children are then immobilized in a spica cast to maintain the hip in a good position.

*Figure 8: An infant wearing a dynamic hip brace (from Wikipedia).*

Children who are diagnosed when they are older than 18 months old or who failed other treatment options are usually treated with open reduction. This involves surgically removing any obstacles to reduction and may also include a pelvic or femoral osteotomy followed by spica casting (Figure 9).



*Figure 9: AP pelvis in a 3-year-old female who underwent staged bilateral open hip reductions with pelvic and femoral osteotomies. The left femur plate will be removed at a later date.*

Long-term follow up is essential as residual dysplasia may occur as often as 10-20% even after successful treatment of DDH. Occasionally, additional surgeries are required later in life to ensure proper development of the hip socket in order to minimize the risk of early osteoarthritis.

## RISK FACTORS AND PREVENTION

The exact causes of DDH are unknown, but it is likely a combination of several environmental and genetic factors. Known risk factors include:

- Inadequate intra-uterine space (e.g. firstborn child, oligohydramnios, etc.)
- Female sex,
- Family history,
- Breech presentation.

## MISCELLANY

In the United States, DDH is commonly treated at a very early age, but in low-income countries this condition may go undiagnosed and untreated. While this does not affect normal child development, older children and young adults with untreated hip dislocations often have decreased quality of life due to pain and limitation of function.

DDH was previously referred to as congenital dislocation of the hip (CDH). This terminology changed over the last decade due to the recognition that some infants will have a normal hip examination at birth but develop hip disease during the first year of life.

## KEY TERMS

Developmental dysplasia of the hip, Hip dysplasia, Hip dislocation, Acetabular dysplasia, Pavlik harness.

## SKILLS

Recognize the variable presentation of DDH. Understand the different clinical presentations that depend upon patient age and severity of hip instability. Explain treatment options for DDH, including the Pavlik harness, bracing, closed and open reduction techniques.

# LEGG-CALVE-PERTHES DISEASE

Legg-Calve-Perthes disease, commonly known as Perthes disease, is a hip disorder affecting children that is caused by decreased blood flow to the head of the femur. This results in osteonecrosis (also known as "avascular necrosis") of the proximal femoral epiphysis (femoral head), with resorption, reossification, and remodeling of the bone. Especially in children under the age of 6, Perthes disease may resolve without sequelae; in older children, however, the bone may fail to remodel to a normal shape leading to disruption of the articular surface and degenerative joint disease later in life.

## STRUCTURE AND FUNCTION

The blood supply to the femoral head is made up of three arteries: the medial and lateral femoral circumflex arteries, and the artery of the ligamentum teres.

In Legg-Calve-Perthes disease, the blood supply is disrupted by an unknown process, and the bone cells begin to die. This process is termed osteonecrosis.

Loss of blood supply to bone results in ischemia and cell death. In turn, cell death leads to less bone remodeling and poorer structural properties of bone. These weaker properties increase the risk of collapse with load.

With time, new blood supply reaches the femoral head, and necrotic bone is removed. This results in fragmentation and reossification.

Notably, and in contrast to the adult form of idiopathic osteonecrosis, Perthes disease in younger children may resolve without sequelae. That is, the bone may remodel to a fairly normal shape with no lingering symptoms. On the other hand, the regenerated bone may also remodel to a non-spherical shape producing an abnormal range of motion, an irregular articular surface, and, ultimately, degenerative joint disease of the hip.

The exact cause of the disruption of the blood supply is not known, although it is believed to be multifactorial. Genetic abnormalities, trauma, coagulopathy, collagenopathy, hyperactivity, and passive smoking exposure are seen with higher prevalence in patients with Perthes, and are therefore thought to possibly have a causative role.

Another theory links systemic delay in growth and development to the development of Perthes. Delayed bone age has been seen among patients with Legg-Calve-Perthes disease, and therefore, endocrine dysfunction has been implicated as a possible cause.

## PATIENT PRESENTATION

The most common age of presentation for Perthes is about 8 years of age, though patients may be as young as 2 or as old as 12. (In patients 12 years of age or older, the natural history of the condition mirrors that of adult osteonecrosis.)

A history of trauma to the painful extremity is sometimes present.

Children with Legg-Calve-Perthes disease typically present with a limp that worsens with activity. Decreased range of motion and pain that radiates into the groin, proximal thigh, or knee are seen as well. There is loss of passive hip rotation on physical exam. Children may often conceal their limp or not complain of it, but parents and caretakers will notice it. In some cases, the child presents due to an unrelated trauma to the affected limb, and the disease is found incidentally.

Children with Legg-Calve-Perthes disease might exhibit an abnormal gait pattern in which they avoid abduction or internal rotation of the leg.

Additionally, in longstanding cases characterized by limited use of the leg over a prolonged period of time, atrophy may set in. Thus, the affected limb may be smaller than the contralateral side.

The definitive diagnosis of Perthes disease is made by x-ray. The disease is primarily classified by radiographic findings, using the Waldenstrom classification. The four stages of Perthes (Figure 1-4) are denoted as *initial, fragmentation, reossification,* and *healed.*

In the initial stage, the osteonecrosis process begins. The x-rays may show a dense and flattened femoral head with joint space widening. There may be only mild symptoms in this stage; parents may notice an altered gait or mild limp.



*Figure 1: The initial stage of Perthes disease. There is irregularity of the left femoral head.*
*(Figures 1- 4 are provided by Dr Wudbhav Sankar, Associate Professor of Orthopaedic Surgery at The Perelman School of Medicine/University of Pennsylvania and The Children's Hospital of Philadelphia)*

The fragmentation stage is characterized by revascularization and bone resorption producing collapse. Hip symptoms are most pronounced in this stage, which may present for a year or longer.

*Figure 2: In the fragmentation phase, the femoral head is resorbed and collapses.*

In the reossification stage, new bone appears as the necrotic bone is resorbed. This is the longest stage and may persist for two years or more.



*Figure 3: Perthes of the left hip in the third stage: re-ossification.*

In the final stage, the femoral head completes its remodeling. This stage is labeled the "healed" stage – a term that refers specifically to the completion of the bone's response to the original insult. The word "healed" does not necessarily reflect the state of the hip joint overall. That is, although the osteonecrosis process may have ended and the fragmented areas have reconsolidated, symptoms may still persist and later degenerative changes may still appear if the shape of the femoral head has not been restored to normal.

*Figure 4: Perthes of the left hip in the fourth and final stage, in which the infarction is "healed." As shown, however, the shape of the head has not been fully restored.*

## OBJECTIVE EVIDENCE

On x-ray, the femoral head is typically lateralized and more radiodense, and in later stages of disease the femoral head appears flattened.

Other radiographic findings can include sclerosis and flattening of the acetabulum.

Another option for imaging is MRI, which has a higher diagnostic accuracy than x-ray, especially in the initial stage when radiographic findings may not be obvious. Scintigraphy is a method which uses Technetium scanning and can be helpful for diagnosis in the earliest stages of disease.

Laboratory findings are used in the diagnosis of Legg-Calve-Perthes disease primarily to rule out other diseases or illnesses. All laboratory values are typically within normal limits.

## EPIDEMIOLOGY

Legg-Calve-Perthes disease is a relatively rare disease, with a prevalence of approximately 1 in 10,000 children, but can have devastating long-term consequences on the child's mobility and quality of life.

The age of onset of Legg-Calve-Perthes disease is most commonly between 4 and 8 years old. In general, it occurs in children under the age of 15 years old. Boys are more commonly affected, with a ratio of male to female of approximately 5:1. In about 10% of all cases, both hips are affected.

Legg-Calve-Perthes disease is more common in children of central European descent, and less common in East Asian and African American populations.

## DIFFERENTIAL DIAGNOSIS

As Perthes is an idiopathic (cause-unknown) disease, diagnoses are made only after all other *known* causes of osteonecrosis have been ruled out. These include hemoglobinopathies, like sickle cell disease or thalassemia,

leukemia, lymphoma, idiopathic thrombocytopenic purpura, or hemophilia. Secondary causes of osteonecrosis such as a history of corticosteroid use, traumatic dislocation, septic arthritis, or untreated developmental dysplasia of the hip should be ruled out as well.

Other diseases that cause epiphyseal dysplasia can be mistaken for Perthes, such as multiple epiphyseal dysplasia or spondyloepiphyseal dysplasia. However, these conditions are typically synchronous and symmetric in appearance unlike Perthes. In children under the age of 3, Meyer's dysplasia should be considered as well. Meyer's dysplasia is a rare condition in which there is delayed ossification of the proximal femur. Magnetic resonance imaging showing multiple centers of ossification of the femoral heads without edema can confirm the diagnosis.

## RED FLAGS

The presence of bilateral osteonecrosis is suggestive of other disease processes, such as multiple epiphyseal dysplasia. A child with a limp can also be the presentation for septic hip arthritis or a slipped capital femoral epiphysis, both of which should be promptly diagnosed. Additionally, symptoms such as fever, anemia, or leukocytosis should not be missed as these can be clues for ruling out other important conditions.

## TREATMENT OPTIONS AND OUTCOMES

Treatment of Legg-Calve-Perthes disease is guided primarily by the stage of disease (itself usually correlated with patients' age).

The main goal of treatment is maintaining range of motion and containment of the femoral head within the acetabulum. By achieving these goals, one can prevent or minimize deformity of the joint, and achieve greater long-term function.

"Containment" minimizes loss of sphericity as the bone remodels. Containment can be achieved either operatively or nonoperatively.

Nonoperative management of Perthes centers on offloading the hip. If the disease is in the fragmentation phase, the child should be non-weight-bearing. Motion is maintained by daily exercises at home or with physical therapy. Containment can be also achieved with the use of an abduction brace or Petrie cast (Figure 5).



*Figure 5: Application of a Petrie Cast to maintain abduction and promote containment.*

In some cases, operative treatment is needed to promote motion with soft tissue releases, or to improve containment with an osteotomy (Figure 6).

Overall, the long-term prognosis for children with Perthes is good; most children are able to return to daily activities without symptoms or limitations.

The most important predictors of long-term prognosis are age at the time of diagnosis and the eventual shape and congruity of the femoral head within the acetabulum. Younger patients typically do better, and more spherically-shaped heads and greater congruity of the head within the hip joint portend better outcomes.



Figure 6a: An x-ray of a varus osteotomy for Legg-Calve-Perthes Disease. The normal neck-shaft angle is shown in red in the left hip; the new smaller (varus) angle on the right is shown in green.
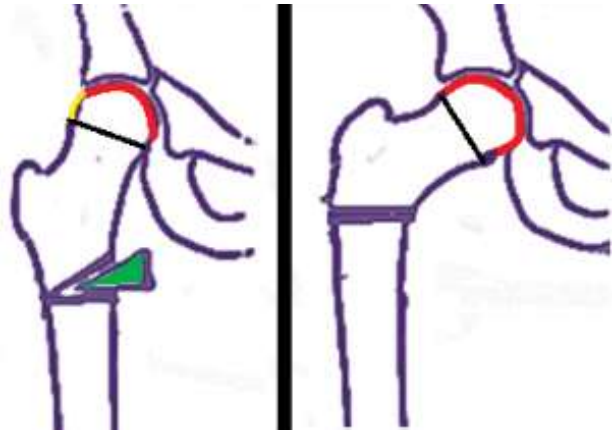
Figure 6b: As shown in the schematic, some of the head is not contained in the acetabulum (yellow region). Cutting out a wedge of the bone (green triangle) will produce a smaller angle between the femoral neck and shaft. This will redirect more of the head into the acetabulum. This greater contact between the ball and the socket will help mold the femoral head into its proper rounded form.

## RISK FACTORS AND PREVENTION

A few proposed risk factors include passive exposure to smoke and hyperactivity, although there is no known mechanism linking these risk factors to the development of Legg-Calve-Perthes disease.

## MISCELLANY

Although the exact cause of Perthes disease is unknown, many environmental and holistic risk factors have been proposed. One association has been noted between Perthes and attention deficit-hyperactivity disorder. Additional investigation is also being carried out regarding Perthes and the role of nutrition.

Perthes disease was first described by Henning Waldenstrom. (Dr. Waldenstrom believed the condition was a variant of tuberculosis. The disease is named after the three physicians who independently found that it was not related to tuberculosis: Arthur Legg, Jacques Calve, and Georg Perthes.) Legg-Calve-Perthes disease thus follows "Stigler's Law of Eponymy," which asserts that no scientific discovery is named after its original discoverer. (Indeed, although the law was named by Stephen Stigler, it was discovered by the sociologist Robert K. Merton.) Note that Henning Waldenstrom was not the discoverer of the eponymous Waldenstrom's Macroglobulinemia; that was Jan G. Waldenstrom, his son.

## KEY TERMS

Osteonecrosis (Avascular necrosis), Bone remodeling

**SKILLS**

Be able to complete a history and physical exam of the hip in a child. Interpret an x-ray of Perthes.

# CHAPTER 11.

## SLIPPED CAPITAL FEMORAL EPIPHYSIS

Slipped capital femoral epiphysis (SCFE, pronounced "skiffy") occurs when there is abnormal movement of the femoral metaphysis relative to the epiphysis along the physis (growth plate). Thus, SCFE is technically a growth plate fracture. Because the femoral head is secured by the acetabulum, a sufficiently large force applied across the physis can cause the neck and shaft to "slip" away from the head (Figures 1 and 2). SCFE is most commonly (but not exclusively) seen in overweight, adolescent males. SCFE is usually treated with percutaneous pin fixation, to prevent further slippage.
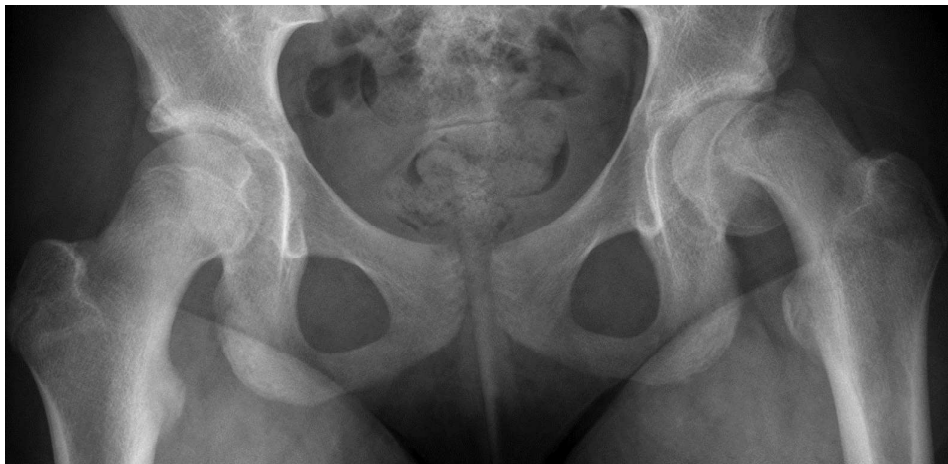


*Figure 1: A slipped capital femoral epiphysis of the left hip is shown on the radiograph. (Case courtesy of Prof Frank Gaillard, Radiopaedia.org, rID: 8229)*



*Figure 2: The relationship of the femoral head to the neck after a slip has been likened to an ice cream scoop falling off its cone.*

## STRUCTURE AND FUNCTION

SCFE is usually seen during periods of rapid growth: in boys between the ages of 12 and 16 and girls between the ages of 10 and 13. This suggests that some factor related to growth makes the growth plate susceptible. Sometimes SCFE develops gradually, over several weeks or months, without overt injury; by analogy, this is a "pathological fracture" of the growth plate, suggesting an underlying disorder of the growth plate tissue.

Several studies have revealed pathologic physeal changes in SCFE, including replacement of the normal growth plate with abnormal cartilaginous and fibrous tissue. There may be widening of the physis along with hypocellularity and loss of columnar organization. The slip generally occurs through the proliferative and hypertrophic zones of the physis, the site where chondrocytes and normal components may have been replaced by ground substance. After fixation and stabilization of the slip, these pathologic findings seem to revert to more normal structure, at least partially.

In most cases of SCFE, the femoral neck and shaft slip anteriorly and rotate externally relative to the femoral head. Following this migration, hip flexion may cause impingement of the anterior femoral metaphysis leading to cyst formation, damage to the labrum or acetabular cartilage, and other degenerative changes.

The etiology of SCFE is unknown in most cases and is thought to possibly be multifactorial. In general, the slip is caused by an inability of the proximal femoral physis to resist physiologic loads.

Endocrinopathies that weaken the physis are thought to be responsible for 5% to 8% of SCFE cases. These include hypothyroidism, panhypopituitarism, growth hormone deficiency, parathyroid derangements, and hypogonadism. In patients with Down syndrome, the increased prevalence of hypothyroidism might explain the increased prevalence of SCFE. Renal osteodystrophy may also weaken the physis due to associated secondary hyperparathyroidism. Nonetheless, routine screening for endocrinopathies is not recommended for all patients with SCFE.

A history of radiation to the hip or pelvis as well as genetic and immunological factors may also have a role in physeal pathology, though the exact contributions are unclear.

Finally, other anatomic factors that increase risk of physeal failure increase risk of SCFE, particularly in patients with concomitant obesity. This includes reduced femoral anteversion (or retroversion) whereby an abnormally-reduced neck-shaft angle positions the physis more vertically and subjects it to greater shearing forces.


## PATIENT PRESENTATION

The prototypical patient with SCFE is an obese, but otherwise healthy, adolescent male between the ages of 8 and 15. Patients who present younger than 8 or older than 15 years of age are more likely to have an underlying endocrinopathy. Patients often present with hip/groin pain, limp, and decreased range of motion at the hip. Notably, half of the children with SCFE do not report pain at all and some may report pain only at the distal thigh or knee. (This is due, most likely, to irritation of the obturator nerve near the hip.) Others may simply complain of altered gait.

Patients may identify an inciting event for their symptoms, though these episodes rarely involve high-energy trauma. Symptoms may have persisted for days or weeks prior to the child's initial presentation. Both hips must be evaluated, as many cases of SCFE initially present with bilateral disease.

The patient will often present with decreased internal rotation, abduction, and flexion of the affected hip.Increasing slip severity can cause a propensity towards "obligate external rotation", meaning the hip rotates externally as the examiner flexes the hip. The child may also have an antalgic gait with progressive external rotation of the foot and knee. In more severe slips, the affected limb may also appear shorter than the contralateral side.

On presentation, SCFE can be classified according to whether the patient is able to bear weight on the affected side. In a "stable SCFE", the patient is able to bear weight. In an "unstable SCFE", the patient cannot bear weight or walk; this is usually associated with a more severe slip. This classification is particularly important for prognostic considerations as stable SCFEs have lower risk of osteonecrosis and other complications.

## OBJECTIVE EVIDENCE

Anteroposterior (AP) and lateral plain radiographs of both hips are typically the only imaging necessary for cases of suspected SCFE. Cross-table lateral films may be taken instead of a frog-leg lateral in cases of unstable SCFE in order to avoid further displacement or pain to the patient.

Frog lateral views are generally the most reliable means for detection of SCFE. As the slip progresses, it becomes evident on the AP view as well. Possible findings include widening of the physis, anterior displacement and external rotation of the femoral neck and shaft relative to the head, decreased height of the capital femoral epiphysis, and increased distance between the metaphysis and acetabular teardrop.

In a normal hip, part of the femoral head is intersected by Klein's line drawn along the lateral border of the femoral neck on a frog lateral image (Figure 3). In SCFE, the femoral head sits "below", or inferomedial to, Klein's line, though the reliability of this sign has been questioned recently.



Figure 3: Frog lateral x-ray of a 12-year-old male with several weeks of right groin pain who was found to have right SCFE. Klein's line is shown in green bilaterally. This line should intersect the lateral part of the superior femoral epiphysis as shown on the left. If the line fails to contact bone in the epiphysis, as shown on the right, SCFE can be diagnosed.

The magnitude of the slip is often described in terms of femoral head displacement as a percentage of the femoral neck diameter (mild is less than 33%, moderate is 33% to 50%, and severe is greater than 50%). Alternatively, the magnitude of the slip angle, measured between the femoral epiphysis and neck on the lateral view, can be described (mild is less than 30 degrees, moderate is 30 to 60 degrees, and severe is greater than 60 degrees).

Though additional imaging is generally unnecessary, magnetic resonance imaging (MRI) can detect distortion of the physis with bone marrow edema before the development of radiographically detectable SCFE. This is termed a "pre-slip" (Figure 4). Computed tomography (CT) is rarely needed but can allow for evaluation of physeal closure in patients who present very late in the course of SCFE.

*Figure 4: MRI can detect a slipped epiphysis even without much displacement. (Image courtesy Journal of Children's Orthopaedics Vol. 11, No. 2 https://doi.org/10.1302/1863-2548-11-160276)*

## EPIDEMIOLOGY

SCFE has an annual incidence of 2 to 13 per 100,000 and is 1.5 to 2 times more common in males. This condition typically affects boys ages 12 to 15 years and girls ages 10 to 13 years. SCFE is more common among patients with higher body weights. About 50% of SCFE patients are at or above the 90th percentile for weight, and about 70% are over the 80th percentile. The current literature suggests that more than half of children with SCFE are obese. Twenty percent of SCFE patients present with initial bilateral involvement. Another 10% to 20% develop a contralateral slip an average of 18 months after the initial slip.

## DIFFERENTIAL DIAGNOSIS

SCFE should be suspected in all children with open physes, a limp, and complaints of hip, groin, thigh, or knee pain until proven otherwise. Other conditions that may be considered include transient synovitis, Legg-Calve-Perthes disease, septic arthritis, osteomyelitis, and fracture. Various metabolic and systemic disorders may also be associated with SCFE, such as obesity, endocrinopathies, renal osteodystrophy, and anatomic variation of the hip joint.

## RED FLAGS

Any adolescent patient that presents with a limp and complains of pain in the hip, groin, thigh, or knee should be considered to have SCFE until proven otherwise. Specifically, complaints of thigh or knee pain merit examination of the hip. Such an exam might prevent delayed diagnosis, misdiagnosis, and unnecessary imaging or procedures. Additionally, both hips should be examined because of the high incidence of bilateral SCFE.

Diagnosis of SCFE in pre- or post-pubertal patients should raise suspicion for underlying metabolic or systemic abnormalities. Inability to bear weight on the affected extremity may indicate an unstable SCFE, a condition that requires more urgent intervention.

## TREATMENT OPTIONS AND OUTCOMES

Both stable and unstable SCFE require surgical management in order to prevent slip progression. In situ fixation is performed in most cases. Forceful manipulation is never recommended as this may increase the risk of osteonecrosis. This is particularly important for stable slips which, by definition, cannot be easily reduced.

In situ fixation is most commonly achieved with cannulated screws under fluoroscopic guidance. One screw is typically sufficient for stable slips (Figure 5). Two screws are occasionally used in unstable cases to provide increased fixation strength. For the single screw approach, the screw should be inserted perpendicular to the physis and in the center of the epiphysis in both the AP and lateral planes. This "center-center" location reduces the risk of penetration through the posterior femoral neck and resultant osteonecrosis. It also increases the surgeon's ability to achieve good screw purchase in the femoral head.
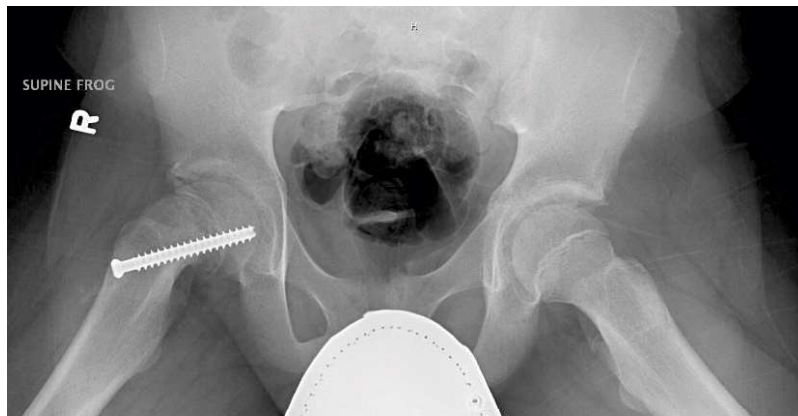


Figure 5: Post-operative frog lateral x-ray of the patient in Figure 3 demonstrating in situ fixation of right SCFE.

Prophylactic pinning of the contralateral (normal-appearing) hip is controversial. In brief, if the contralateral hip is pinned before it slips, by definition there will be no displacement. Preventative pinning may be beneficial because displacement is associated with a higher risk of degenerative joint disease. On the other hand, there are risks associated with prophylactic surgery in an unaffected hip, and if that surgery was indeed not necessary – something that will never be known – those risks were assumed in vain. Prophylactic fixation is performed more commonly in SCFE patients with a higher risk of developing a contralateral slip, including patients with significant growth remaining, endocrinopathies, or history of pelvic radiation.

For severe slips, in situ pinning may result in residual deformity, impingement, and degenerative changes. Additionally, sufficient stabilization of a severe slip with in situ pinning is difficult because it is not always possible to obtain the proper screw trajectory. Accordingly, closed reduction maneuvers have been described for severe, unstable SCFE. These should be performed only by experienced surgeons and only with gentle manipulation. The goal of this manipulation is not anatomic alignment, rather sufficient reduction to allow for a more appropriate screw trajectory. While outcome studies suggest osteonecrosis is associated with closed reduction maneuvers, it is not clear that this elevated risk is due to the reduction maneuver itself. It's possible as well that reduction is reserved for the more severe slips, and thus the elevated risk of osteonecrosis seen is a manifestation of underlying disease severity. Open procedures are technically demanding operations and usually require a trochanteric osteotomy to visualize and correct the deformity. Even in experienced hands, there is a risk of osteonecrosis and other post-operative complications. Thus, treatment of these severe, unstable slips remains a matter of debate.

Several series have reported good or excellent outcomes in 90% to 95% of patients treated with in situ fixation. In series with less favorable results, outcomes seem to worsen with increasing slip severity and longer follow-up. Osteoarthritis is a common long-term consequence of both treated and untreated SCFE. Arthritis is caused by both anatomic and biomechanical changes that damage the articular cartilage of the hip joint. More severe slips appear to be at risk for earlier onset and increased severity of osteoarthritis.

Two potential complications of in situ fixation are osteonecrosis and chondrolysis. Osteonecrosis may be iatrogenic or the result of an unstable slip itself. While stable slips rarely lead to this complication, osteonecrosis is seen in about 55% of unstable cases. Chondrolysis, namely rapid and progressive loss of articular cartilage, can be caused by unrecognized pin penetration. Patients with osteonecrosis or chondrolysis tend to have poor outcomes and early osteoarthritis. Other complications include screw failure or impingement, slip progression, leg length discrepancy, and fracture of the proximal femur.

## RISK FACTORS AND PREVENTION

Though the specific cause of SCFE is often unknown, a number of factors are thought to increase the risk of a slip. These include obesity, endocrinopathies, renal osteodystrophy, prior radiation therapy, and anatomic abnormalities of the hip joint. Among patients with an endocrine abnormality, SCFE is 6 to 8 times more prevalent than it is in the general population. Accordingly, children with Down syndrome are at elevated risk due to the higher prevalence of hypothyroidism in this population. An underlying metabolic or systemic disorder should be considered in SCFE patients who are younger than 8 years, older than 15 years, or underweight.

## MISCELLANY

On standardized examinations, if the question stem describes an obese adolescent who complains of leg pain or limps following minor trauma or sports activity, the correct answer likely involves SCFE.

Exam writers also expect you to recognize SCFE on the differential diagnosis of knee pain in the case of a "classic SCFE patient."

Some studies of adult total hip arthroplasty patients suggest that 40% of these individuals initially suffered some type of pediatric hip disorder, SCFE chief among them.

Slipped capital femoral epiphysis, pediatric hip, osteoarthritis, osteonecrosis.

## SKILLS

Perform a thorough physical examination of the hip. Identify risk factors for SCFE. Diagnose SCFE on plain radiographs. Identify potential complications (osteonecrosis & chondrolysis).

# ADOLESCENT IDIOPATHIC SCOLIOSIS

Scoliosis is a rotational deformity of the spine in both the coronal and sagittal planes (Figure 1). A diagnosis of idiopathic scoliosis is made when the coronal plane Cobb angle (see Figure 2 ) is >10 degrees on plain film radiographs and there is no underlying spinal cord pathology, neuromuscular disorder, or congenital malformation present. Adolescent idiopathic scoliosis is diagnosed in children between the ages of 10 and 18 years old and represents 80 percent or more of all cases. (Historically, if scoliosis is diagnosed in a patient 4 years of age or younger, it would be designated 'infantile idiopathic scoliosis' with the term 'juvenile idiopathic scoliosis' referring to patients ages 4-10, but more recently all cases of scoliosis diagnosed before the age of 10 are classified as 'early onset scoliosis' [discussed in another chapter.])

Depending on the severity of the curve and age of the patient, scoliosis can be managed with observation, bracing, or surgery. While scoliosis is not generally associated with pain during adolescence, more advanced curves (>40 degrees) can be associated with higher rates of low back pain in adulthood.
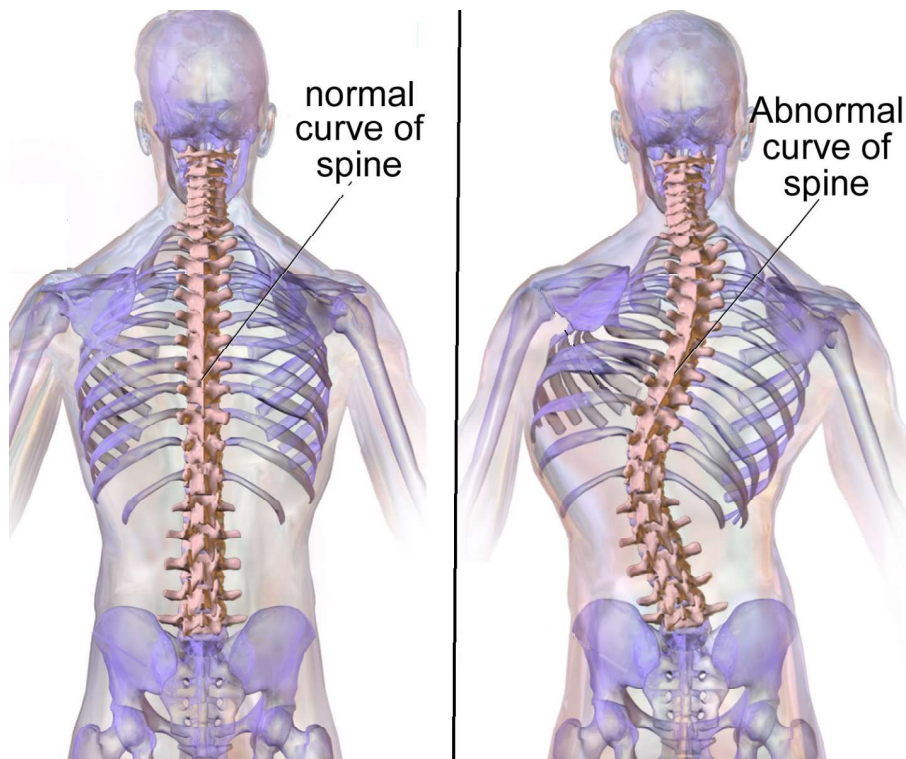


*Figure 1: A normal spine and one with scoliosis (Images from Medical gallery of Blausen Medical 2014. WikiJournal of Medicine 1 (2). DOI:10.15347/wjm/2014.010. ISSN 2002-4436. https://commons.wikimedia.org/w/index.php?curid=27796937)*
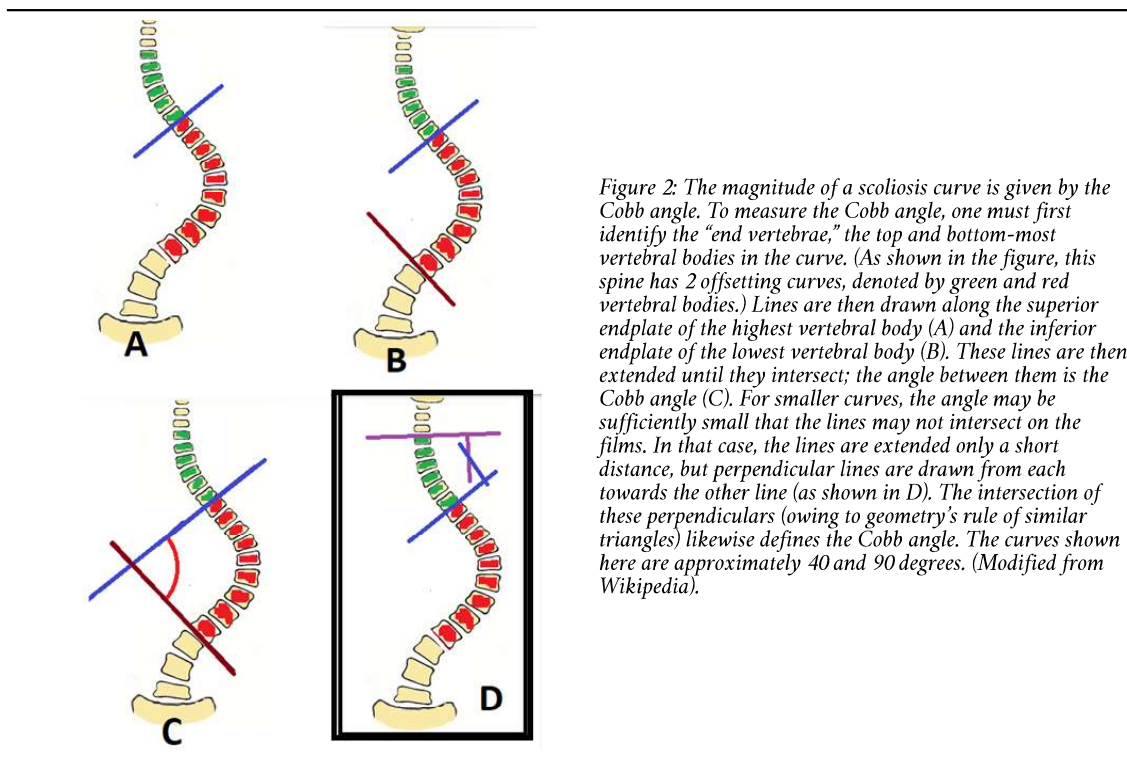
Figure 2: The magnitude of a scoliosis curve is given by the Cobb angle. To measure the Cobb angle, one must first identify the "end vertebrae," the top and bottom-most vertebral bodies in the curve. (As shown in the figure, this spine has 2 offsetting curves, denoted by green and red vertebral bodies.) Lines are then drawn along the superior endplate of the highest vertebral body (A) and the inferior endplate of the lowest vertebral body (B). These lines are then extended until they intersect; the angle between them is the Cobb angle (C). For smaller curves, the angle may be sufficiently small that the lines may not intersect on the films. In that case, the lines are extended only a short distance, but perpendicular lines are drawn from each towards the other line (as shown in D). The intersection of these perpendiculars (owing to geometry's rule of similar triangles) likewise defines the Cobb angle. The curves shown here are approximately 40 and 90 degrees. (Modified from Wikipedia).

## STRUCTURE AND FUNCTION

The spine consists of 7 cervical, 12 thoracic, 5 lumbar, and 5-7 sacro-coccygeal vertebrae. Normal alignment is critical to maintain the balance of the axial skeleton between the head and pelvis. This alignment is measured in both the coronal and sagittal plane using a plumb line starting at the base of C7. In neutral coronal balance, the C7 plumb line overlaps the center of the sacrum. When the C7 plumb line is to the <u>left</u> of the sacrum, there is negative coronal balance and when it is to the <u>right</u> there is positive coronal balance. In neutral sagittal balance the C7 plumb line touches the posterior-superior corner of S1. If the C7 plumb line falls anterior to S1 there is positive sagittal balance, and if it falls posterior to S1 there is negative sagittal balance.

In the coronal plane, the spine should be straight (<10 degrees of curvature), but on the sagittal plane the lumbar spine typically has 20-55 degrees of lordosis (inward curving, as seen from the side), while the thoracic spine has a physiologic kyphosis (an outward curve or hunching of the spine) of 20-45 degrees.

## PATIENT PRESENTATION

Adolescent idiopathic scoliosis is classically described as a painless condition. Low back pain is becoming increasingly more common among adolescent patients, and some of them will have scoliosis; but scoliosis should not be the default explanation of the symptoms.

A common complaint of adolescent idiopathic scoliosis patients is the appearance of the back and posture from a pronounced rib "hump," trunk shift, and uneven shoulders. Appearance issues can range from mild dysphoria to debilitating psychosocial distress.

Scoliosis may progress with growth. It is therefore important to establish the patient's growth history and document growth at each follow-up visit. On average, boys grow until the age of 16 and girls grow until the age of 14, or 2 years after menarche. Predicting growth is difficult, but some tools to help predict remaining growth include the Risser sign and Sanders stage (discussed below).

A complete physical exam begins with inspection of the patient standing upright facing away from the examiner. Scapular prominence, waist crease asymmetry and arm-side space can be observed. From a lateral view, the hypokyphosis of the thoracic spine can also be noted. Shoulder asymmetry, trunk shift and pelvic asymmetry can be observed and palpated. During the examination, it is important to identify a potential leg length discrepancy that can mimic scoliosis. If leg length difference is noted, the examiner can place a block under the shorter leg to assess if the curve corrects.

The Adam's forward bending test is an easy screening test. It is performed by having the patient lock his or her knees in extension and attempt to bend forward and touch toes. The patient is then observed from behind, to note the presence of an asymmetries of the rib cage and shoulder blades. Any rib prominence should be measured using a scoliometer (Figure 3). A rib prominence deformity greater than 7 degrees should prompt an evaluation by a pediatric spine specialist. As a rule of thumb, 7 degrees of prominence on Adam's forward bending test corresponds curve measuring about 10 to 20 degrees on x-ray.
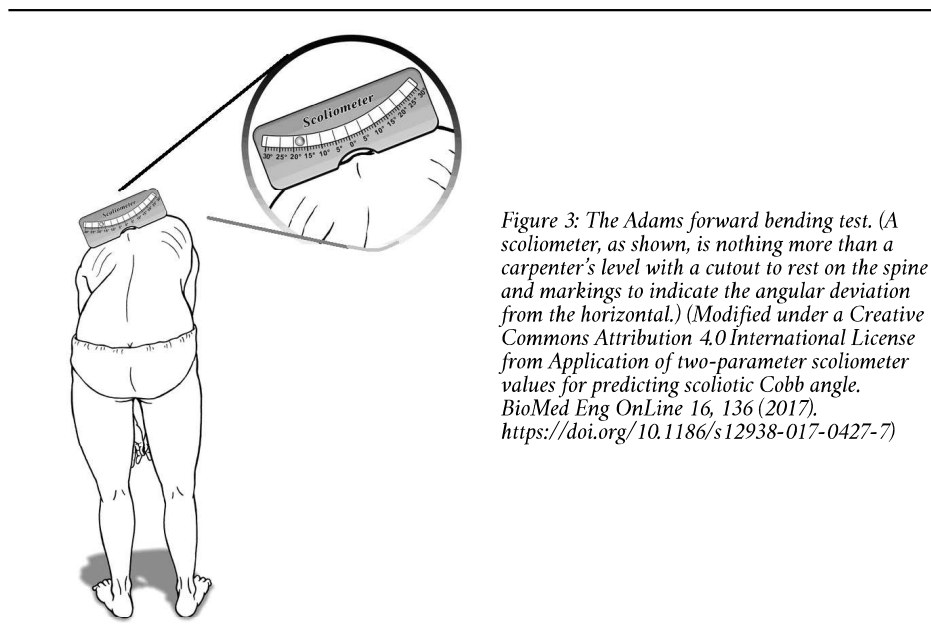


*Figure 3: The Adams forward bending test. (A scoliometer, as shown, is nothing more than a carpenter's level with a cutout to rest on the spine and markings to indicate the angular deviation from the horizontal.) (Modified under a Creative Commons Attribution 4.0 International License from Application of two-parameter scoliometer values for predicting scoliotic Cobb angle. BioMed Eng OnLine 16, 136 (2017). https://doi.org/10.1186/s12938-017-0427-7)*

## OBJECTIVE EVIDENCE

There are important radiographic parameters to help with diagnosis and management of adolescent idiopathic scoliosis:

- **Cobb angle:** The angle between intersecting lines drawn perpendicular to the two end vertebrae (see Figure 1).
- **End Vertebrae:** Top and bottom vertebrae maximally tilted into the concavity.
- **Neutral vertebra:** Vertebra that are not rotated. Identified by symmetric pedicles in the coronal radiograph.
- **Stable vertebra:** Most cephalad vertebra distal to end vertebra that is most closely bisected by the central sacral vertical line.
- **Apical vertebrae:** Central vertebra within a curve, typically least tilted and most rotated.
- **Risser sign:** Radiographic predictor of growth based on the amount of iliac crest apophysis ossification (Figure 4).
- **Sanders staging:** Radiographic predictor of growth based on hand x-ray (Figure 5). In general, the bone epiphysis in the hand fuse from distal (phalangeal bones) to proximal (distal radius) and thus supports a staging system that ranges from 1 (skeletally immature) to 8 (skeletally mature).
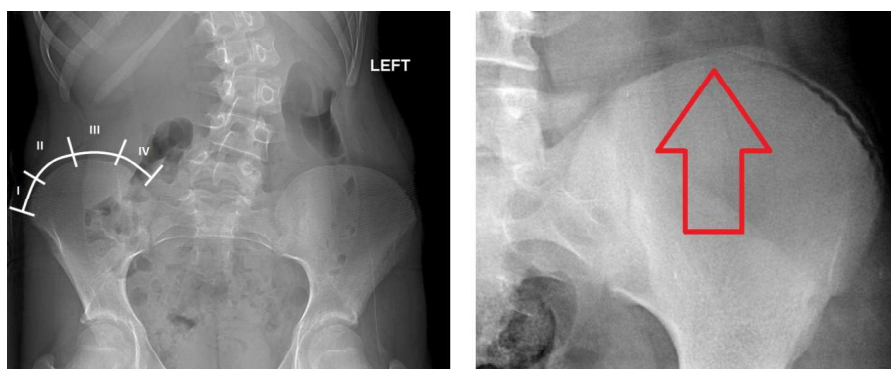
Figure 4: The Risser classification grades skeletal maturity based on the degree of ossification of the iliac crest apophyses. It begins with Risser Stage 0, where there is no ossification center at the level of iliac crest apophysis, and ends with stage 5, where there is complete ossification. Stages 1, 2, 3 and 4 are defined as 0 to 25% ossification, 25 to 50% ossification, 50 to 75% ossification, and 75% but less than 100% ossification, respectively. A Risser Stage 0 is shown in the x-ray at left, with a line indicating where the Stages 1 to 4 are defined. At right, a pelvic x-ray shows a Risser Stage 3 apophysis (Case courtesy of Dr Bruno Di Muzio, Radiopaedia.org, rID: 38297)
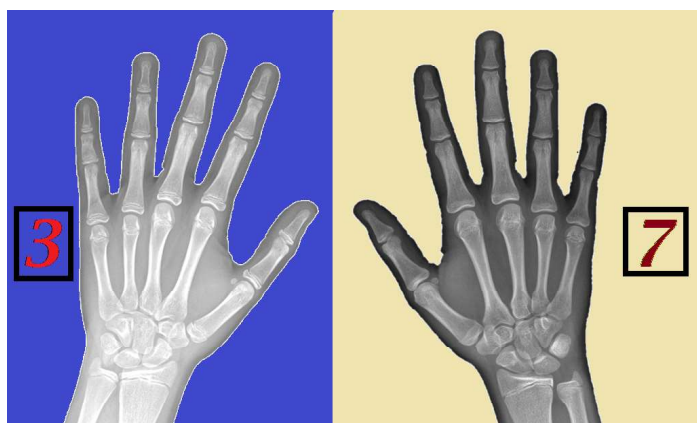


Figure 5: A hand x-ray corresponding to Sanders Stage 3 on the left and Stage 7 on the right. Note that in Stage 7, all physes are closed except for the distal radius physis, whereas the phalangeal and metacarpal physes are open in Stage 3.

## EPIDEMIOLOGY

Adolescent idiopathic scoliosis is relatively rare, with a prevalence of 3% in the general population. The prevalence of severe curves (>30) is much lower, approximately 0.03%. When considering only large curves, females are ten times more commonly affected than males, but the ratio is closer to 1:1 for cases with smaller curves.

There is thought to be a genetic component to adolescent idiopathic scoliosis, though the mode of inheritance is unknown. The risk of having adolescent idiopathic scoliosis is increased 50-fold when both parents have a history of scoliosis. In females whose mother has a curve >15º, the risk is 27%.

## DIFFERENTIAL DIAGNOSIS

So-called neuromuscular scoliosis is found when there are irregular spinal curvatures in the presence of the central nervous system or muscular disorders. In contrast to adolescent idiopathic scoliosis, neuromuscular curves tend to progress more rapidly, involve more vertebral levels, progress after maturity, and are usually associated with pelvic obliquity.

Scheuermann's Kyphosis is a spinal deformity causing rigid thoracic hyperkyphosis (>45 degrees). In general, adolescent idiopathic scoliosis is associated with "flat back," or loss of normal kyphosis and lordosis.

A leg length discrepancy can cause an apparent scoliosis. If present, leg length discrepancy will result in pelvic obliquity, and with it, a compensatory curve to keep the head and upper body over the center of the pelvis (see Figure 6).
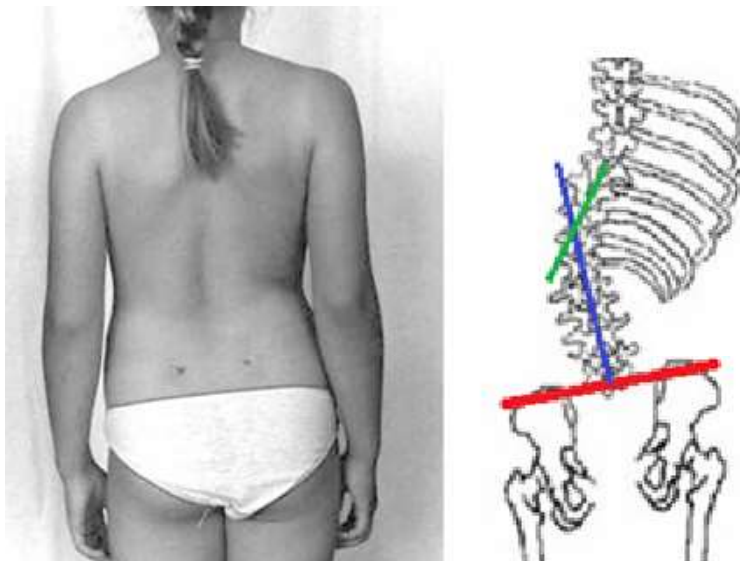


*Figure 6: At left, a photograph of a patient with pelvic obliquity. This is defined (in the diagram at right) by the red line drawn across the iliac crests. The lumbar spine is accordingly tilted as well (blue line), with a compensatory curve above (intersection of green and blue lines) to keep the head centered over the body. (modified from https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3282518/)*

## RED FLAGS

It is important to identify red flags during adolescent idiopathic scoliosis evaluation, which will warrant further investigation such as MRI to assess for intraspinal abnormalities such as syrinx, Chiari malformation, or cord tethering that may need to be addressed prior to curve correction. These include left thoracic curves (right thoracic curves are far more common), apical kyphosis (kyphosis is rare in adolescent idiopathic scoliosis), rapid progression of a curve, structural abnormalities such as a hemivertebrae, neurologic findings such as pathologic reflexes or radicular pain, and foot deformities.

## TREATMENT OPTIONS AND OUTCOMES

The plan of treatment depends on the severity of the curve and patient age. After achieving skeletal maturity, curves <30 degrees are unlikely to progress in adulthood. In contrast curves >50 degrees are likely to progress 1 degree per year; and even higher rates are seen during pregnancy and menopause.

Curves smaller than 20 degrees have a low probability of progressing and require only surveillance with periodic physical exams until skeletal maturity. (After skeletal maturity is reached, curves smaller than 20 degrees do not need close monitoring.)

Curves measuring more than 20 degrees on initial presentation require intervention. The goal in treating patients with curves of more than 20 degrees but less than ~50 degrees (see Figure 7) is to prevent progression – not correction. This goal can often be achieved with thoraco-lumbar bracing. A landmark multicenter, prospective, randomized trial known as the Bracing in Adolescent Idiopathic Scoliosis Trial (BrAIST) demonstrated a 72% success rate at preventing curve progression to 50 degrees or more compared to 48% in the observation cohort. Given the effectiveness of bracing, the trial was terminated early. Multiple societies, including the Scoliosis Research Society (SRS), Pediatric Othopaedic Society of North America (POSNA), American Academy of Orthopaedic Surgeons (AAOS), and American Academy of Pediatrics (AAP) all recommend bracing in cases of adolescent idiopathic scoliosis with growth remaining and curves between 20-40 degrees to prevent progression and avoid surgical intervention. Needless to say, patient compliance is key: brace therapy is effective only if the brace is worn.
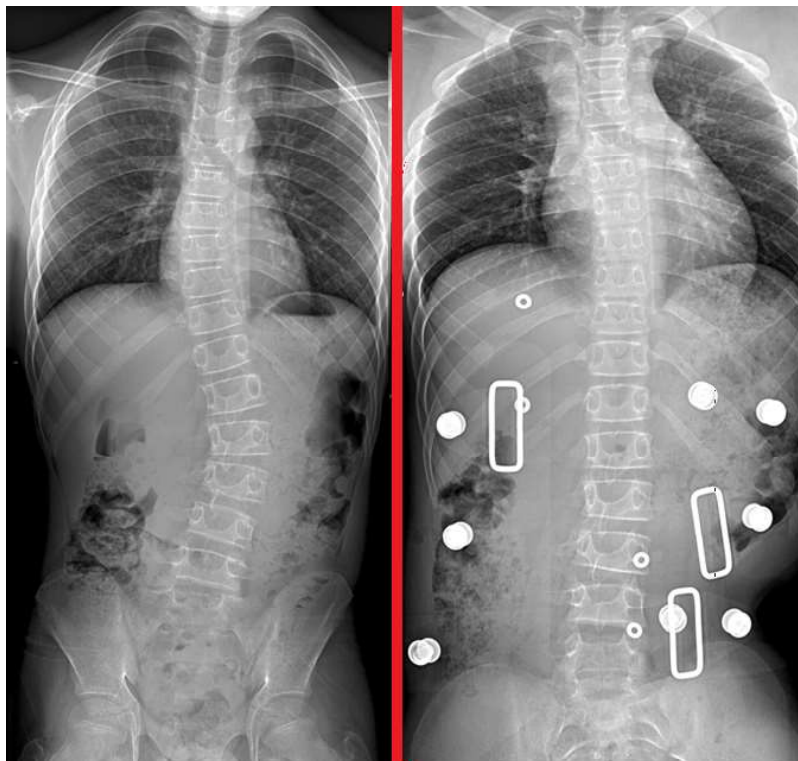


*Figure 7: Anteroposterior plain film radiograph of a patient with major lumbar curve of 36 degrees prior to bracing [left] and a radiograph of the same patient in brace demonstrating curve correction while in a brace [right].*

For patients with curves measuring more than 45 to 50 degrees, surgery may be the best option to correct the deformity and halt curve progression. In addition to the severity of the curve, it is ideal that patients be close to or have reached skeletal maturity.

(The precise indication for surgery is based on expert opinion. According to the website of the American Academy of Orthopaedic Surgeons in August 2020, "Most scoliosis surgeons agree that children who have very severe curves (45-50° and higher) will need surgery to lessen the curve and prevent it from getting worse.")

Surgical techniques are variable and continue to evolve with emerging technology, but the ultimate goal is to help correct deformity and achieve a solid fusion. A common technique used today is stabilization of the spine with pedicle screws and long rods until posterior spinal fusion is achieved (Figure 8).

*Figure 8: Surgical correction of scoliosis with a posterior spinal fusion, T4 to L3.*

Surgery is usually successful. In some patients who undergo posterior spinal fusion at a young age, the anterior column of the spine can continue to grow, creating a rotation deformity. This complication can be prevented by delaying surgery until skeletal maturity or performing anterior based spinal fusions to halt growth of the anterior vertebrae.

## RISK FACTORS AND PREVENTION

Currently, there are no known preventable risk factors for adolescent idiopathic scoliosis. As such, much effort has been focused on screening to help with early diagnosis and bracing to prevent the need for surgery.

A few studies have reported on the relationship with scoliosis and exercise and/or posture. Although healthy ergonomic posture and core strengthening are encouraged, there is no Level 1 evidence that demonstrates prevention or reversal of scoliosis based on posture and exercise or physical therapy programs.

Although adolescent idiopathic scoliosis is considered to be painless, residual curves (especially those measuring 50 degrees or more) may be associated with increased incidence of low back pain in adulthood.

## MISCELLANY

Scoliosis need not interfere with high level athletic performance. Usain Bolt (Figure 9), one of the fastest sprinters in history, reported in his autobiography that he has scoliosis.



*Figure 9: Usain Bolt (From Wikipedia https://commons.wikimedia.org/w/index.php?curid=7828074)*

## KEY TERMS

Scoliosis, Cobb angle, Risser/Sanders Assessment of Skeletal maturity, Bracing, Posterior spinal fusion

## SKILLS

Define scoliosis. Be able to measure the coronal plane Cobb angle on plain film radiographs. Identify coronal and sagittal plane global balance. Recognize red flags on history and physical exam that warrant advanced imaging with MRI in patients that present with Adolescent idiopathic scoliosis. Understand criteria for bracing and surgery based on curve severity and skeletal maturity.

# EARLY ONSET SCOLIOSIS

Scoliosis is defined as a three-dimensional deformity of the spine that includes not only a more noticeable lateral deviation, but also rotation of the vertebrae within the (lateral) curvature. Scoliosis can be further classified by the age of a patient. Early onset scoliosis is defined as scoliosis diagnosed before the age of 10 years old due to any cause (Figure 1). Early onset scoliosis can be categorized by etiology: namely, congenital, syndromic, neuromuscular and idiopathic. The scoliosis might be present without a known cause too; this is termed idiopathic scoliosis. Idiopathic scoliosis is rare in children under the age of 10. (Adolescent idiopathic scoliosis is discussed in its own chapter.)



*Figure 1: Clinical photograph of a two-year-old girl with severe thoracic early-onset scoliosis (Courtesy of Treatment strategies for early-onset scoliosis. EFORT Open Reviews. 3. 287-293. 10.1302/ 2058-5241.3.170051.)*

## STRUCTURE AND FUNCTION

Growth of the spine and the chest wall are critical for lung growth and development. Without normal growth, alveolar hypoplasia and disturbance of chest wall function causes a restrictive lung disease and possible respiratory failure.

The "golden time" for lung growth is from birth to the age of 5 years old and coincides with the period of most rapid growth of the thoracic spine and rib cage. Bronchial tree and alveolar complement are maximally developed by 8 years of age, and the thoracic volume at 10 years of age is 50% of the expected adult volume.

Patients with congenital anomalies causing their early onset scoliosis often have rib fusions or other anomalies that can disturb the normal biomechanics of chest wall, leading to decreased forced vital capacity (FVC). The term *thoracic insufficiency syndrome* (TIS) has been coined to describe the inability of the thorax to support normal respiration and lung growth.

Because the earlier that a spinal deformity appears, the greater the disturbance on thoracic cage, patients with early onset scoliosis have greater lung dysfunction and increased mortality rates compared to the general population, in proportion to the magnitude of the spinal deformity.

Early onset scoliosis can be categorized by etiology, as follows: congenital, syndromic, neuromuscular and idiopathic.

### Congenital early onset scoliosis

Congenital scoliosis (Figure 2) is defined as scoliosis caused by at least one bony anomaly of the spine which is present at birth (though the diagnosis may not be made until later in childhood). Congenital anomalies include failure of vertebral formation, such as hemivertebrae or wedged vertebrae, or failure of separation or segmentation, such as block vertebrae or unilateral bars. A unilateral unsegmented bar with a contralateral hemivertebrae is the anomaly known for the worst risk of progression of congenital scoliosis. A positive family history is found in 1% of patients with congenital spinal deformities, but a genetic cause has not been discovered.

The neural axis, vertebral column, and other organ systems develop simultaneously at about five to eight weeks of gestation. Thus, congenital scoliosis may be associated with an intraspinal anomaly such as a tethered cord, diastematomyelia (a longitudinal cleft in the spinal cord), diplomyelia (duplication of the spinal cord), syringomyelia (fluid-filled cavity in the spinal cord), or Arnold-Chiari malformation (herniated cerebellar tonsils at the foramen magnum). Because the vertebral malformations form during embryogenesis at the same time the heart and kidneys are also developing, about 60% of patients will have abnormalities affecting these systems. Accordingly, a renal ultrasound and echocardiogram is often obtained to evaluate for concomitant pathology.

A constellation of symptoms often diagnosed in patients with congenital scoliosis is the VACTERL syndrome, consisting of: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities. A patient must have at least three of the features to be diagnosed with VACTERL syndrome.



*Figure 2: A 3-dimensional reconstruction of a CT scan demonstrating congenital scoliosis with fused ribs. (Courtesy of Treatment strategies for early-onset scoliosis. EFORT Open Reviews. 3. 287-293. 10.1302/2058-5241.3.170051.)*

### Syndromic early onset scoliosis

Syndromic scoliosis is defined as scoliosis that appears in conjunction with a recognized pediatric syndrome, such as Marfan's, Ehlers-Danlos, trisomy 21 (Downs syndrome), Prader-Willi, Retts, neurofibromatosis, Noonan, or osteoporosis imperfecta.

When a child is diagnosed with a syndrome known to be associated with scoliosis, it is important that physicians screen for scoliosis. The natural history of scoliosis associated with these syndromes varies, so the curves they cause progress at different rates and require personalized treatment approaches.

### Neuromuscular early onset scoliosis

Neuromuscular scoliosis is defined as scoliosis caused by a neurologic disorder of the central nervous system or muscle, such as muscular dystrophy, cerebral palsy, spina bifida, spinal muscular atrophy, or Freidrich's ataxia.

The likelihood of developing neuromuscular scoliosis usually depends on the extent of nerve and muscle involvement with a specific disorder. Not all children with a neuromuscular disorder will develop scoliosis, but it is especially common in those who are not able to walk.

Neuromuscular scoliosis differs in particular from other types of scoliosis as the deformity is often a long, sweeping curve, typically involving the thoracic spine, the lumbar spine, as well as the pelvis.

### Idiopathic early onset scoliosis

Idiopathic scoliosis is defined as scoliosis without known cause; that is, when congenital, syndromic and neuromuscular causes have been excluded. Children with idiopathic early onset scoliosis are considered otherwise healthy.

Idiopathic scoliosis can be of the infantile type (diagnosed at less than 3 years of age). This is very rare. Juvenile Idiopathic scoliosis is that diagnosed between 3 years and 10 years of age. (The most common form of idiopathic scoliosis, the Adolescent type, refers to scoliosis occurring after 10 years of age in the setting of an otherwise healthy child. This, by definition, is not a type of early onset scoliosis.)


## PATIENT PRESENTATION

Evaluation of a patient with concern for early onset scoliosis begins with a comprehensive prenatal and birth history, as well as a detailed review of other medical conditions.

It is important that neurologic, pulmonary, urogenital, cardiovascular, and gastrointestinal systems have been evaluated if there is concern for congenital scoliosis.

If surgery is ultimately planned, evaluation should also include the current nutritional and pulmonary status of the patient.

A physical exam should start with measurements of height and weight. A full neurologic exam tests sensation, motor function and reflexes in the upper and lower extremities. Increased or decreased tone should be noted in patients with neuromuscular conditions. The skin should be examined for cafe-au-lait spots associated with neurofibromatosis, hair tufts or skin dimples near the sacrum associated with congenital scoliosis, and hypermobility associated with connective tissue disorders.

Coronal and sagittal balance is assessed by observing the patient from the front and the side, to see if they are tilted to one side (coronal imbalance), or have increased or decreased lordosis or kyphosis (sagittal imbalance). The examiner should also assess whether the torso is level, by palpating the iliac crests and the top of the shoulders.

The Adams forward bend test (Figure 3) has the patient bend at the waist, reaching toward their toes while the examiner stands behind the patient, looking at the back for asymmetric axial rotation of the trunk. If present, this will appear as a rib prominence on one side of the back. When the patient bends forward with the shoulders level with the hips, a scoliometer –a device similar to a carpenter's level, with a cutout to rest on the spine and markings to indicate the angular deviation from the horizontal– is laid to rest atop the most severe part of the deformity. A scoliometer reading of seven degrees or more warrants a referral to a pediatric orthopaedist.
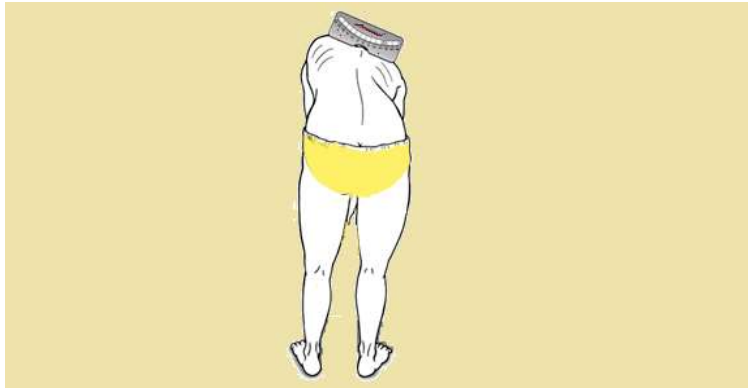
*Figure 3: The Adams forward bending test, showing a scoliometer resting on the child's back as she bends forward. As shown, the scoliometer is approximately 20 degrees off the horizontal, a clearly abnormal value. (Modified under a Creative Commons Attribution 4.0 International License from Application of two-parameter scoliometer values for predicting scoliotic Cobb angle. BioMed Eng OnLine 16, 136 (2017). https://doi.org/10.1186/s12938-017-0427-7)*

Note that performing the Adams forward bend test can be challenging in many children with EOS that are not independently ambulating; others may be unable to stay still for the scoliometer readings. In these cases, the spine exam can be performed while the child is sitting, with the examiner paying particular attention to shoulder or waistline asymmetry

## OBJECTIVE EVIDENCE

The diagnosis of scoliosis is established with a plain radiograph in the posteroanterior plane. A lateral radiograph is also useful for evaluating for sagittal plane abnormalities and where a diagnosis of scoliosis is suspected. These two views should be obtained in most cases.

Radiographs are important to the diagnosis of congenital scoliosis because bony anomalies can be identified that are causing scoliosis to occur. In the absence of congenital anomalies, plain radiographs are useful for assessing the severity of scoliosis and the risk of progression.

The Cobb angle (Figure 4) describes the angle of the spinal curvature for each curve present in the spinal column resulting in a scoliosis. A patient may have cervicothoracic, thoracic, thoracolumbar, or lumbar curve(s) depending on which region of the spine is involved in scoliosis.
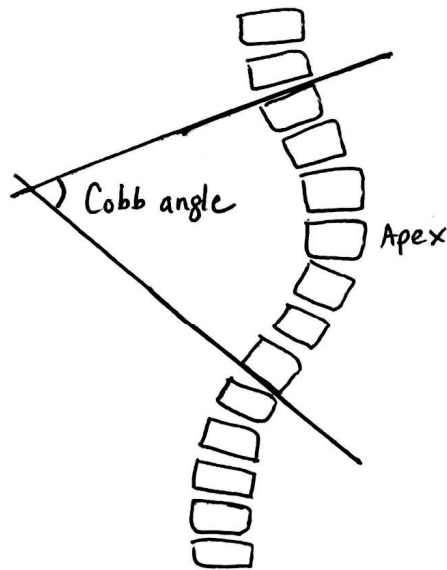
*Figure 4: The Cobb angle, as shown, quantifies the magnitude of the curve.*

To measure the Cobb angle, one must first decide which vertebrae are the end vertebrae of the curve deformity, which means the vertebra whose endplates are most tilted towards each other. Lines are then drawn along the endplates, and the angle between where the two lines intersect is the Cobb angle.

The vertebrae are also evaluated for the presence or absence of rib head overlap with the vertebral body at the apex of the curve (the vertebra that is located at the farthest point laterally from the midline of the body on the convexity of the curve). This assessment is termed the rib phase (Figure 5).

Phase 1 ribs do not overlap the vertebral bodies, while Phase 2 ribs do overlap the vertebral bodies. Patients with Phase 2 ribs are more likely to have progressive scoliosis, and therefore should be treated more aggressively.
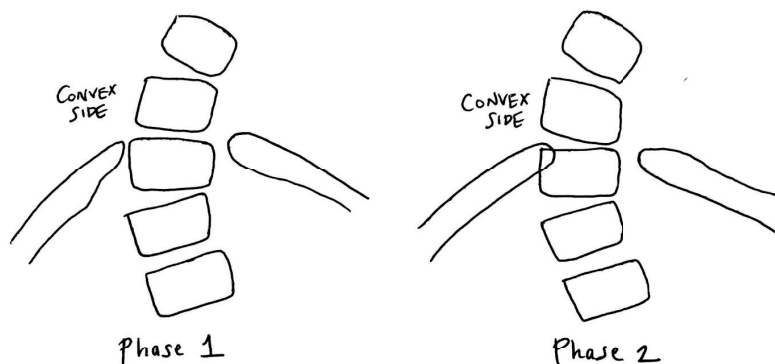


*Figure 5: Phase 1 ribs, as shown to the left, do not overlap the vertebral bodies, whereas Phase 2 ribs, at right, do overlap the vertebral bodies.*

An MRI of the entire spine is indicated for all patients with early onset scoliosis to evaluate for intraspinal abnormalities that may not yet be detectable based on clinical exam. About 20-40% of early onset scoliosis patients will have an abnormality, such as a tethered cord, Chiari malformation, or syrinx. A CT, especially with a three-dimensional reconstruction, can be useful to better delineate bony abnormalities and for surgical planning.

## EPIDEMIOLOGY

More than 100,000 children in the United States are diagnosed with scoliosis annually. The majority of those children are diagnosed with adolescent idiopathic scoliosis rather than early onset scoliosis. Because early onset scoliosis comprises a group of conditions with diverse etiologies and natural histories, the exact prevalence of early onset scoliosis is unknown. Idiopathic early onset scoliosis is estimated to account for less than one percent of all scoliosis.

## RED FLAGS

There are several "red flags" in the history, physical exam, and radiographs of a patient that should concern the treating provider.

An abnormal neurologic finding or a left thoracic curve can be predictive of an underlying pathologic condition of the spinal cord, although in early onset scoliosis, left thoracic curves are more common and less likely to be pathologic than in AIS.

A painful scoliosis should prompt suspicion for an osteoid osteoma, a benign bone tumor that in the spine is most commonly found in the thoracic and lumbar posterior elements and releases prostaglandin.

A rapidly-progressing scoliosis should raise concern for a tethered cord, also known as "tethered spinal cord syndrome." A tethered cord syndrome is present when there is a pathological development of excess fibrous connective tissue (fibrosis) in the filum terminale that fixes (or tethers) on the caudal spinal cord to the sacrum, limiting its movement. When a child grows, this abnormal attachment causes stretching and tension on the spinal cord. This can cause scoliosis, motor and sensory changes in the legs, back pain, foot deformities, and urinary dysfunction. Scoliosis can be one of the early signs of a tethered cord, so if there is any concern then an MRI should be ordered.

## TREATMENT OPTIONS AND OUTCOMES

Historically, the standard of care for severe, progressive early onset scoliosis was early definitive anterior and posterior spinal instrumented fusion. It was thought that a short but straight spine was superior to a long but curved spine, but now our understanding is that a well-aligned spine with a thoracic cavity large enough to support pulmonary development is the proper goal.

Nutrition is important in early onset scoliosis patients independent of treatment. Patients with severe spine deformity are often underweight, have poor soft tissue coverage over rib or spinal bony prominences/implants, and thus are a setup for complications. A nutrition consult can be helpful in advance of any treatment or at the time of the occurrence of wound complications in order to best optimize the patient pre-operatively or promote healing moving forward.

*Non-operative Treatment*

The first approach to a patient with early onset scoliosis is to use non-operative methods in an attempt to delay surgery. Some patients will in fact improve with non-operative treatment alone, such that later surgery is not needed. About 80% of children with idiopathic infantile scoliosis, for instance, will have resolution of scoliosis.

Serial casting and bracing have both been used. The concept of serial casting is based on the fact that growth can drive correction of the three-dimensional deformity. Patients with documented progression of scoliosis but low magnitude curvature (less than 60 degrees) or low risk for anticipated progression are candidates for serial casting.

A cast is applied on a special table that applies traction to the patient's head and legs while anesthetized. The traction distracts the facet joints so that there can be increased movement between vertebrae, then the cast

is applied with a mold over the ribs in order to rotate the spine and chest wall opposite the direction of the deformity. Windows are made in the cast anteriorly and posteriorly to allow room for normal expansion of the thoracic and abdominal cavities with respiration and eating. Casts are changed every 2 to 4 months based on age and growth and can be eventually replaced by a brace if the curve reduces to 10 to 20 degrees.

Outcomes of casting are best when casting is initiated at a younger age and when performed for less severe, idiopathic curves. Concerns and potential complications of casting include skin breakdown, negative effects on quality of life, repetitive anesthesia events for cast changes, and superior mesenteric artery or brachial plexus compression from the cast. Casts may not be tolerated in patients with poor pulmonary function or sensory disorders. Bracing can be similarly used to control non-congenital early onset scoliosis deformities with a goal of delaying surgery, or after serial casting is successfully completed. When a brace is used in a rapidly growing child younger than 5 years old, the patient may develop rib deformity or loss of the normal sagittal profile from pressure from the brace.

*Operative Treatment*

Currently, the concept of allowing continued growth of the spine and chest instead of performing a spinal fusion, while still managing spinal deformity, is of great interest in early onset scoliosis patients. Growth-sparing techniques have been classified into distraction-based techniques (meaning distracting posteriorly on the spine to lengthen the spine and enlarge the thoracic cavity), convex compression-based growth inhibition (meaning inhibiting vertebral body growth on the convexity of the curve to correct a curve), and guided growth (meaning directing growth to correct a curve over time). Spinal fusion is typically delayed until at least age 10, at which point the thoracic cavity is sufficiently developed to support the child through adult life.

Distraction-based techniques involve attaching rods to the spine or ribs proximally and to the spine or pelvis distally, while avoiding the spine in between the anchored segments. The rods are then distracted or lengthened by various mechanisms in order to produce growth of the spine and in turn the thoracic cavity. The options for distraction-based instrumentation include the "vertical expandable prosthetic titanium rib," also known as the VEPTR, (Figure 6); a "traditional growing rod" (TGR) (Figure 7); and the magnetically-controlled growing rod (MCGR) (Figure 8).
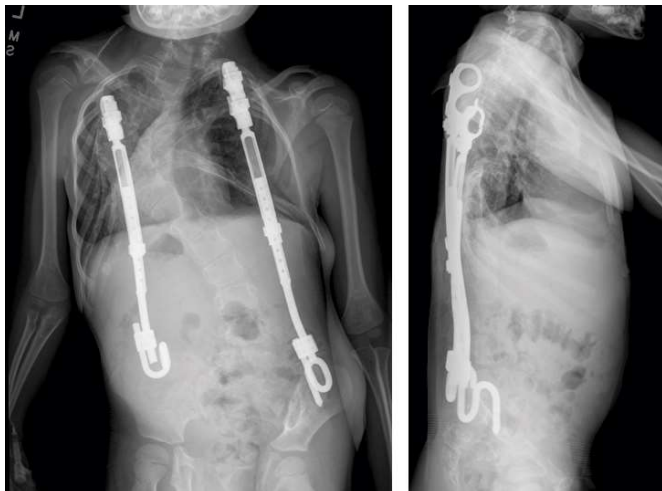


Figure 6: A patient with bilateral vertical expandable prosthetic titanium rib devices, VEPTRs, placed for congenital scoliosis and thoracic insufficiency syndrome. The VEPTR is a rib-distraction device that was designed specifically to address the thoracic insufficiency by expanding the thoracic cavity. As the child grows, VEPTR adjustment (under anesthesia) is performed every 6-8 months until the child reaches skeletal maturity.
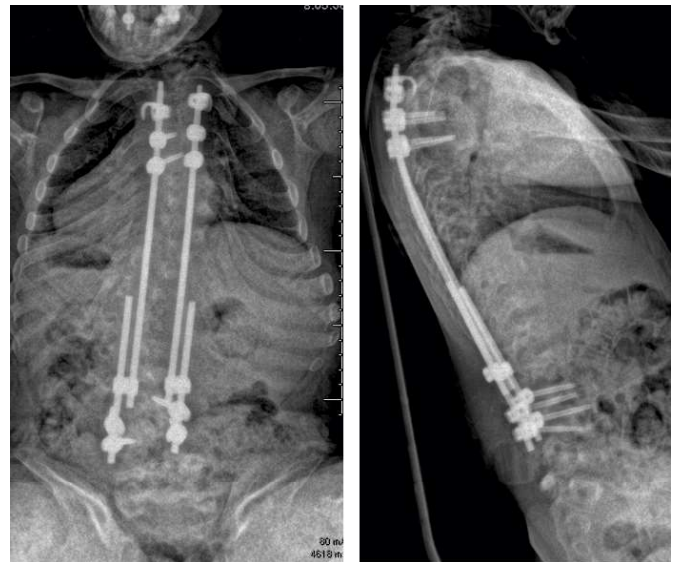
Figure 7: A patient with bilateral traditional growing rods. In the technique, a combination of hooks, wires and screws are anchored both proximally and distally to the spine via a short fusion and are connected by tunneled rods beneath the soft tissue. This technique must be distracted surgically under anesthesia as well.

*Figure 8: A patient with a unilateral magnetically-controlled growing rod, with rib anchors proximally and spine anchors distally. Magnetically-controlled growing rods are similar to traditional growing rods, with the notable difference being that the anchors can be distracted using a magnet in the out-patient setting.*

Complications of these techniques include implant prominence which can eventually result in skin breakdown, surgical site infection, implant breakage, implant migration, and failure to lengthen, all of which result in return trips to the operating room and additional anesthetic events. Repeat lengthening surgery is problematic as with each additional surgical procedure the likelihood of a complication rises in an already risky/sick patient population. (This makes the MCGR so appealing: it helps avoid trips to the operating room.)

Convex compression-based growth inhibits deformity progression by applying a compressive force on the convex side of the scoliosis deformity.

Guided growth(Shilla system) relies on fixation at the apex of a scoliosis curve with gliding fixation proximally and distally so that the spine can continue to grow above and below while gradually correcting the scoliosis. Osteotomies are performed at the apex to correct as much deformity as possible and pedicle screws are placed at the apex over three or four vertebral segments.

Resection and short fusion can be the treatment methodology employed when attempting to manage specific types of congenital scoliosis. A hemivertebra causing a progressive scoliosis in a very young child may be completely resected with a short (2-level) spinal fusion using pedicle screws/hooks to correct deformity and minimize the need for repeat surgical procedures. However, there are risks of neurological injury with hemivertebra resection, thus the decision to resect versus growth-sparing techniques may be a complicated one.


## KEY TERMS

Early onset scoliosis, congenital scoliosis, neuromuscular scoliosis, syndromic scoliosis, idiopathic scoliosis, casting, growth-sparing spine surgery


## SKILLS

Name the etiologies of early onset scoliosis. Understand the importance of spinal growth on thoracic cavity growth and pulmonary development. Describe the Adams forward bend test and how to use a scoliometer. Describe VACTERL syndrome. Understand the work-up, associated conditions or syndromes, and red flags important for a patient with concern for early onset scoliosis. Understand the measurements used in radiographic assessment of early onset scoliosis and measure the Cobb angle in plain x-rays.

# SCHEUERMANN'S KYPHOSIS

Scheuermann's kyphosis is a rigid sagittal plane deformity within the thoracic, thoracolumbar, or lumbar spine. The cause of Scheuermann's kyphosis (often named as "Scheuermann's disease," or simply "Scheuermann's") is not known. This condition is characterized by uneven growth of the vertebrae in the sagittal plane. Posterior overgrowth results in wedging of the vertebrae and a rounded, hunched back that does not fully correct with active extension. Approximately 50% of cases of Scheuermann's kyphosis are associated with back pain.

## STRUCTURE AND FUNCTION

Kyphosis refers to a convex curvature of the spine; the corresponding term, lordosis, refers to a concave curvature (see Figure 1). Normal thoracic kyphosis averages approximately 35 degrees with the cervical and lumbar spine both being lordotic.
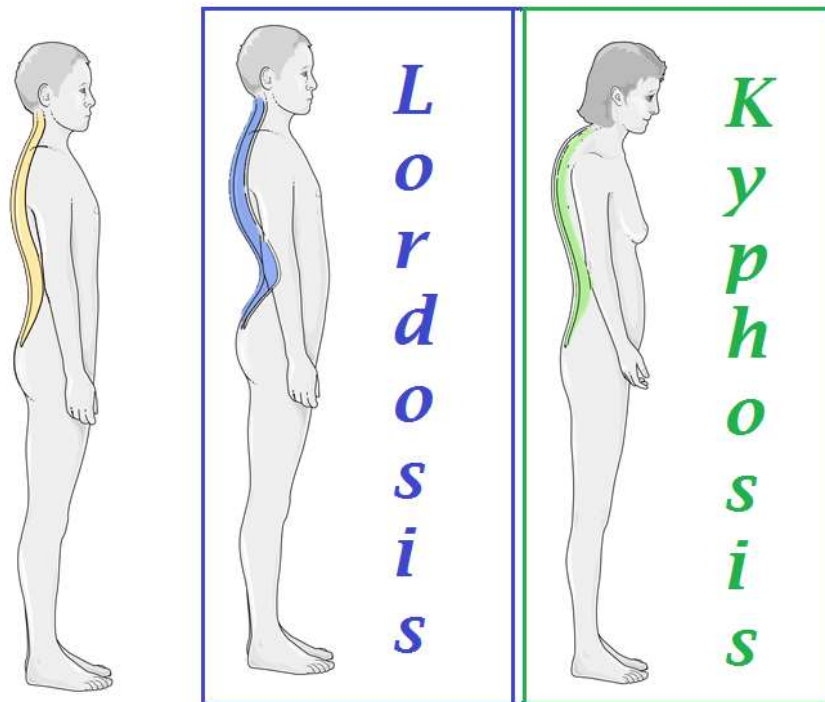


Figure 1: Normal sagittal plane alignment is shown at left, with excessive lordosis (blue arrow) and kyphosis (green arrow) shown in the center and right panels, respectively. (Image modified from Wikipedia)

Scheuermann's kyphosis is an increased amount of kyphosis (>45 degrees) and is defined radiographically. (Because some kyphosis is normally present, technically speaking this condition should be called "Scheuermann's hyperkyphosis.") The radiographic definition is a patient having three or more contiguous vertebrae with at least 5 degrees of anterior wedging. There are vertebral endplate abnormalities resulting in disc space narrowing that may be caused by an error in collagen aggregation. Schmorl's nodes, which are disc herniations into the vertebral endplate, are present.

The true etiology of Scheuermann's kyphosis is unknown but multiple theories exist. Scheuermann's theory was that the growth disturbance is due to osteonecrosis of the vertebral ring apophysis. This causes a growth arrest in the anterior vertebral body and radiographically is depicted by wedging. Schmorl's theory suggests disc material herniating through the vertebral end plate causes loss of height & ultimately the anterior wedging. Growth hormone abnormalities could possibly be a causative factor. Relative osteoporosis can lead to a compression deformity, thus causing the anterior vertebral wedging and increased kyphosis. There are possible genetic causes, as there is a high rate of heritability and an autosomal dominant inheritance pattern.

## PATIENT PRESENTATION

The most evident clinical manifestation of Scheuermann's kyphosis comes with the physical exam. Patients will have increased kyphosis that is exacerbated with bending forward, (see Figure 2). The patients may also have compensatory hyperlordosis of the lumbar or cervical spine. This can lead to tightness in the hamstrings and iliopsoas muscles. Patients with more severe curves also have a higher incidence of back pain. Neurologic deficits, while rare, require a full neurologic exam.



*Figure 2: Clinical photograph of a 22-year-old male with a very extreme case of Scheuermann's disease. (courtesy Wikipedia)*

## OBJECTIVE EVIDENCE

After a patient's physical exam is worrisome for Scheuermann's kyphosis, an AP and lateral spine radiograph should be obtained (Figure 3).

*Figure 3: Lateral radiographs of a patient with Scheuermann's kyphosis (Case courtesy of Dr Bruno Lorensini, Radiopaedia.org, rID: 43740)*

On the AP radiograph, the presence or absence of scoliosis should be noted. Lateral radiographs must be scrutinized to detect spondylolysis and spondylolisthesis.

Scheuermann's kyphosis is defined by a rigid thoracic hyperkyphosis greater than 45 degrees, associated with anterior wedging of three consecutive vertebrae measuring at least 5 degrees per vertebral body (Figure 4).



*Figure 4: Wedging of the vertebral bodies. (Case courtesy of Dr Dalia Ibrahim, Radiopaedia.org, rID: 58862)*

Disc space narrowing, endplate irregularities, and Schmorl's nodes can be noted on the lateral radiograph. Endplate irregularities are more common in thoracolumbar and lumbar Scheuermann's kyphosis compared to vertebral wedging.

Sagittal balance can be noted on the lateral radiograph by using the C7 plumb line and the posterior sacral vertical line.

The entire spine must be included on the x-ray as Scheuermann's kyphosis can extend all the way to the thoracolumbar regions. Thoracolumbar Scheuermann's is a far less common form, but is associated with increased back pain and more likely to be progressive.

A supine hyperextension lateral radiograph over a bolster may be obtained to differentiate Scheuermann's kyphosis from postural kyphosis. As opposed to postural kyphosis, Scheuermann's kyphosis is relatively inflexible on the hyperextension lateral radiograph.

MRI may be obtained at the discretion of the surgeon to identify disc herniations, spinal cord abnormalities, spondylolysis, spondylolisthesis, and spinal stenosis, among other spinal abnormalities. Any neurological deficit or symptom should be evaluated with an MRI.

## EPIDEMIOLOGY

Scheuermann's kyphosis affects men and women equally, with a prevalence between 0.4%-10%. Scheuermann's kyphosis is the most common type of structural kyphosis in adolescents with a typical onset between the ages of 10-12. Thoracic Scheuermann's kyphosis is by far the most common, which is classified as a curve apex between T6-T8.

## DIFFERENTIAL DIAGNOSIS

The main differentiation that must be made is between Scheuermann's kyphosis and postural kyphosis. Postural kyphosis will correct with extension, and radiographs will reveal the absence of anterior vertebral wedging.

Severe hyperkyphosis can be caused by vertebral compression fractures; this would be on the differential diagnosis for older females much more than in the pediatric or adolescent male population normally affected by Scheuermann's.

Congenital kyphosis may be found if vertebrae are malformed or fused. A congenital kyphosis in the absence of neurological disorders is rare.

Nutritional kyphosis can result from rickets, usually due to a vitamin D deficiency.

If there is lower back pain, spondylolysis and spondylolisthesis must be ruled out.

## RED FLAGS

Any neurologic deficit or complaint should be evaluated with a full physical examination, radiograph, and advanced imaging. Back pain along with constitutional symptoms should be investigated fully. Spinal deformity may happen in the setting of malignancy, therefore imaging should be thoroughly evaluated by a radiologist as well as the ordering physician.

## TREATMENT OPTIONS AND OUTCOMES

The treatment of mild to moderate (less than 50-80 degrees) Scheuermann's kyphosis is non-operative. Non-operative treatment includes stretching and physical therapy with routine radiographic follow up. Therapy and exercises include postural improvement, thoracic extensor strengthening, and core strengthening.

Bracing with an extension orthosis has been attempted (Figure 5). Bracing requires significant patient compliance with brace wear of 16-23 hours per day. As is the case with scoliosis, bracing is chosen not so much to affect a correction but to stop progression; thus, it is indicated only if there is remaining skeletal growth anticipated.



*Figure 5: A thoraco-lumbar brace. (Photo modified from Wikipedia)*

Operative treatment should be considered only in severe curves. Other indications for surgery include progressive deformity, neurologic deficit, spinal cord compression, and severe pain. Although there is no precise Cobb angle above which surgery is recommended, curves of 80 degrees or more in patients who have failed nonoperative treatment are usually indicated for surgery.

Surgery includes posterior instrumented spinal fusion with pedicle screw fixation (Figure 6). Anterior surgery is much less common with improvements in posterior surgical intervention. With severe, rigid curves posterior (Ponte) osteotomies may be performed. About 5 to 10 degrees of correction is anticipated with each posterior osteotomy performed. More aggressive osteotomies (pedicle subtraction or vertebral column resection) can be used with increased risk of complications. Intraoperative neuromonitoring is the standard of care in patients with spinal deformity.



*Figure 6: Clinical photos and lateral x-rays of a patient with Scheuermann's kyphosis before and after posterior instrumented spinal fusion. (Courtesy Surgical treatment of Scheuermann's disease by the posterior approach. Case series. Coluna/Columna, 14(1), 14-17. https://doi.org/10.1590/S1808-1851201514010R120)*

Patients with curves less than 60 degrees typically have a benign course and a good clinical outcome with observation alone. Patients with kyphosis greater than 100 degrees have clinically significant impairment in pulmonary function.

The Scoliosis Research Society and Harms Study Group both reported an overall complications rate of approximately 15% in Scheuermann's Kyphosis.

Neurologic complications with posterior spinal fusion have a reported rate of 0.6-0.8%, which is slightly higher than the rates in idiopathic scoliosis. There is conflicting evidence on whether combined anterior/posterior procedures have higher neuromonitoring changes compared to posterior only procedures.

Proximal and distal junctional kyphosis are complications that can be mitigated by selecting appropriate fusion levels, avoiding overcorrection >50% of original curve, construct choice, and correcting sagittal balance.

## MISCELLANEOUS

While the natural history of Scheuermann's kyphosis tends to be benign, patients are more likely to pursue jobs that require less strenuous physical activity. Also, there is a cosmetic deformity. In all, there are important psychosocial considerations beyond the medical aspects of the condition.

## KEY TERMS

Kyphosis, Schmorl's node

## SKILLS

Evaluate lateral x-rays of the spine and identify abnormal alignment. Identify signs of abnormal spinal alignment.

PART III.

# MUSCULOSKELETAL ASPECTS OF PEDIATRIC SYNDROMES

# DOWN SYNDROME

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Down syndrome, or Trisomy 21, is the most common genetic disorder caused by a chromosomal abnormality. The presence of three copies of chromosome 21, rather than the usual two, leads to characteristic facial features, developmental delay, intellectual disability, and impaired immune function. Down syndrome is also associated with an increased risk of congenital heart defects, epilepsy, leukemia, and other diseases. Approximately 20% of all patients with Down syndrome have an associated musculoskeletal condition. The main effects of Down syndrome on the musculoskeletal systems are due to ligament laxity, excessive joint flexibility, and loss of muscle tone. Beyond generalized laxity and hypotonia, musculoskeletal manifestations of Down syndrome include cervical spine instability, hip subluxation, patellofemoral instability, scoliosis, and flatfoot deformity (also known as pes planus or pes planovalgus).

## STRUCTURE AND FUNCTION

Trisomy 21 is usually due to duplication of one of the parental copies of chromosome 21 (most commonly the mother's). This occurs due to failure of the diploid chromosome to separate into haploid gametes during meiosis (termed "nondisjunction"). In rare cases, Down syndrome is due to a chromosomal translocation.

The ligament laxity and excessive joint flexibility seen in Down syndrome likely arise from the production of abnormal or excess Type VI collagen. The production of Type VI collagen is encoded by a gene found on the 21st chromosome.

There is an increased incidence of polyarticular rheumatoid factor-negative arthritis in children with Down syndrome. This typically causes erosive changes of the small joints of the hands and wrists. The mechanism of this arthropathy is not known.

## PATIENT PRESENTATION

Growth in children with Down syndrome is commonly delayed and stunted, as are many other developmental milestones. The typical facial appearance includes flattening of the nasal bridge, with upward-slanting eyes and epicanthal folds. The mouth is typically small with a large tongue, which commonly leads to obstructive sleep apnea. The neck may be stout or shortened, often with redundant skin posteriorly. The hands are shortened with a single palmar crease (Figure 1). The feet are usually flat (pes planus) and may have an unusually large space between the 1st and 2nd toes (Figure 2). Classically, the gait is waddling and broad based.
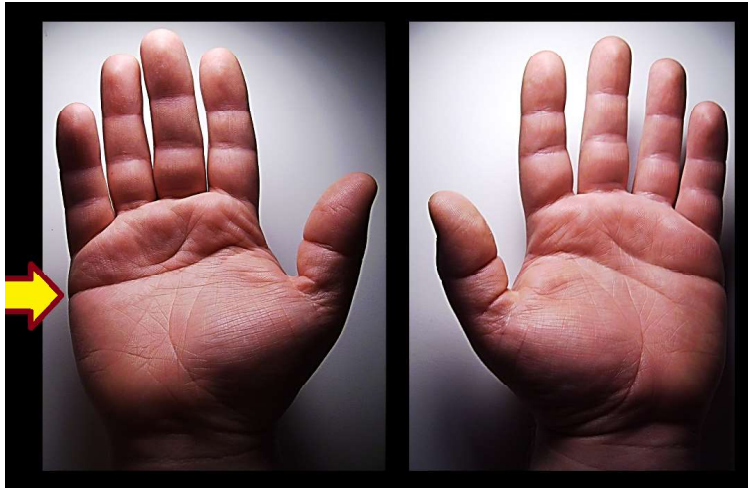
*Figure 1: Shown at left is a hand with a single palmar crease (yellow arrow); a normal hand crease pattern is shown at right. (image courtesy Wikipedia)*



*Figure 2: Clinical photograph of the feet of a boy with Down syndrome, showing a large gap between the first and second toes bilaterally. (image courtesy Wikipedia)*

Most patients with Down syndrome have some degree of intellectual disability. Hearing and vision problems are fairly common; the incidence of epilepsy is also elevated.

Congenital heart disease is common in individuals with Down syndrome. This usually takes the form of a septal defect, although valve pathology may be seen. More serious defects can include tetralogy of Fallot. Hypothyroidism, type 1 diabetes mellitus, duodenal atresia, and leukemia are also seen at higher rates in patients with Down syndrome. Additionally, the overall occurrence of infection is greater, leading to elevated surgical site infection risk as compared to the general population.

Approximately 20% of all patients with Down syndrome have an associated musculoskeletal condition. Occipito-cervical and atlanto-axial instability are common. Atlantoaxial instability can be assessed on lateral cervical spine radiographs or sagittal CT scans by measuring the atlanto-dens interval (Figure 3). This interval should be 5mm or less.
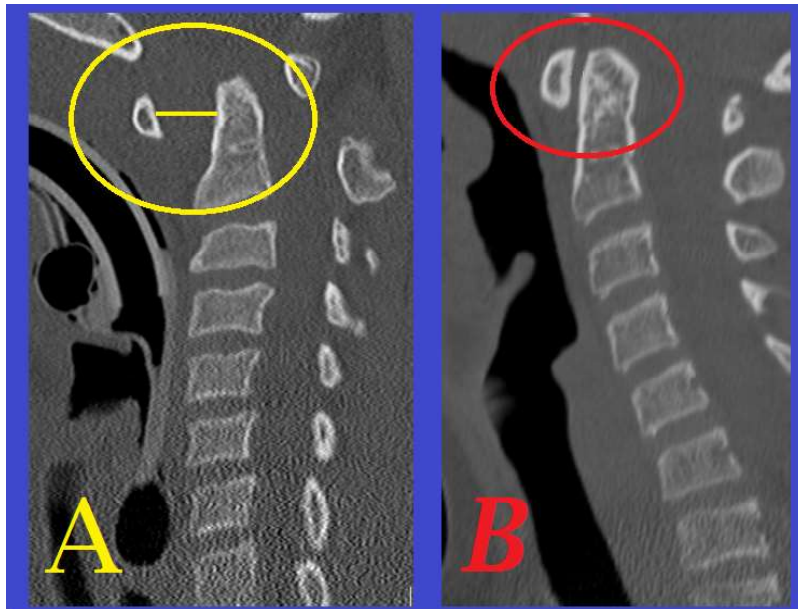
*Figure 3: Mid-sagittal slice of a cervical spine CT scan. The image on the left (A) is of a 14-year-old boy with Down syndrome who was experiencing myelopathic symptoms due to atlantoaxial instability. The atlanto-dens interval, indicated by the yellow line, is ~13mm. The image on the right (B) is of a normal C-spine with a normal atlanto-dens interval for comparison.*

Down syndrome is associated with high rates of scoliosis and slipped capital femoral epiphysis (Figure 4).

Hypotonia and ligamentous laxity commonly lead to hip instability and recurrent patellofemoral dislocation in these individuals (Figure 5).
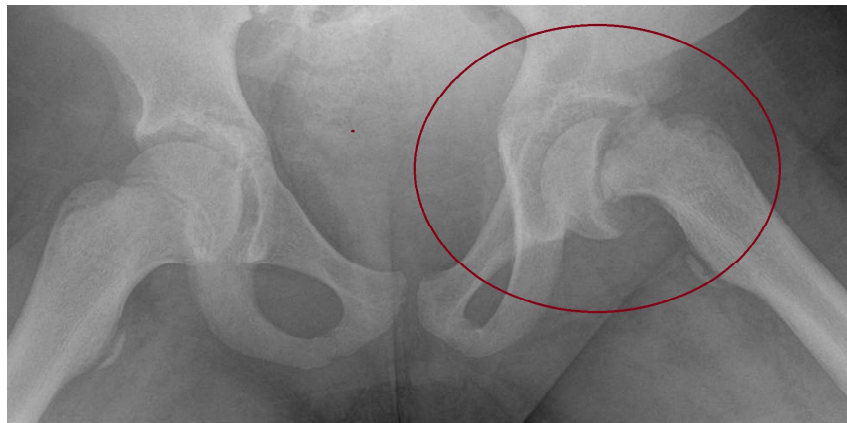


*Figure 4: Frog-leg lateral radiographic view of the bilateral hips in a patient with Down syndrome, demonstrating a left slipped capital femoral epiphysis.*

*Figure 5: Anteroposterior radiographic view of the left knee in a patient with Down syndrome, demonstrating lateral dislocation of the patella (red arrow).*

## OBJECTIVE EVIDENCE

Chromosomal analysis in Down syndrome reveals three copies of chromosome 21, which is diagnostic for this syndrome.

On an AP pelvis radiograph, the iliac wings are commonly flared, with flat acetabula. Prior to the advent of chromosome analysis for diagnosis, the pelvis radiograph was often used to confirm the diagnosis of Down syndrome.



*Figure 6: Pelvic radiograph showing "flaring" of the iliac wing, outlined in red, with the normal contour outlined in yellow. (Case courtesy of Radiopaedia.org, rID: 43579)*

## EPIDEMIOLOGY

Down syndrome is the most common chromosomal abnormality, occurring about once per 700 to 1000 live births. This occurrence, however, increases with the mother's age at conception. Down syndrome does not have a predilection for certain races or geographic regions. Life expectancy for patients with Down syndrome averages around 50-60 years. Individuals with Down syndrome also suffer premature aging, with a high prevalence of Alzheimer's disease in those who reach mid-adulthood.

## DIFFERENTIAL DIAGNOSIS

Diagnosis can usually be made based on the typical phenotypic characteristics described above. However, other conditions exist that may overlap phenotypically including congenital hypothyroidism, trisomy 18 and

Zellweger syndrome. When the diagnosis is in question, a chromosomal analysis may provide confirmation.

## RED FLAGS

Down syndrome is itself a red flag for atlantoaxial instability. This must be considered before any surgical procedure requiring intubation is contemplated. Many clinicians recommend radiographic evaluation of the cervical spine before patients with Down syndrome are cleared to play sports, though this is not universally accepted.

When evaluating a Down syndrome patient with a limp or vague lower extremity pain, pelvis radiographs must be studied closely for a subtle slipped capital femoral epiphysis of the hip. This condition is otherwise easily missed, as these patients may have trouble articulating their symptoms.

## TREATMENT OPTIONS AND OUTCOMES

When treating musculoskeletal manifestations in Down syndrome, non-operative modalities should be exhausted prior to surgical intervention as complication rates tend to be higher in this population.

Pes planovalgus and patellar subluxation can be treated initially with stabilization braces and physical therapy. Hip subluxation can be treated initially with hip abduction braces. When non-operative treatments fail, any surgical strategy must consider that patients with Down syndrome have generalized hypotonia and ligamentous laxity; thus, typical soft-tissue stabilization procedures alone are often ineffective. Osteotomies, such as tibial tubercle osteotomies for patellar instability or acetabular osteotomies for hip dislocation, are needed to address the generalized hypotonia and ligamentous laxity typically encountered.

If a slipped capital femoral epiphysis occurs in a Down syndrome patient, pinning the unaffected side prophylactically is usually undertaken at the same time.

Treatment of various degrees of atlantoaxial instability is controversial. Although the normal atlanto-dens interval is 5 mm or less, asymptomatic patients with an interval measuring between 5–10 mm can simply be observed initially. Surgical fusion and stabilization should be reserved for when symptoms develop.

Fusion options include C1-C2 fusion, or even occiput-C2/C3 if greater instability exists. However, it should be cautioned that complication rates in Down syndrome patients following fusion are high; overall complication rates can approach 50%, and mortality rates are quoted as high as 25%.

The spinal curvature that occurs in many patients with Down syndrome often resembles that of idiopathic scoliosis and treatment is similar: bracing and observation for small curves and surgical fusion for larger curves (Figure 7).
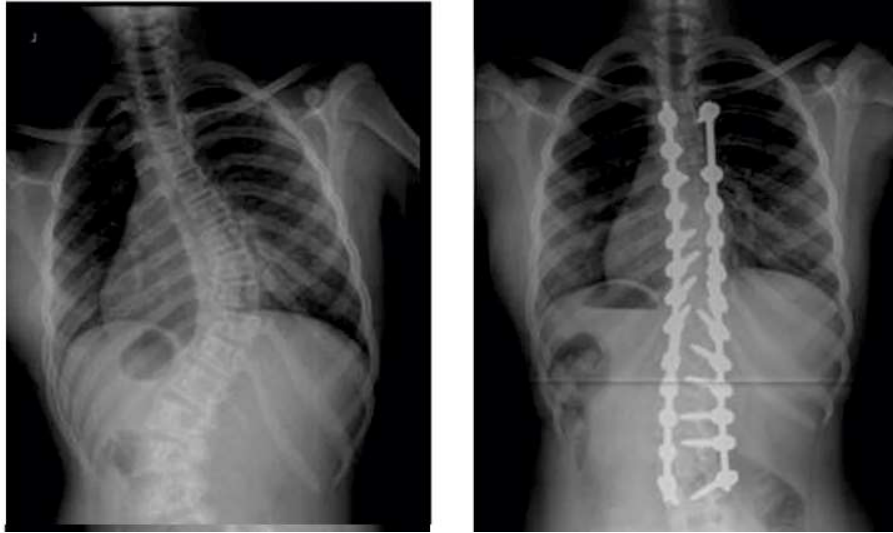


*Figure 7: preoperative (left) and post-operative (right) views of a patient with Down syndrome and scoliosis. (image courtesy Scoliosis 10, 14 (2015). https://doi.org/10.1186/s13013-015-0035-x)*

Although life expectancy for individuals with Down syndrome is shorter than normal, many patients routinely live into their 6th decade of life. Reasons for earlier mortality include cardiac disease and infections.

## RISK FACTORS AND PREVENTION

The most well-known risk factor for Down syndrome is advanced maternal age. This risk is < 0.1% when the mother is < 30 years old at conception, and increases more than 10-fold as the mother reaches her late 30s to early 40s. Parents of a child with Down syndrome are at increased risk of a subsequent child being affected. There is no known modifiable risk factor, other than maternal age, at which prevention can be aimed. For rare cases of Down syndrome due to a translocation, parents who carry the translocation are at an increased risk of having another child with the condition.

## MISCELLANY

Down syndrome is named after John Langdon Down, who first described the condition in 1862. However, it wasn't until the late 1950s that the genetic cause of the condition was identified, after the advent of technology that made karyotyping possible.

# OSTEOGENESIS IMPERFECTA

Osteogenesis imperfecta is a connective tissue disease characterized by extremely fragile bones due to an autosomal dominant genetic defect in type 1 collagen production. There are four main types of osteogenesis imperfecta: type I is the most common and the mildest form of the disorder, and is caused by an inadequate production of type 1 collagen. The remaining types are characterized by abnormal type 1 collagen formation. In addition to bone disorders, osteogenesis imperfecta is associated with blue sclera (Figure 1); lax ligaments; hearing impairments; facial abnormalities; short stature; scoliosis or kyphosis; and dental abnormalities.



Figure 1: The classic "blue sclera" of osteogenesis imperfecta (courtesy Wikipedia).

## STRUCTURE AND FUNCTION

Osteogenesis imperfecta is a disease caused by a genetic mutation that causes abnormal type 1 collagen cross-linking. Patients either have insufficient production of type 1 collagen or they produce an abnormal version of type 1 collagen.

The lack of normal type 1 collagen leads to insufficient production of osteoid (the organic matrix of bone). The bones in osteogenesis imperfecta are brittle and fracture easily (hence the synonym "Brittle Bone Disease"). In some cases, the bones may be bowed or otherwise severely deformed.

Four main types of osteogenesis imperfecta have been identified. Osteogenesis imperfecta type I is the most common and mildest form; type II is the most severe. In most cases, osteogenesis imperfecta is inherited in an autosomal dominant pattern. Type III patients are most commonly seen in clinics because of their frequent fractures and need for surgery.

Although the name "osteogenesis imperfecta" implies imperfection (to say the least) in the bones, the disease can in fact affect all connective tissues in which type 1 collagen is found. Clinically, patients usually present with ligamentous laxity. Other clinical findings include basilar invagination of the skull (which may cause neurological symptoms), blue sclera, and abnormalities of the teeth (termed "dentinogenesis imperfecta"). Patients can also have structural abnormalities in and around the heart such as mitral valve prolapse and aortic dissection.

In the vast majority of cases, osteogenesis imperfecta results from a mutation in the COL1A1 or COL1A2 genes which encode proteins involved in the formation of type 1 collagen.

Mature collagen contains a large triple helix region in which two alpha1 chains (from the COL1A1 gene) and one alpha2 chain (from COL1A2) assemble together to form this triple helix. Normally, in the triple helix region of the chains, glycine occurs every third amino acid position in the chain in a glycine-X-Y pattern, where X and Y are frequently proline or hydroxyproline (Figure 2). This pattern allows the collagen fibrils to coil into the triple helix and helps define the unique biochemical properties of the protein.
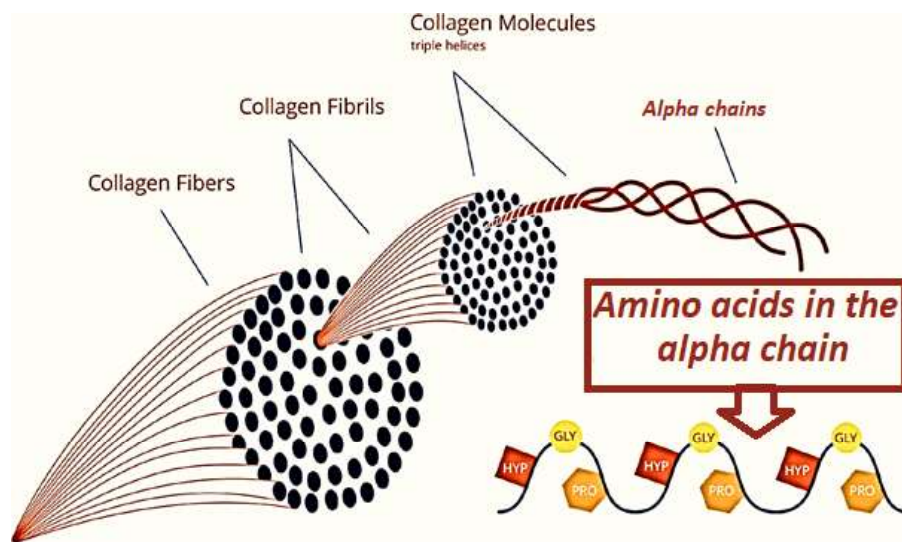


*Figure 2: Multiple collagen fibrils form into collagen fibers. Amino acids on the alpha chain shown here are proline (PRO), glycine (GLY) and hydroxyproline (HYP). Substitution of the glycine by a larger amino acid, as in osteogenesis imperfecta, inhibits the formation of the normal helix. (Courtesy of Journal of Children's Orthopaedics https://doi.org/10.1302/1863-2548.13.180190)*

Glycine is the simplest amino acid, with its side chain simply consisting of a hydrogen. With this simplicity, having glycine occur at every third spot in the amino acid chain allows the alpha chains to form a tight "twist." This spatial arrangement wouldn't otherwise be possible if an amino acid with a bulkier side chain were substituted for glycine.

In the most well-studied genetic abnormality in osteogenesis imperfecta, a point mutation in the COL1A1 and COL1A2 genes leads to the substitution of another amino acid in place of glycine. This impedes the correct association of alpha chains when forming the triple helix. This abnormal protein is then either hydrolyzed by the cell or simply fails to function properly.

Both autosomal dominant and autosomal recessive forms of the disease have been described.

## PATIENT PRESENTATION

Any organ system in which type 1 collagen plays a structural role can be affected. The severity and phenotype vary widely depending on the type of osteogenesis imperfecta. While four types of osteogenesis imperfecta have been classically described (see Table 1), as many as 15 more have been added since.

| Type | Description |
|------|-------------|
| I | · Most common type<br>· Least severe type<br>· Blue sclerae |
| II | · Lethal (respiratory compromise); most do not survive past early infancy |
| III | · Most sever form that is compatible with life<br>· Early and numerous fractures leading to deformed limbs<br>· Progressive in nature<br>· Normal sclerae |
| IV | · Not as severe as Type III<br>· Fractures are common<br>· Bowed long bones<br>· Normal Sclerae |

Classically, patients with osteogenesis imperfecta have diminished growth. The face may have an atypical triangular shape, and the eyes may have blue sclera. The teeth are commonly affected, termed "dentinogenesis imperfecta," due to abnormal dentin. This condition can lead to discolored, soft teeth with a predilection for dental caries. Hearing loss is commonly seen as well, which may be due to conduction abnormalities from the bones in the middle ear, sensorineural abnormalities from abnormalities of the inner ear organs, or a combination of the two.

The lungs and cardiovascular system may be affected. Patients with osteogenesis imperfecta can have abnormal valvular tissue leading to mitral valve prolapse or aortic regurgitation. The connective tissue of vessel walls is weakened which can lead to easy bruising.

Often musculoskeletal complaints will cause the patient to present to the orthopedist. Ligamentous laxity is characteristic, which leads to hypermobility of the joints. Basilar invagination, a condition in which the occipito-cervical junction gradually deforms and the edges of the foramen magnum fold inward, may be seen in some subtypes of osteogenesis imperfecta, leading to neurologic compromise. Scoliosis may be present to some degree, which may be quite severe in some subtypes.

Generalized bone brittleness and fragility are common. Recurrent fractures may lead one to suspect the diagnosis in otherwise mild forms of the disease. Olecranon apophyseal fractures are usually pathognomonic for osteogenesis imperfecta (Figure 3). Notably, the risk of fracture drops dramatically after puberty.
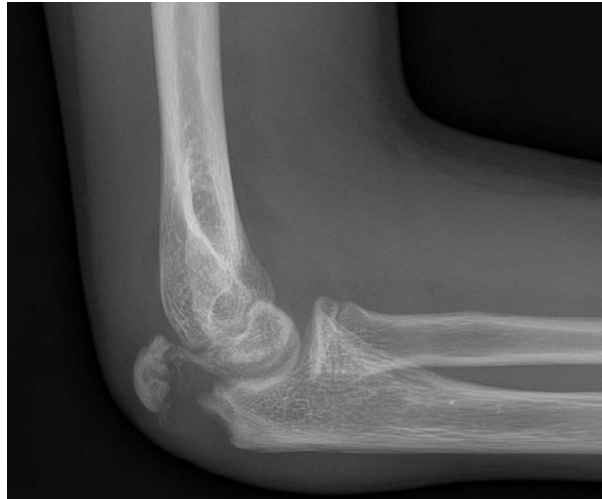
*Figure 3: Lateral radiographic view of the right elbow in a patient with osteogenesis imperfecta, demonstrating an olecranon apophyseal fracture.*

Severe bowing of the limbs is usually present in more severe forms (Figure 4A). The legs may exhibit the classic "saber shin" appearance due to anterior bowing of the tibiae (Figure 4B). The femora may be severely bowed. Compression fragility fractures of the spine lead to the classic "codfish vertebrae" appearance on lateral spine radiographs (Figure 4C).
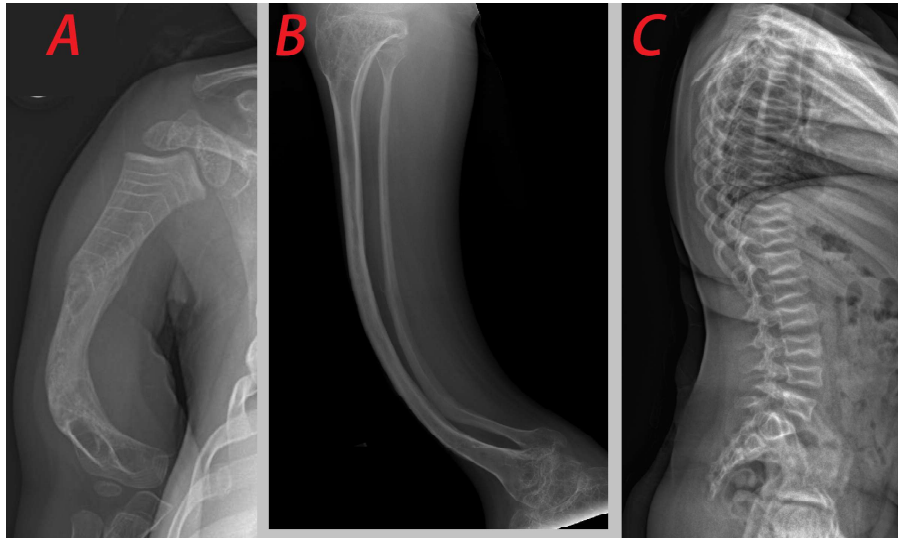


*Figure 4: A) Anteroposterior radiographic view of the right humerus in a patient with osteogenesis imperfecta. Note the severe bowing deformity present. Also noticeable are multiple growth arrest lines in the proximal half of the humerus. B) Lateral radiographic view of the tibia in a patient with osteogenesis imperfecta. Note the anterior bowing of the tibia, the so-called "saber shin" deformity. C) Lateral radiographic view of the spine of a patient with osteogenesis imperfecta. Note the biconcave appearance of multiple vertebral bodies due to multiple compression deformities over time. This appearance is termed "codfish vertebrae."*

## OBJECTIVE EVIDENCE

A diagnosis of osteogenesis imperfecta can usually be made based on family history and typical radiographic findings. Although DNA analysis may reveal mutations, especially in COL1A1 and COL1A2, due to the variety of mutations that lead to an osteogenesis imperfecta phenotype, there is no one simple lab test that can lead to diagnosis.

Radiographic analysis of patients with osteogenesis imperfecta will show osteopenia and thinned cortices. There may be radiographic evidence of multiple prior fractures – these may be pseudarthroses (a "false joint") if the bones failed to heal.

Long bones are typically thin and have variable degrees of progressive bowing and general deformity. As mentioned above, the radiocapitellar joint may be dislocated, and ossification of the interosseous membrane can be seen. The pelvis may show coxa vara due to the stress seen at the proximal femur and an inability to remodel correctly in response to this stress.

In patients who have been treated with bisphosphonates, multiple radio-dense lines may be seen in long bones which correlate to growth arrest and subsequent recovery during treatment (Figure 5).



*Figure 5: Anteroposterior radiographic view of the tibia and fibula in a patient with osteogenesis imperfecta. Radiographic growth arrest lines are appreciated in the proximal and distal metaphyses of both the tibia and fibula.*

Multiple compression fractures in the spine may lead to a biconcave appearance of the vertebral bodies, the classic "codfish vertebrae" appearance, or even a complete flattening of the bone called platyspondyly (Figure 6) if the compression was more severe.
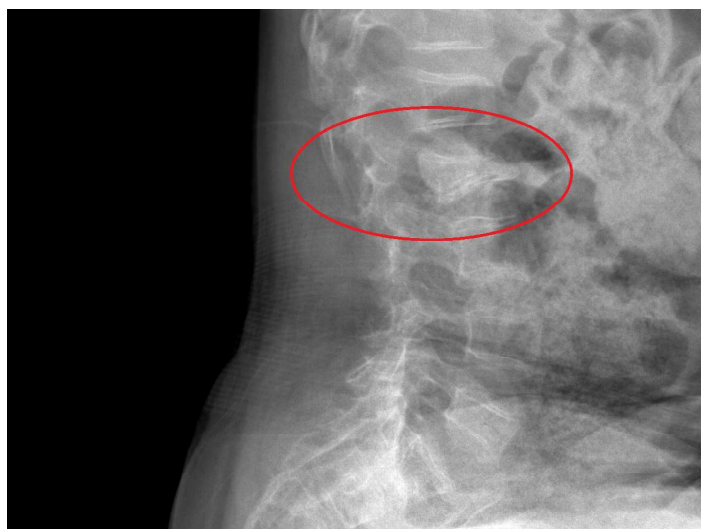
*Figure 6: Lateral radiographic view of the spine of a patient with osteogenesis imperfecta. Note the multiple vertebral compression deformities and flattening of the vertebra ("platyspondyly"), especially at L2.*

Basilar invagination can be evaluated with lateral cervical spine radiographs or sagittal views on CT scan or MRI. Basilar invagination is present when the dens protrudes superior to the McRae line, drawn from the caudal aspects of the basion and opisthion (Figure 7).
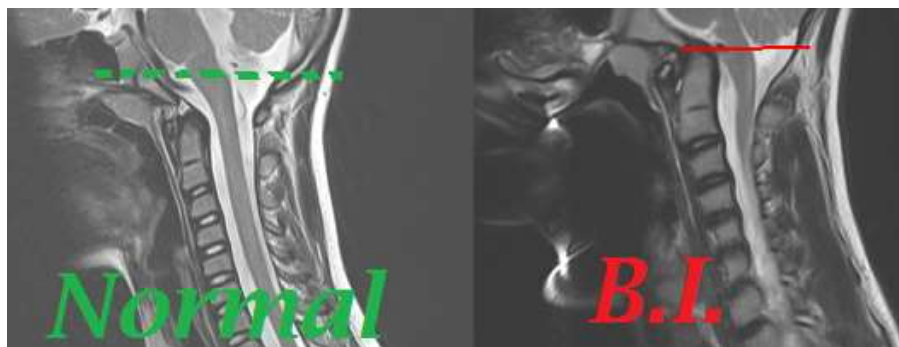


*Figure 7: Basilar invagination at right, normal at left as shown on mid-sagittal cuts of the cervical spine of two different patients. On the right, basilar invagination is present. Shown in green and red is the approximate location of the foramen magnum. Note that in the image on the right, the dens (also known as the odontoid process) is protruding cranially, through the foramen magnum to the level of the basion. The dens normally lies about 5 mm below this point.*

## EPIDEMIOLOGY

Considering all subtypes, osteogenesis imperfecta occurs about once per 15,000–20,000 live births with no known variability across races and geographic regions. Type I osteogenesis imperfecta is the most common, found in one in 30,000 live births; osteogenesis imperfecta type II, the most severe form, is happily the rarest. Osteogenesis imperfecta affects males and females in equal numbers. In most cases, osteogenesis imperfecta is inherited in an autosomal dominant pattern. Osteogenesis imperfecta type II can occur as a spontaneous genetic mutation, which is then transmitted in an autosomal dominant pattern. Rare forms of osteogenesis imperfecta are inherited as autosomal recessive genetic traits.

## DIFFERENTIAL DIAGNOSIS

Any disorder that presents with frequent or recurrent fractures may share some phenotypic characteristics with osteogenesis imperfecta. Importantly, if a child presents with multiple fractures, child abuse (non-accidental trauma) must be investigated and ruled out. Indeed, the possibility of child abuse must not be ignored merely because a diagnosis of osteogenesis imperfecta is present!

Other conditions which may have a musculoskeletal presentation similar to osteogenesis imperfecta include vitamin-D homeostasis deficiencies and hypophosphatasia, celiac disease, some forms of Ehlers-Danlos syndrome, Menkes disease, and I-cell disease.

## RED FLAGS

- Any child presenting with skin lesions and/or fractures otherwise unexplained must be worked up for non-accidental trauma. As mentioned above, just because a diagnosis of osteogenesis imperfecta is made does not mean child abuse is not present.
- Myelopathy or other motor/sensory changes, ataxia, decreased mental status, or apnea suggest possible basilar invagination.
- Easy fatigability, shortness of breath and murmurs must prompt an evaluation for mitral valve prolapse, aortic regurgitation, aortic dissection.
- The risk of malignant hyperthermia is increased in patients with osteogenesis imperfecta. Vigilance on the part of the anesthesia and surgical teams is required when patients are undergoing surgery.

## TREATMENT OPTIONS AND OUTCOMES

Treatment of the clinical effects of osteogenesis imperfecta is specific to each patient and subtype of disease. Many clinicians recommend early bracing to correct deformities and/or physical therapy in an effort to prevent fractures from occurring.

Bisphosphonates are also commonly used in patients with osteogenesis imperfecta, which have been shown to decrease the incidence of fractures as well as pain from impending fractures and brittle bones. Bisphosphonates work by inhibiting osteoclasts, which thereby effectively decreases bone resorption and leads to thicker cortical walls.

Certain fracture patterns treated non-operatively in children without osteogenesis imperfecta may also often be treated non-operatively in children with osteogenesis imperfecta. In an effort to prevent further fractures in this fragile population, the caveat with non-operative treatment is that light splints may be favored over rigid casts, and immobilization times generally are shortened.

Fractures of the femur and tibia may be treated with rods that span the entire bone to allow simultaneous support and continued growth. Load sharing devices that protect the whole bone should be used. Plates should be avoided due to the propensity for creating stress risers and increasing the risk of peri-implant fracture.

In the setting of bowed long-bones, single or multiple re-alignment osteotomies can be performed. These are secured with traditional (non-telescopic) rods (Rush) or telescopic rods (Fassier-Duval) which allow the rod to expand as the child continues to grow (Figure 8). A "bisphosphonate holiday" must be taken around the time leading up to long bone surgery and for some time thereafter, as bisphosphonates can impair the fracture and osteotomy healing process.

*Figure 8: Anteroposterior radiographic views of the bilateral lower extremities showing telescopic rods that have been placed after osteotomies to correct deformity and allow for continued growth.*

Basilar invagination, if severe, may be treated with surgical decompression and fusion from the occiput to either C2 or C3. This is done to prevent neurologic compromise. Scoliosis in children with osteogenesis imperfecta cannot be well controlled with bracing, as the bones are simply too fragile to allow that. Surgical fusion is sometimes performed when the curve progresses beyond 45 degrees, but may prove challenging due to the poor bone quality.

Life expectancy in osteogenesis imperfecta relates to the subtype of disease: Type II disease often results in perinatal death, whereas a normal lifespan can be expected in more mild Type I cases. Respiratory failure is the most common cause of death in patients with osteogenesis imperfecta.

Outcomes following fracture are poorer compared to the general population. The healing at the fracture site will not be as strong as fracture healing in an individual without osteogenesis imperfecta. This lack of strength is due to the defects in type I collagen and the important role that type I collagen plays in fracture remodeling.

Although patients with osteogenesis imperfecta experience a much higher fracture burden than the general population, most patients with the condition lead normal lives, work normal jobs, and have families. In general, patients with osteogenesis imperfecta should be counseled on optimizing bone health; this includes avoidance of smoking, excessive alcohol intake, appropriate calcium and vitamin D intake, and avoidance of high-risk activities that would put them an increased risk of skeletal trauma.

## RISK FACTORS AND PREVENTION

As osteogenesis imperfecta is a disease due to a genetic mutation, children of parents with known osteogenesis imperfecta are at higher risk of having the disease.

Respiratory failure (due to scoliosis, narrowed thorax, and basilar invagination) is the most common cause of death in patients with osteogenesis imperfecta.

## KEY TERMS

osteogenesis imperfecta, COL1A1, COL1A2, blue sclera

# MUSCULAR DYSTROPHY

Muscular Dystrophy is a group of more than 30 genetic diseases that are characterized by the production of abnormal muscle proteins leading to progressive weakness and loss of muscle mass. There are various types of Muscular Dystrophy and the severity of symptoms, location, and age of occurrence vary between the various types. The most common form of muscular dystrophy is Duchenne Muscular Dystrophy (DMD). Duchenne Muscular Dystrophy primarily affects boys and is caused by the absence of dystrophin, a protein involved in maintaining skeletal muscle. Onset is at approximately 3 years of age and is progressive: the initial presentation is lower limb muscle weakness and gait impairment but eventually, there is loss of muscle strength in the upper limbs and impairment of the diaphragm and heart leading to cardiopulmonary failure.

## STRUCTURE AND FUNCTION

In the cytoplasm of normal skeletal muscle, there is a protein called dystrophin that connects the cytoskeleton of a muscle fiber to the surrounding extracellular matrix through the cell membrane. Dystrophin plays an important role in cell signaling, muscle cell membrane stabilization, and the transmission of force from the contracting sarcomere to the muscle cell membrane. In Duchenne Muscular Dystrophy, dystrophin is absent. In Becker Muscular Dystrophy, dystrophin is present but in lower quantity compared to normal muscle cells.

A lack of dystrophin leads to necrosis and fibrosis of the muscle cells (Figure 1). On gross examination, healthy skeletal muscle tissue in Muscular Dystrophy is replaced with fibrous and fatty tissue. On microscopy, immunoblot preparations with stains for dystrophin will show absent dystrophin in Duchenne Muscular Dystrophy and decreased intensity of the signal in Becker Muscular Dystrophy due to the quantitative decrease in dystrophin.
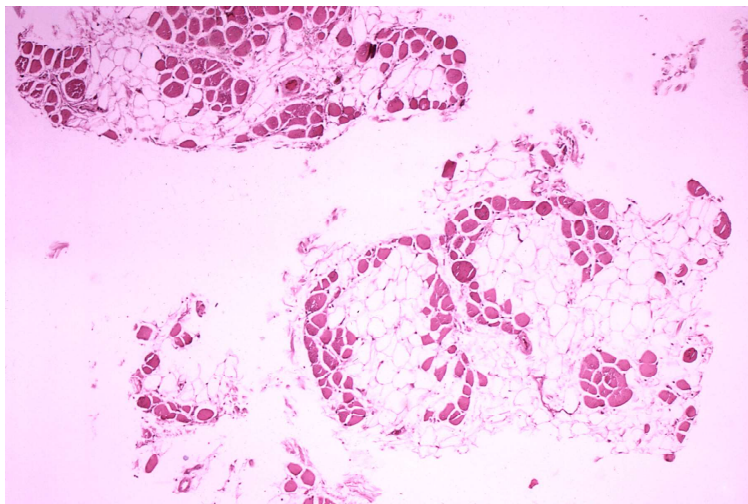


*Figure 1: Biopsy of the calf muscle from a person with Duchenne Muscular Dystrophy showing replacement of muscle fibers by fat cells.*

The muscular dystrophies are genetic disorders which may be inherited in an autosomal dominant, autosomal recessive, or X-linked recessive fashion. Duchenne Muscular Dystrophy is due to a point deletion causing a nonsense mutation in the gene which encodes the dystrophin protein. The result is an inability to produce dystrophin. This mutation is inherited in an X-linked recessive manner (Figure 2). Becker Muscular Dystrophy,

also X-linked recessive, is due to a mutation in the non-coding region of the gene encoding dystrophin, which does not shift the translation reading frame like the Duchenne Muscular Dystrophy mutation. The mutation in Becker Muscular Dystrophy leads to the production of a truncated version of dystrophin, and smaller quantities of the protein are produced.
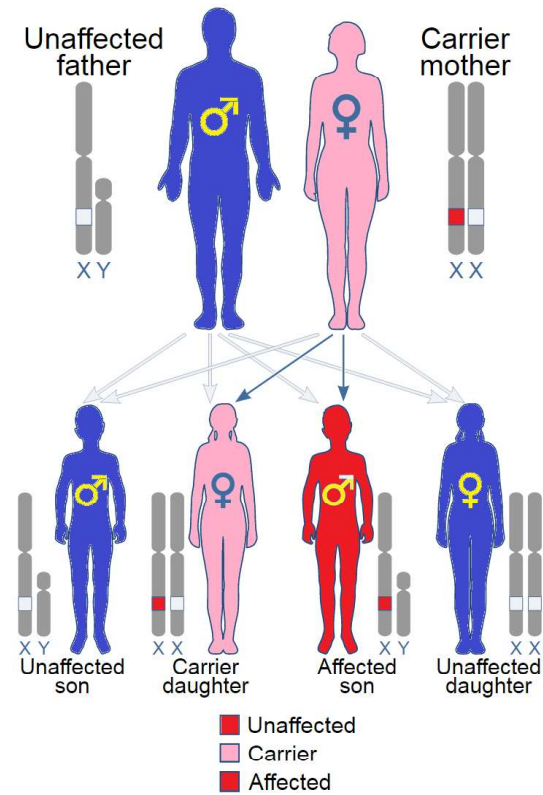


*Figure 2: X-linked recessive transmission. (image modified from Wikipedia)*

## PATIENT PRESENTATION

Duchenne Muscular Dystrophy generally presents between 3 and 6 years of age. The condition first affects muscles near the hip and shoulder, leading to difficulty in jumping, running, climbing stairs, and toe walking.

In most patients, the classically-described calf pseudohypertrophy may be seen. This phenomenon is due to fatty and fibrotic infiltration in the calf muscles. Increased lumbar lordosis may be present, as this compensates for the gluteal muscle weakness present (Figure 3).
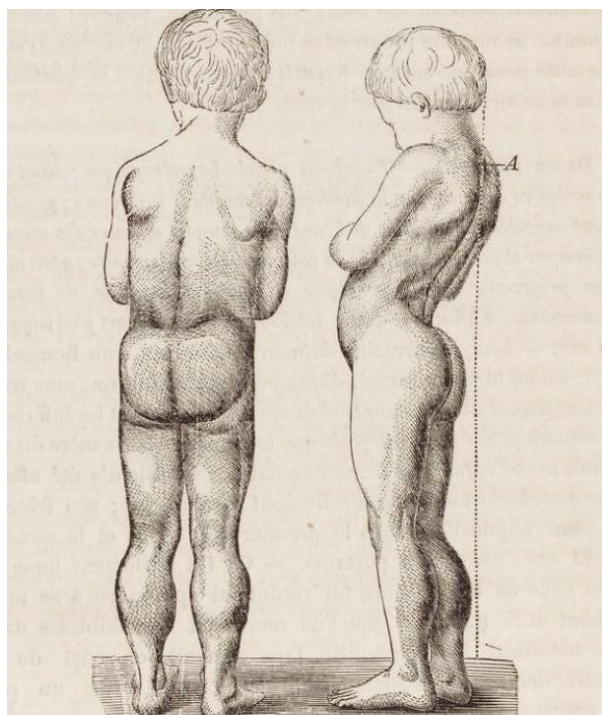
*Figure 3: A drawing of a 7-year-old boy with muscular dystrophy. Note the lower limb pseudohypertrophy, relative thinness of the arms and increased lumbar lordosis. (Image from Duchenne's original manuscript)*

To compensate for the muscle weakness first affecting the hip, when patients are rising from the floor, they will walk their hands up their legs and push their knees into extension to assist in standing themselves up. This maneuver is termed "Gower's sign" (see Figure 4). If nearby objects such as tables, chairs or walls are present, patients may use these to achieve standing posture as well
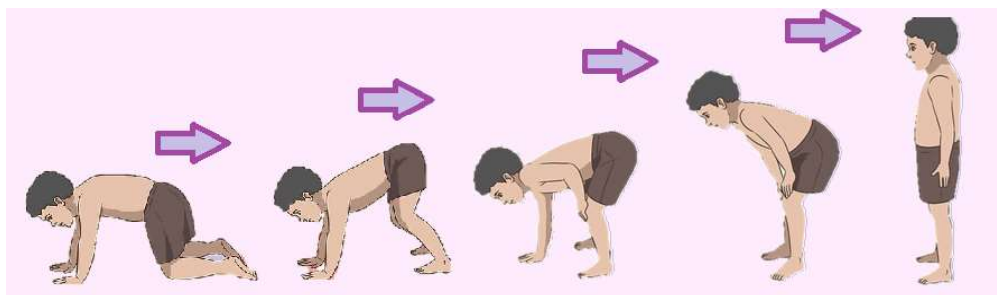


*Figure 4: Gower's sign is seen when children with proximal muscle weakness stand up by first placing their hands on the ground and then walking towards their hands and pushing up along their own legs. Patients with weak lower limbs are able push up against their own bodies due to relative preservation of distal arm muscle strength. (Modified from https://www.wikihow.com/Diagnose-Muscular-Dystrophy#/Image:Diagnose-Muscular-Dystrophy-Step-2.jpg).*

As patients age, the muscle weakness becomes more severe and progressive, leading to upper limb girdle weakness and trunk weakness which often results in scoliosis. Eventually, patients lose the ability to ambulate independently and will become wheelchair-bound by 10-15 years old. The progressive weakness leads to respiratory issues and often cardiomyopathies.

Symptoms in patients with Becker Muscular Dystrophy present later than those in Duchenne Muscular Dystrophy, often after age 7, due to a less severe disease course. Becker Muscular Dystrophy patients maintain ambulatory ability well into their teenage years.

Becker Muscular Dystrophy patients have a higher incidence of cardiomyopathy than those with Duchenne Muscular Dystrophy. Respiratory problems in Becker Muscular Dystrophy are also common, but occur later than Duchenne Muscular Dystrophy.

## OBJECTIVE EVIDENCE

Elevated levels of creatine phosphokinase (CPK), leaking from damaged muscle, is a specific test for muscular dystrophy. Due to the abnormal muscle composition and progressive skeletal muscle breakdown, CPK levels in patients with Duchenne Muscular Dystrophy are elevated and can be as high as 200x normal values. Becker Muscular Dystrophy patients also show elevated CPK on lab testing, but levels are not as elevated as in Duchenne Muscular Dystrophy.

Genetic testing is available for detecting the abnormal gene which codes dystrophin. A polymerase chain reaction assay can detect deletions of the dystrophin gene.

A muscle biopsy may also be taken for the purposes of diagnosis (as shown in Figure 1), but genetic tests are the more common route to diagnosis.

When the child develops scoliosis, radiographs of the spine typically may show a long, C-shaped or S-shaped curve which may differ from the typical patterns seen in adolescent idiopathic scoliosis (Figure 5).



*Figure 5: An anteroposterior radiograph of the spine in a patient with Duchenne Muscular Dystrophy showing a scoliosis with a characteristic long, sweeping S-shaped curve.*

## EPIDEMIOLOGY

Because Duchenne Muscular Dystrophy and Becker Muscular Dystrophy are X-linked recessive, only male patients are affected, except in very rare cases (for fascinating reasons, including missing, damaged or inactivated second X chromosomes — all beyond the scope of this text).

The incidence of Duchenne Muscular Dystrophy is one in 5,000 male live births. Becker Muscular Dystrophy is around 1/10th as rare, with an incidence of one in 30,000 male live births.

## DIFFERENTIAL DIAGNOSIS

Differential diagnoses for Duchenne Muscular Dystrophy and Becker Muscular Dystrophy include the other muscular dystrophies, such as limb girdle muscular dystrophy or myotonic muscular dystrophy. Other progressive neuromuscular conditions can have similar symptoms as Muscular Dystrophy, such as spinal muscular atrophy (SMA). One way to tell them apart clinically is that in Duchenne Muscular Dystrophy and Becker Muscular Dystrophy, deep tendon reflexes are preserved, unlike in SMA. Guillain-Barre syndrome (GBS) can present with progressive weakness as well, but tends to progress faster. GBS also characteristically lacks deep tendon reflexes, and creatine phosphokinase levels will be normal.

## RED FLAGS

- Inability to climb, run, jump, or keep up with peers as a child should prompt investigation into Muscular Dystrophy.
- Shortness of breath or murmurs necessitate a cardiac evaluation to rule out cardiomyopathy or respiratory failure.
- The risk of malignant hyperthermia is increased in patients with Muscular Dystrophy. Vigilance of the anesthesia and surgical teams is required when patients are undergoing surgery.

## TREATMENT OPTIONS AND OUTCOMES

Corticosteroids may be used to treat children with progressive symptoms. Corticosteroid therapy has been shown to slow overall progression. It can delay the deterioration of ambulatory and lung function and can delay the progression of scoliosis. However, corticosteroid treatment is not without well-known side-effects, which include weight gain and Cushingoid appearance, osteonecrosis, headaches, cataracts, GI symptoms, and stunting of growth.

Supporting respiration during sleep with a ventilator has also been shown to lengthen lifespan in Muscular Dystrophy patients. In some patients, a tracheostomy may be required to facilitate this, but most individuals with Muscular Dystrophy can use Continuous Positive Airway Pressure (CPAP) or Bi-level Positive Airway Pressure (BIPAP) machines.

Physical therapy and orthotics also play a large role in the treatment of Muscular Dystrophy patients. Maintaining strength and range of motion may serve to prolong ambulation and prevent contracture formation. Targeted orthoses and braces may help maintain mobility and independence. When ambulatory function is lost, custom or adaptive equipment including a motorized wheelchair allows patients to maximize independence.

The degree of surgical treatment necessary for the care of patients with Muscular Dystrophy is still debated. Contracted joints (ankle plantarflexion contractures due to tight Achilles, knee contractures due to tight hamstrings, etc.) may be targets for surgical lengthening in an effort to prolong ambulatory ability, but the rehabilitation phase of surgical treatment must not be overlooked. Vigilance with early post-operative physical therapy is necessary to prevent deconditioning and limit the progress achieved by the surgical intervention itself.

The development of scoliosis is extremely common in this patient population, especially after ambulation is lost. Bracing should not be used in these patients – it does not prevent curve progression, and more importantly, may restrict pulmonary function in a patient with already tenuous pulmonary status.

A scoliosis measuring 20 to 30 degrees is usually an indication for posterior spinal fusion (with an anterior release sometimes added for severe, stiff curves). Often, fusion to the pelvis must be performed to correct

pelvic obliquity. Rapid progression of the curve or severe restrictive lung disease (manifested as low forced vital capacity, the amount of air that can be forcibly exhaled from the lungs after taking the deepest breath possible) are also used as indications for surgery.

The goals of surgical treatment in scoliosis for Muscular Dystrophy patients is to prevent further spine deformity, thus preventing further worsening of restrictive lung disease, and a balanced spine over a balanced pelvis to facilitate sitting posture, comfort, and care.

Patients with Duchenne Muscular Dystrophy may survive, on average, into their mid-20s. This life expectancy, however, is greatly dependent on the quality of care, with some patients who have access to excellent care and caregivers living into their 30s or even 40s.

The cause of death is usually respiratory failure, highlighting the need for appropriate respiratory care in these patients (including nighttime ventilation and well-timed spine fusion). Heart failure due to dilated cardiomyopathy is another common cause of death. Those with Becker Muscular Dystrophy, due to a milder and more delayed constellation of symptoms, live much longer.

Patients with Duchenne Muscular Dystrophy have a higher complication profile following surgical procedures. As mentioned above, malignant hyperthermia is common in these patients, and so appropriate steps must be taken by the anesthesia and surgical teams prior to operating.


## RISK FACTORS AND PREVENTION

Duchenne Muscular Dystrophy and Becker Muscular Dystrophy are both genetic diseases inherited in an X-linked recessive manner. Thus, females who are carriers of the disease have a 50% chance of passing the disease on to their offspring. However, around 1/3rd of cases are due to a spontaneous mutation.


## MISCELLANY

The gene which codes for Dystrophin is one of the longest, representing almost 0.1% of the entire human genome.

Although the condition now known as Duchenne Muscular Dystrophy was described by Giovanni Semmola in 1834 and Gaetano Conte in 1836, Duchenne Muscular Dystrophy is named after Guillame-Benjamin-Amand Duchenne, a French physician who described the condition in a series of patients. Notably, Duchenne was the first physician to perform a surgical biopsy, that is, taking tissue from a living patient for the purposes of diagnosis.

Since Duchenne Muscular Dystrophy and Becker Muscular Dystrophy are both due to mutations in a single, known gene, ongoing research is focused on targeted gene therapy in an effort to replace the mutated gene in affected individuals via a viral vector.


## KEY TERMS

Duchenne muscular dystrophy, Becker muscular dystrophy, Dystrophin, Gower's sign

# SPINA BIFIDA

Spina bifida is a spectrum of congenital malformations in which the neural tube does not close during embryonic development. Spina bifida represents a range of diseases including **spina bifida occulta**, in which the posterior bony elements fail to fuse but the dura and neural elements remain unaffected; **meningocele**, in which the posterior bony elements fail to fuse and the dura and arachnoid tissue form a sac that protrudes from the bony defect; and last, **myelomeningocele**, where posterior bony elements fail to fuse, and the sac that protrudes through the bony defect includes not only dura and arachnoid but spinal cord and neural elements (Figure 1). Spina bifida occulta is so named because it is "occult," or hidden by a layer of skin that covers the malformation of the bone. Spina bifida occulta is present in 10% or more of the general population and rarely causes signs or symptoms. Some individuals with meningocele may have few or no symptoms yet some may have varying degrees of paralysis and bladder and bowel dysfunction. Myelomeningocele uniformly results in partial or complete dysfunction of the nerves below the spinal opening, which leads to paralysis and bladder and bowel dysfunction.
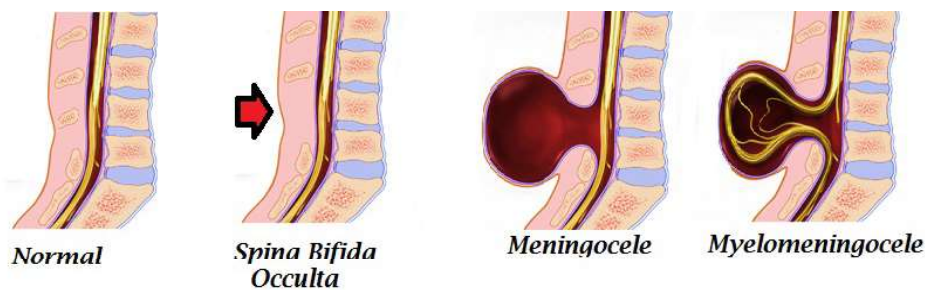


*Figure 1: The spectrum of disease in spina bifida, ranging from fusion failure of posterior bony elements (spina bifida occulta); to protrusion of the dura and arachnoid tissue but without the spinal cord (meningocele); or protrusion of the spinal cord and neural elements within the dural sac (myelomeningocele).*

## STRUCTURE AND FUNCTION

Spina bifida (Latin, meaning "spine in two parts") is caused by a developmental failure of fusion of the vertebral arches, with or without protrusion of the spinal cord (Figure 2).

Spina bifida can occur at any level of the spine, but most lesions are found in the lumbo-sacral region (Figure 3).
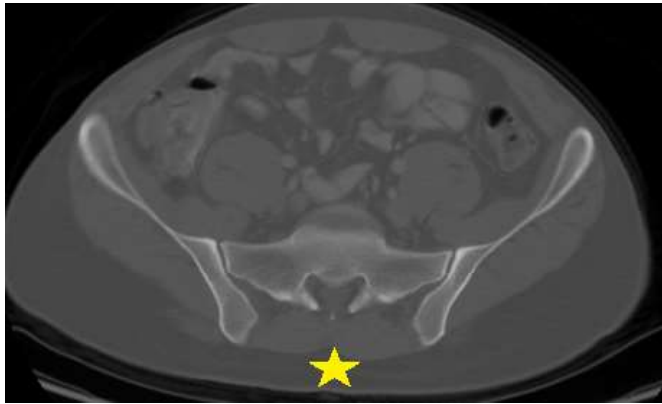
Figure 2: CT scan of the pelvis showing a failure of fusion. As shown (by the star), there is a gap in the posterior aspect of the spinal canal. (Case courtesy of Dr Hani Makky ALSALAM, Radiopaedia.org, rID: 8909)
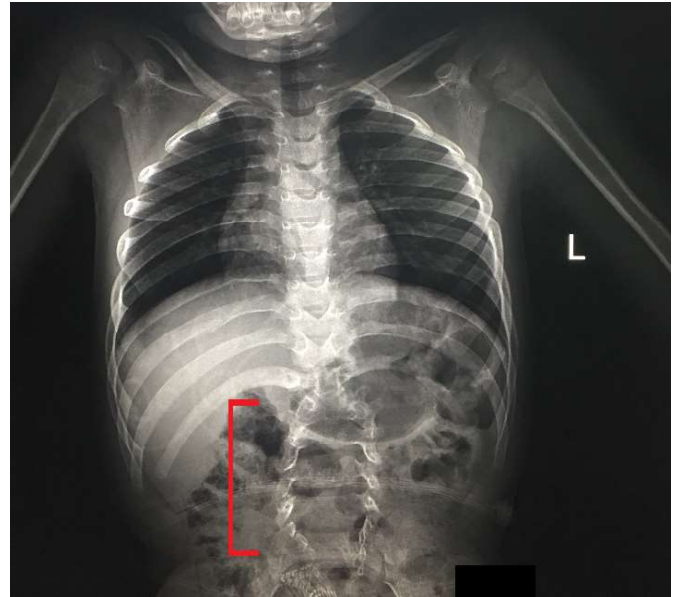


Figure 3: Failure of fusion of the lumbo-sacral spine. (Case courtesy Radiopaedia.org, rID: 73276)

In cases of myelomeningocele, neurological function is impaired at the spinal level at which the defect is found. The lowest normal level determines function (Table 1). Patients with an L5 functional level, for example, will be community ambulators. An L3 level (with impaired quadricep function) limits a patient to ambulation in the home with an orthosis. Note, however, that body habitus and motivation to walk can influence function, and both body habitus and motivation vary from child to child. Accordingly, the relationships shown in the table are not ironclad. Nonetheless, independent of body habitus and motivation, quadriceps function is needed for community ambulation.

*Table 1: Functional Levels of Myelomeningocele*

| Lowest Level Intact | Muscle Function Present | Ambulatory Status | Notes |
|---|---|---|---|
| L2 | Some hip flexion (iliopsoas) may be preserved | Minimal ambulation possible with a reciprocating gait orthosis | |
| L3 | Hip flexion and hip adduction | Some household ambulation with reciprocating gait orthosis or hip, knee, ankle, and foot orthosis | Highest risk of hip instability and dislocation |
| L4 | Knee Extension (quadriceps), ankle dorsiflexion/ inversion (tibialis anterior) | Household ambulation, may require Ankle-Foot Orthosis and/or crutches | Biggest difference in mobility prognostication is between L3 and L4 levels due to quadriceps function |
| L5 | Hip Extension and hip abduction (gluteal muscles), knee flexion (medial hamstrings), toe dorsiflexion (extensor hallucis longus and extensor digitorum longus muscles) | Community ambulation, may require Ankle-Foot Orthosis and/or crutches | Medial hamstring function helpful for community ambulation |
| S1 | Ankle plantarflexion (gastrocnemius-soleus complex) | Community ambulation, may require Ankle-Foot Orthosis | |
| S2 | Toe plantar-flexion (flexor hallucis longus and flexor digitorum longus muscles) | Community ambulation, may require Ankle-Foot Orthosis | |

In addition, the more rostral (toward the head) the defect is, the more severe the secondary orthopaedic manifestations, including scoliosis, hip dysplasia, knee contractures, and foot deformities.

The risk of developing scoliosis correlates with the spinal level; those with thoracic lesions develop scoliosis in nearly all cases. Kyphotic deformity may be present and may be very focal in nature – a so-called "Gibbus deformity."

Flexion and adduction contractures near the hip are common in myelomeningocele patients. Muscles responsible for hip abduction and extension receive innervation largely from L4 and L5 nerve roots, while hip flexors and adductors are innervated by L1-L3 nerve roots. Accordingly, if the patient is functional through the L3 level, the hip flexors and adductors will work while hip extensors and abductors will not. The resulting muscle imbalance leads to hip subluxation, dysplasia and, in extreme cases, posterosuperior dislocation.

Even without dislocation, contractures can cause sitting imbalance and pelvic obliquity. In turn, this can cause pressure sores and skin breakdown.

Various knee deformities can be seen in myelomeningocele patients (Figure 4). Flexion and, less frequently, extension contractures may be seen. Patients with a mid-lumbar functional level often develop knee valgus due to their relative hip abductor weakness.



Figure 4: Radiograph of a knee in a patient with a
high spinal lesion causing Charcot arthropathy.

Deformity of the foot and ankle is frequently encountered among patients with myelomeningocele. Clubfoot, often bilateral, is the most common of these deformities. Clubfoot caused by myelomeningocele is typically more rigid than that seen in idiopathic clubfoot and therefore less amenable to treatment by casting. Equinovarus, equinus, cavovarus, calcaneal, calcaneovalgus deformities and congenital vertical talus may also be seen.

Children with myelomeningocele are often osteopenic and lack protective sensation below the functional level. The combination of weak bones and a lack of protective sensation increases the chances of suffering pathologic fractures.

Conditions such as spinal cord tethering, hydrocephalus, Arnold-Chiari malformation, or syrinx may be present or may develop in the neural axis.

Failure of the neural tube to fuse posteriorly during embryonic development can be caused by genetic and environmental factors. Maternal folate deficiency is a well-known risk factor. From a genetic standpoint, certain chromosomal abnormalities may lead to spina bifida, such as trisomy 13 and 18 (Patau and Edwards syndrome, respectively). Many single gene mutations have also been linked to spina bifida.

## PATIENT PRESENTATION

Spina bifida may be diagnosed in utero by ultrasound. Myelomeningocele and meningocele may be directly visualized by examining the infant's back where the lesion is obvious (Figure 5).
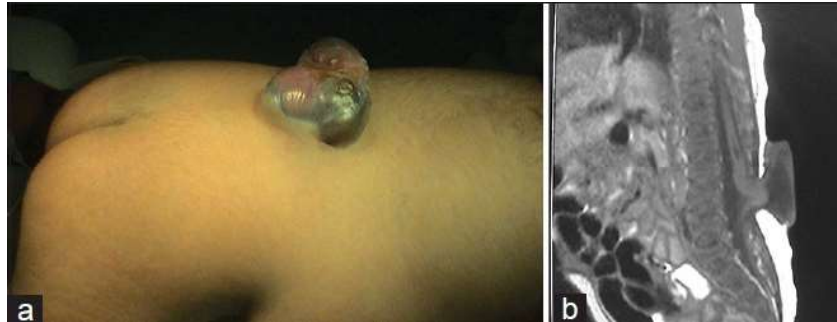


*Figure 5: Clinical photo of a child with myelomeningocele (A) with corresponding MRI (B). (Image courtesy Bao N, Lazareff J. How I Do It: Management of spina bifida in a hospital in The People's Republic of China. Surg Neurol Int 23-Jul-2015;6)*

As the name implies, spina bifida occulta may not be obvious. At times, there is a hairy patch or skin dimpling present. However, the overlying skin will often appear normal. The defect may be directly palpated, revealing lack of midline fusion of the bony posterior spinal elements. Note that most people with spina bifida occulta have no signs or symptoms; it is discovered incidentally when x-rays, CT scans, or MRI studies are obtained for other reasons.

When examining patients with spina bifida, it is good practice to avoid wearing latex gloves as the incidence of latex allergy is high in this cohort of patients.

The scoliosis associated with spina bifida may be progressive, so serial examination is recommended. Progression may be rapid if the scoliosis is in part due to a neural axis abnormality, such as tethering of the cord.

As noted, the combination of osteopenia and lack of sensation increases the risk of pathological fractures. The classic presentation of a fracture associated with myelomeningocele is swelling, erythema, and warmth. These signs may be confused with infection. Thus, in the case of swelling, erythema, and warmth, both infection and fracture must be ruled out.

The skin below a functional level must be checked regularly, as decreased sensation and contractures can lead to abnormal pressure points and ulcers.

A vigilant neurological exam is warranted whenever these patients are seen. As noted, conditions such as spinal cord tethering, hydrocephalus, Arnold-Chiari malformation, or syrinx may be present or may develop in the neural axis. Any change in neurologic exam or suspicion should prompt a neurosurgical referral.

## OBJECTIVE EVIDENCE

Various lab and imaging studies may be utilized to diagnose spina bifida in utero. Screening is often performed via α-fetoprotein assay of the mother's serum. To confirm diagnosis, ultrasound may be used to visualize the fetal spine. Amniocentesis may also be employed to confirm a diagnosis, in which α-fetoprotein and acetylcholinesterase levels are measured.

In patients with decreased to no ambulatory capacity, radiographs of the lower extremities will reveal osteopenia. Radiographs of the spine in patients with kyphoscoliosis often show a neuromuscular type curve, which may be long and C-shaped. The focal kyphotic "Gibbus deformity" may be seen on lateral spine imaging. Radiographs of affected hips may show underdeveloped acetabula with posterosuperior dislocation.

## EPIDEMIOLOGY

Spina Bifida is the most common major birth defect. About 10-20% of people have spina bifida occulta. The risk of overt spina bifida is ~0.1% in the general population; there is a 5% chance of spina bifida in a given fetus if an older sibling had spina bifida and 40% if two siblings are affected. The incidence of spina bifida varies greatly by region, due to both environmental and racial factors. Caucasians are at a higher risk of giving birth to a child with spina bifida.

## DIFFERENTIAL DIAGNOSIS

Spina bifida is usually diagnosed definitively, and thus there is no true differential diagnoses. Still, it is instructive to consider how the neuromusculoskeletal manifestations in patients with myelomeningocele may overlap with other pediatric conditions.

Clubfoot in myelomeningocele patients may appear identical to idiopathic clubfoot, but is often much more rigid.

Cavovarus feet seen in some patients may appear similar to those who are afflicted with Charcot-Marie-Tooth disease.

The scoliosis and pelvic obliquity can appear similar to curves seen in other neuromuscular conditions, such as cerebral palsy or muscular dystrophy. The focal kyphotic deformity sometimes seen in these patients may appear similar to spinal deformity seen in patients with spinal tuberculosis (Pott's disease) or MPS I (Hurler syndrome).

## RED FLAGS

- Red, hot, swollen limb in a patient with myelomeningocele suggests fracture or infection until proven otherwise.
- Facial swelling, shortness of breath, flushing, and other symptoms of anaphylaxis may be due to latex allergy and exposure.
- Changes in neurologic examination may be caused by spinal cord tethering, syrinx or other cord abnormalities and should prompt a neurosurgery referral.

## TREATMENT OPTIONS AND OUTCOMES

There is no cure for spina bifida. The best treatment is prevention (through folate supplementation).

Fetal surgery – to repair myelomeningocele in utero – has shown promise to limit clinical impairments but is "considered experimental" according to the National Institute of Neurological Disorders and Stroke.

The treatment of patients with spina bifida involves the coordination of multiple different specialties, especially for patients with myelomeningocele, to address the complications caused by loss of neurological function.

Neural axis abnormalities require neurosurgical care, and bowel and bladder issues may require pediatric general surgery and urology.

Physical therapy and orthotics play a major role; depending on the functional level in myelomeningocele patients, braces might be necessary to maintain ambulatory ability. Focused rehabilitation exercises help to prevent formation of contractures. Appropriate fitting for crutches, walkers, or wheelchairs is necessary.

As is the case with other neuromuscular scoliotic deformities, bracing for spina bifida scoliosis is not effective. Thus, surgical fusion may be necessary if the deformity is severe or progressive. Posterior spinal fusion often includes fusion to the pelvis to correct pelvic obliquity. Anterior spinal release and fusion might be needed because the posterior spinal elements may be absent or dysplastic, compromising available bone stock for posterior instrumentation.

Kyphotic deformities are often progressive and treated surgically to prevent posterior skin breakdown and formation of pressure sores. However, due to the neural axis abnormalities common in these patients, a previous shunt may be present. It is of vital importance that shunt function is checked, as shunt compromise during surgical correction can lead to acute hydrocephalus in these patients, often resulting in death.

Soft tissue contractures about the hip greater than 40 degrees are often treated with surgical release. The optimal treatment of dislocated hips in patients with myelomeningocele is still debated. Most surgeons will treat hip dislocations in these patients without surgery. Some advocate for surgical reduction of a unilateral dislocated hip in a patient who is a community ambulatory in an effort to preserve and prolong ambulatory ability. However, re-dislocation rates after surgery are high, leading some authors to advocate for non-operative management.

Flexion contractures are common, and contractures of more than 20 degrees often require surgical management for those who are home or community ambulators. Hamstring lengthening, distal femoral hemiepiphysiodesis, and distal femoral extension osteotomy are commonly employed surgical techniques in this regard.

Serial casting for knee extension contractures may be attempted. If this fails, surgical quadriceps lengthening may be utilized. The goal of treatment of extension deformities is usually to reach a comfortable position for ease of sitting position in the wheelchair.

Clubfoot in myelomeningocele patients may be treated initially with the Ponsetti method in an attempt to provide non-operative correction, but the recurrence rates are high. Thus, surgical treatment with soft tissue releases or tendon transfers is often necessary to lessen the risk of recurrence.

Because prolonged casting can worsen the underlying osteopenia, fractures are immobilized for relatively shorter periods. Further, given the lack of protective sensation in many of these patients, frequent cast changes and vigilant attention to adequate padding must be employed to prevent skin breakdown.

Overall outcomes in spina bifida are strongly influenced by non-musculoskeletal issues, especially neurological complications such as Chiari malformations (with possible hydrocephalus), meningitis, and learning disabilities. Frequent urinary tract infections and pyelonephritis are common and may lead to sepsis.

Within the musculoskeletal system, patients with myelomeningocele are at a higher risk for infection. Pressure sores may develop into pressure ulcers due to lack of protective sensation.

Progressive kyphotic deformities can lead to pressure ulcers overlying a focal kyphotic segment. An ulcer markedly increases the risk of an infection that may travel to the central nervous system, owing to the posterior element defects. Untreated progressive scoliosis may lead to respiratory failure and restricted cardiac output. Because of the lack of adequate soft tissue posteriorly, myelomeningocele patients undergoing spinal fusion surgery are at a much higher risk of surgical site infection (Figure 6). These patients are also at a higher risk of non-union after fusion.

*Figure 6: Clinical photograph of wound breakdown with exposed hardware in a patient after spinal fusion surgery.*

## RISK FACTORS AND PREVENTION

Maternal folate insufficiency is a well-known risk factor associated with spina bifida. The recommended folate intake for women of childbearing age is 0.4mg/day. Although sufficient folate intake does not completely abate the risk of having a child with spina bifida, it can greatly decrease the chances by about 70%.

Taking certain anti-convulsant medications, such as valproic acid, increase the risk of having a child with spina bifida. In women who take such mediation, the recommended daily folate intake is about ten times higher.

Obesity and maternal diabetes also increase the risk of giving birth to a child with spina bifida. To compensate for that, the recommended daily folate intake is higher in affected women.

## MISCELLANY

In the United States, many foods are fortified with folic acid in an attempt to reduce the incidence of children born with spina bifida. The nationwide push to fortify many grains and grain-based foods decades ago has led to a decrease in spina bifida birth rates.

Fetal surgery is proposed to work by shielding the nerves from amniotic fluid, which is thought to be toxic. For that reason, surgery must be performed before 25 weeks gestation.

## KEY TERMS

Spina bifida occulta, meningocele, myelomeningocele

# CEREBRAL PALSY

Cerebral palsy (CP) is defined by the US National Institute of Neurological Disorders and Stroke as a group of neurological disorders "that appears in infancy or early childhood and permanently affects body movement and muscle coordination. CP is caused by damage to or abnormalities inside the developing brain that disrupt the brain's ability to control movement and maintain posture and balance. The term cerebral refers to the brain; palsy refers to the loss or impairment of motor function."

## STRUCTURE AND FUNCTION

CP is a static encephalopathy (a non-progressive brain disorder) that usually manifests as a disorder of movement. Babies with cerebral palsy do not roll over, sit, crawl or walk normally; older children with CP can present with poor coordination, stiff and weak muscles.

CP is primarily an upper motor neuron or movement disorder – however, depending on the size and location of the brain lesion, CP may affect speech, swallowing, and/or cognition. Cognitive problems can be a feature of CP, but are not always found. Any injury that occurs to the brain before the age of 2 may cause cerebral palsy.

CP may be classified *physiologically* (Table 1), *anatomically* (Table 2), or *functionally*. The physiological classification is based on the affected location of the brain lesion. The anatomic classification is defined by the affected limbs. A functional classification is based largely on the individual's mobility. Because certain levels of function that might be described in a classification system – jumping, for example – are not expected to be present in very young children, functional classification is not maximally reliable until approximately 6 years of age or older.

*Table 1: Physiologic Classification of Cerebral Palsy*

| Type | Location of Lesion in Brain | Description |
|---|---|---|
| Spastic | Pyramidal structures | · Increased muscle tone, especially with rapid stretching<br>· Hyperreflexia<br>· Compared to other physiologic types, Spastic CP patients benefit more from orthopedic surgical treatments |
| Dyskinetic/ Athetoid | Extra-pyramidal structures | · Involuntary, choreoathetoid movements<br>· Dystonia<br>· Less frequent than in the past due to improvement in prenatal care with Rh-immune globulin treatment of Rh incompatibility |
| Ataxic | Cerebellum | · Impaired balance<br>· Impaired coordination<br>· Wide-based gait |
| Mixed | Combination of the above | |

*Table 2: Anatomic Classification of Cerebral Palsy*

| Type | Affected Anatomy | Notes |
|------|-----------------|-------|
| Diplegic | Lower extremities | ·     Often still have some upper extremity involvement but not as severe as lower extremities<br>·     Brain lesion is midline |
| Hemiplegic | Ipsilateral upper and lower extremity on one side of the body | ·     Walking ability almost universal |
| Quadriplegic | All four extremities | ·     Usually non ambulatory |

The "static" aspect of the encephalopathy implies that the lesion which affects the brain does not expand or change with time – and that is true. However, the musculoskeletal manifestations of CP are not necessarily static. Indeed, the musculoskeletal manifestations of CP often worsen as the child grows. This occurs because normal muscle development depends on the appropriate neurological signals – and in CP, the damaged upper motor neuron fails to provide those signals. Normal bone and joint development, in turn, depend on normal muscle function and development.

The static brain lesions may also give rise to seizures, which is an additional cause of morbidity (and sometimes mortality) in patients with CP.

In simple language, CP is caused by brain damage during development in utero, during birth, or during very early infancy. There are a multitude of known factors that are linked to a higher chance of causing CP (see "Risk Factors," below). In many cases, the cause of CP is unknown.

## PATIENT PRESENTATION

Children with CP will present with delayed developmental milestones, especially with independent sitting and ambulation.

The degree of severity varies greatly. Depending on the physiologic type of CP, general spasticity or dystonia and choreoathetosis (Involuntary muscle contractions causing writhing or twisting movements) may be appreciated.

Contractures of joints are common findings. Gait disturbances such as toe-walking, crouched gait, scissoring (Figure 1), among others, are frequently seen.

Figure 1: A scissor gait pattern. Relative tightness of the thigh adductors produces extreme adduction, causing the knees to cross in a scissors-like fashion.

Torsional deformities of the long bones may be present. These develop via a normal remodeling response to the abnormal muscle forces acting across the joints of developing bones.

People with cerebral palsy often have low bone mineral density and thin cortices of the long bones. Contractures may impede full development of the articular surfaces.

Common deformities include increased femoral anteversion and external tibial torsion. Often, knee valgus, pes planovalgus or pes equinovarus are also present.

The hips in people with cerebral palsy are often subluxated or dislocated posterosuperiorly, due to a combination of abnormal muscle tone leading to abnormal acetabular development, increased femoral anteversion, and hip adduction and flexion contractures.

Neuromuscular scoliosis is also common in CP patients, especially in those with high levels of functional impairment (Figure 2).

*Figure 2: Clinical photograph and anteroposterior radiographs in a patient with CP.*
*Note the severe scoliotic curve, increased kyphosis, and pelvic obliquity.*

## OBJECTIVE EVIDENCE

There are no laboratory tests with which to diagnose CP; it is a clinical diagnosis based on clinical manifestations.

As the child grows, radiographs of the spine, hips, lower/upper extremities, and feet may show the effects of abnormal bone and joint development due to the underlying upper motor neuron lesion.

The spine may show a scoliotic curve, which is usually long and C-shaped (as shown above in Figure 2).

The hips may show increased femoral anteversion with apparent coxa valga, shallow acetabula and femoral head subluxation or dislocation (Figure 3).



*Figure 3: Anteroposterior radiographic views of a patient with CP showing bilateral hip dislocations.*

Radiographs of the long bones may reveal evidence of abnormal axial plane malrotation due to abnormal growth and development.

The feet radiographs may reveal pes planovalgus, calcaneovalgus, or cavovarus deformities as well as hallux valgus and hallux interphalangeus (Figure 4).



*Figure 4: Anteroposterior and lateral radiographic views of a patient with CP and severe calcaneovalgus deformity of the hindfoot and ankle, as well as hallux valgus.*

Gait analysis – often paired with dynamic EMG – is routinely used as objective evidence when evaluating cerebral palsy, specifically for the disorders of movement and gait. Results from gait studies can guide surgical treatment in patients with CP.

## EPIDEMIOLOGY

CP is the most common cause of chronic disability in childhood. It occurs in about 1 to 3 per 1000 live births. However, if the child is born prematurely or with low birth weight, the incidence increases to around 90 per 1000 live births. The incidence may be slightly higher in families of lower socioeconomic status, although this difference is small.

The incidence of CP has remained stable in recent decades, despite improvements in neonatal care that have increased the survival rates among babies at risk for perinatal brain damage. These greater survival rates have been matched with improvements in perinatal care that help avert anoxic brain injury and CP.

## DIFFERENTIAL DIAGNOSIS

The spine and hip pathology seen in CP patients can be seen in other neuromuscular disorders. However, proper history and physical exam can often distinguish between the various diagnoses.

A condition called familial spastic paraparesis may also look very similar to CP phenotypically. In this inherited condition, affected patients display weakness and spasticity, especially in the lower extremities. This condition, in contrast to CP, is progressive – that is, weakness and spasticity usually worsen as the patient ages.

## RED FLAGS

Seizure activity can be seen in patients with CP. This may lead to further brain damage if left untreated. Choking and trouble swallowing may be present in those patients with CP who also have bulbar involvement. This may require G-tube placement for nutrition if the airway cannot be protected and there is a risk of aspiration. (Note that some children who can swallow normally may nonetheless benefit substantially from G-tube placement, simply as a means to ensure adequate nutrition.)

## TREATMENT OPTIONS AND OUTCOMES

The treatment of patients with CP is often complex and multi-disciplinary. Musculoskeletal providers treating CP patients often follow them from infancy through adulthood. Apart from assisting with the coordination of care to other providers, the musculoskeletal needs change throughout the life of the child living with CP.

The skills and services of physical therapists, occupational therapists, and orthotists are central to the treatment of children with CP. Physical therapy (PT) serves to help train and maintain mobility, which often relies on the use of aids such as crutches or walkers. PT programs focusing on stretching and specific muscle strengthening to help with ambulation and mobility are important, as are the use of various braces and orthoses to help prevent joint contractures and maintain appropriate joint alignment for ambulation. The treating orthopedist should also check at each visit to ensure that orthoses are still fitting appropriately, as children may grow out of their orthoses, or sometimes due to worsening deformity.

Medical treatment of the neuromusculoskeletal manifestations of CP is widely used. Common medications prescribed to combat global spasticity include baclofen and diazepam. Baclofen may be taken orally or administered intrathecally with an implantable pump to non-ambulatory patients. (Intrathecal pumps deliver the medication directly to the central nervous system and hence decrease the risk of systemic side effects.) Diazepam is taken orally. Both of these medications allow the musculature to relax but may lead to profound sedation if the dosage is too high. A neurologist skilled in the treatment of CP should follow these patients closely to help and strike the right balance between muscle relaxation and sedation.

Botulinum toxin A ("Botox") may also be given to treat spastic CP patients. This medication is injected at the neuromuscular junction, and acts as an irreversible inhibitor to proteins at the axon terminal responsible for the release of acetylcholine, thereby disrupting the neuromuscular signal chain and preventing muscle fiber depolarization. Even though the toxin acts in an irreversible manner, after 3-6 months the clinical effect wears off due to protein turnover in the cell and new, uninhibited protein targets replace the inhibited ones. Patients treated with botulinum toxin A are then usually prescribed a physical therapy and bracing regimen. It is important to note that this therapy is only intended for spasticity; it is ineffective alone to correct a fixed joint contracture (though it can be a helpful adjunct to physical therapy which can decrease contracture).

When scoliosis is present, bracing may be employed to assist with sitting balance. However, unlike for idiopathic scoliosis, bracing does not prevent progression in neuromuscular scoliosis. Furthermore, unlike idiopathic scoliosis, curves tend to progress in neuromuscular scoliosis after skeletal maturity.

Surgical treatment may become necessary when following patients with CP. Surgery may be performed in an effort to improve ambulatory function, improve standing ability, relieve pain, improve hygiene, prevent worsening scoliosis and thus respiratory decline, prevent pressure ulcers, and sometimes for cosmetic concerns.

Contractures may be treated with tendon or muscle releases or lengthening procedures, especially in the lower extremities to improve gait. The treating surgeon should be warned, however, to avoid overlengthening the Achilles tendon, which may convert one gait problem into another. Crouch gait can be treated with distal femoral extension osteotomies and patellar tendon shortening or advancements. In-toeing or external rotation deformities may be treated with de-rotational osteotomies at the site of pathology (tibia or femur). Foot deformities, such as cavovarus foot, pes planovalgus, calcaneovalgus, and/or hallux valgus may be treated with tendon transfers, osteotomies, and/or fusions, as appropriate (Figure 5). When performing tendon transfers, split tendon transfers are often recommended to prevent over-correction.
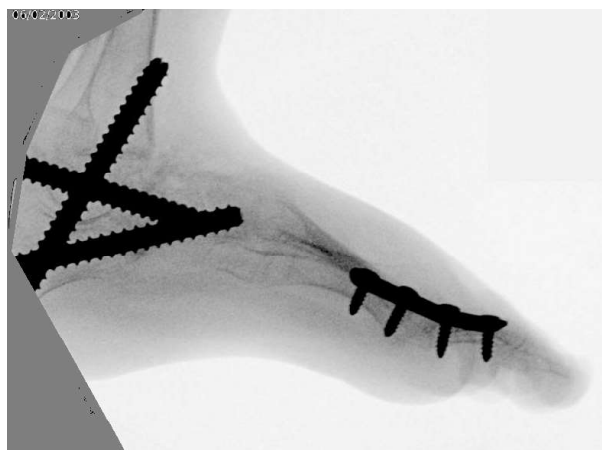
*Figure 5: Post-operative lateral fluoroscopic view of the patient shown in Figure 4. This patient underwent talectomy and hindfoot fusion to address the calcaneovalgus deformity of the hindfoot and ankle, and an MTP fusion to address the hallux valgus.*

Hip subluxation may be treated with adductor tenotomy, but only in mild cases in younger patients. In the setting of more severe subluxation, adductor tenotomy alone is not apt to be effective. Thus, when there is severe subluxation (or worse, when frank dislocation is present), femoral and/or pelvic osteotomies are often necessary in addition to adductor release. A common option for the femoral osteotomy is a varus de-rotation osteotomy of the proximal femur (Figure 6). Options for pelvic osteotomies include Dega osteotomy (if the triradiate cartilage is still open) or a Ganz versus shelf osteotomy (if the triradiate cartilage is closed). In addition, total hip replacement can also be used to address CP-associated hip arthrosis in adult patients (teens and older).
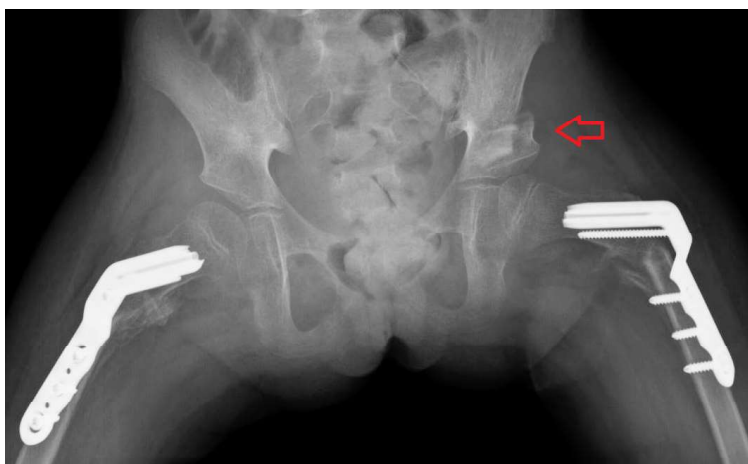


*Figure 6: Post-operative anteroposterior radiographic views of the patient shown in Figure 3. The bilateral hip dislocation was relocated with bilateral femoral varus denotational osteotomies of the femurs, as well as a Dega acetabular osteotomy on the left (red arrow).*

Scoliosis is often progressive and may be treated with posterior spinal fusion. Fusion to the pelvis is recommended if pelvic obliquity is present, especially in non-ambulators to improve wheelchair balance (Figure 7). Very stiff curves and large curves may require anterior release as well.

*Figure 7: Radiographs of the patient shown in Figure 2, after posterior spinal fusion from T2 to the pelvis.*

Life expectancy in CP is inversely correlated with the severity of disease. Patients with mild CP may have a normal life expectancy. By contrast, as many as 10% of severely affected patients will die in childhood. Respiratory failure is the most common cause of mortality in CP patients.

Outcomes following surgery are generally worse than outcomes for similar surgeries in unaffected patients. For example, the infection rate following spinal fusion surgery is much higher for CP patients than it is for idiopathic scoliosis patients.

## RISK FACTORS AND PREVENTION

Many known risk factors exist for CP (Table 3). Many of these environmental risk factors can be mitigated before & during pregnancy. However, controlling for these risk factors does not completely reduce the chances of being born with CP to zero.

*Table 3: Risk Factors of Cerebral Palsy.*

| | |
|---|---|
| **Prenatal Factors** | • TORCH infections<br>   ◦ **Toxoplasmosis**<br>   ◦ **"Other"** Infections (chiefly syphilis but also varicella-zoster, parvovirus B19)<br>   ◦ **Rubella**<br>   ◦ **Cytomegalovirus**<br>   ◦ **Herpes**<br>• Placental complications<br>• Maternal Epilepsy<br>• Maternal Drug and Alcohol Abuse<br>• Teratogenic medications<br>• Radiation exposure<br>• Rh incompatibility |
| **Perinatal Factors** | • Prematurity (birth prior to 37 weeks gestation) – most common<br>• Low birth weight ( < 1,500 g)<br>• Chorioamnionitis<br>• Anoxia |
| **Postnatal Factors** | • Meningitis<br>• Anoxia<br>• Intraventricular hemorrhage<br>• Brain trauma (including child abuse) |

## MISCELLANY

Although the term "cerebral palsy" was not used until the late 1800s, the condition has been described since ancient times. William Osler was the first to coin the term "cerebral palsy." During the polio epidemic, cases of polio and cerebral palsy were often confused, and CP was thought to be due to the polio virus to some extent.

# SPINAL MUSCULAR ATROPHY

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Spinal muscular atrophy is a disease of progressive muscle wasting and motor weakness. In spinal muscular atrophy, the alpha motor neurons in the anterior horn of the spinal cord that are responsible for skeletal muscle control deteriorate and are lost over time. Affected individuals are plagued by muscle weakness that progresses in a proximal to distal fashion. As the motor neurons deteriorate, muscle wasting ensues. Three major types exist (Table 1) and differ based on age of onset and clinical severity of symptoms.

*Table 1: Types of Spinal Muscular Atrophy*

| Spinal Muscular Atrophy Type | Alternate Name | Age of Onset | Description |
|---|---|---|---|
| I | Werdnig-Hoffman disease | Birth | ·    Most severe involvement of spinal muscular atrophy types<br>·    Death by 2 years of age |
| II | Dubowitz disease | 6-18 mo. | ·    Able to sit (some can stand) but not walk<br>·    Commonly reach adulthood |
| III | Kugelberg-Welander disease | After 18 mo. | ·    Least severe of spinal muscular atrophy types<br>·    Normal life expectancy<br>·    Can walk, although often become wheelchair-bound later in life |

## STRUCTURE AND FUNCTION

Spinal muscular atrophy is a genetic disease inherited in an autosomal recessive manner. It is caused by a mutation in the SMN1 (survival motor neuron) gene on chromosome 5 that codes for the so-called survival motor neuron protein – so-called, because without it, motor neurons do not survive.

Even if the SMN1 gene is not functional, a similar gene, SMN2, found nearby on chromosome 5, can code for the production of some survival motor neuron protein. The SMN2 gene is able to produce only about 10-20% of the normal amount of SMN protein (Figure 1). People ordinarily have 2 to 4 copies of the SMN2 gene. The more copies of SMN2 that affected people have, the more SMN protein they produce and the milder their clinical presentation will be. Nonetheless, even with many copies of the SMN2 gene – and some people have up to 8 – the levels of SMN protein are still below normal if the main SMN1 gene is not functional.
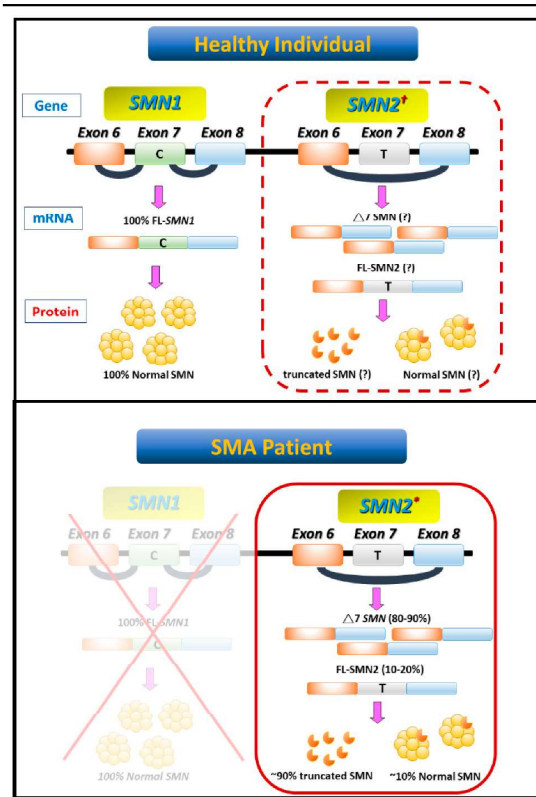
*Figure 1: Genetic phenotype-genotype correlation of spinal muscular atrophy (SMA). In a healthy individual, full-length (FL) survival motor neuron (SMN) mRNA and protein arise from the SMN1 gene. Patients with SMA have homozygous deletion or mutation of SMN1 but retain at least one SMN2; and about 10% of SMN2 transcripts produce full-length (FL) survival motor neuron (SMN) mRNA and protein. (Courtesy of Int. J. Mol. Sci. 2020, 21(9), 3297; https://doi.org/10.3390/ijms21093297)*

## PATIENT PRESENTATION

Patients with spinal muscular atrophy type 1 are the most severely affected, with age of onset at birth and rapid progression, often leading to respiratory failure and death within the first two years of life (Figure 2).
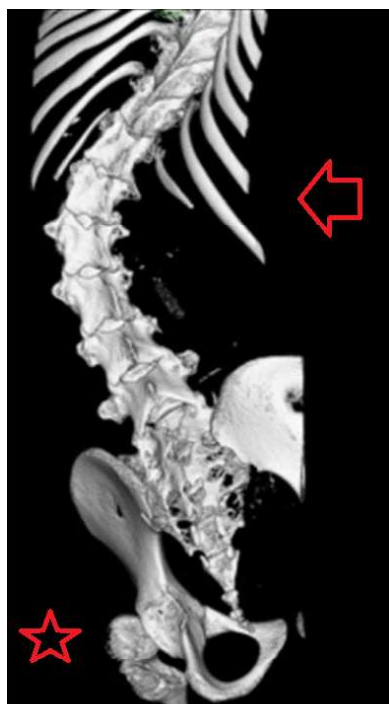


*Figure 2: A reconstructed three-dimensional CT image of a person with spinal muscular atrophy type 1. Note the marked scoliosis (red arrow) but also the dysplasia of the hip (red star). (Courtesy of JA Clin Rep 6, 28 (2020). https://doi.org/10.1186/s40981-020-00334-7)*

In these patients, deep tendon reflexes are absent, and muscle fasciculations may be seen. The spinal musculature is severely affected, leading to poor trunk control, often termed "floppy baby syndrome."

Patients with spinal muscular atrophy type 2 present later, between 6-18 months of age, and have a less severe and less rapidly progressive course. The proximal musculature is affected first, and progresses in a distal manner. Almost all patients will develop scoliosis as weakness progresses (Figure 3). The curve pattern is usually typical of a neuromuscular curve, but is often very flexible compared to scoliotic curves seen in other conditions. Soft tissue contractures around the hip, knee, and ankle are common. Hip dislocations (Figure 4) and equinovarus feet are often encountered in these patients as well. Patients usually do not have the ability to stand independently and rely on the use of a wheelchair for mobility. Deep tendon reflexes are absent.



*Figure 3: Clinical photograph and anteroposterior radiographic view of a patient with spinal muscular atrophy type 2, demonstrating severe scoliosis.*
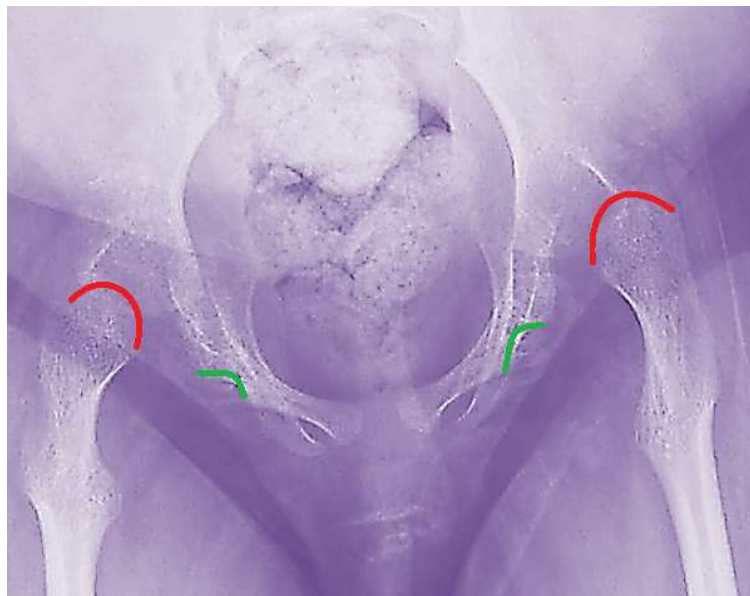


*Figure 4: Anteroposterior radiographic view of the pelvis of a patient with spinal muscular atrophy type 2, demonstrating bilateral hip dysplasia and dislocation. (The femoral heads are outlined in red and the approximate location of where the acetabulum should be is noted in green.)*

Spinal muscular atrophy type 3 patients are the least severely affected, and symptoms present later, during childhood or early adolescence. These children usually maintain the ability to stand independently but will usually become wheelchair-bound as an adult.

## OBJECTIVE EVIDENCE

The diagnosis is suspected based on clinical signs and symptoms, but since spinal muscular atrophy is caused by a genetic mutation in the SMN1 gene, DNA analysis confirms the disease.

Radiographs of the affected spine will show the scoliotic deformity, which is usually a long, sweeping curve commonly with associated pelvic obliquity. Hip radiographs reveal dysplasia, signs of acetabular dysplasia ranging from mild uncovering with instability to dislocation. Generalized osteopenia may be present in non-ambulatory patients.

## EPIDEMIOLOGY

Spinal muscular atrophy, an autosomal recessive genetic disease, is seen in one in 10,000 live births. It is the most common cause of death due to a genetic disease in infants. There is no differential risk based on ethnic group.

## DIFFERENTIAL DIAGNOSIS

Spinal muscular atrophy may be confused phenotypically with muscular dystrophy, and indeed it wasn't until the mid-20th century that spinal muscular atrophy was distinguished as a separate entity. One classic differentiating factor between muscular dystrophy and spinal muscular atrophy is the absence of deep tendon reflexes in spinal muscular atrophy, with maintenance of the reflexes in muscular dystrophy.

## RED FLAGS

Shortness of breath and respiratory distress is a common cause of death in spinal muscular atrophy type 1 infants and may require ventilator support in individuals with type 2 and 3 disease.

Signs of choking and difficulty swallowing is common as the disease progresses, which may require speech therapy support.

## TREATMENT OPTIONS AND OUTCOMES

Medical treatment of spinal muscular atrophy has evolved over the past few years. Nusinersen, a medication that is injected intrathecally (into the spinal canal such that it reaches the cerebrospinal fluid), has been shown to slow the progression of disease in spinal muscular atrophy patients. Nusinersen acts on the SMN2 gene to modulate the splicing of the RNA transcript, leading to increased production of SMN protein.

In 2019, a gene therapy treatment option was approved in the United States: onasemnogene abeparvovec (Zolgensma). The adenovirus-like vector is given intravenously and contains the SMN1 gene. It is currently approved for patients under 24 months of age.

Ventilator support is a critical aspect of treatment as spinal muscular atrophy patients suffer from respiratory muscle atrophy. Much like patients with muscular dystrophy, nighttime ventilation is often required either via tracheostomy or CPAP/BiPAP.

Patients with spinal muscular atrophy type 2 may develop progressive scoliosis. Bracing is sometimes used in an effort to slow progress but will not halt it. In young patients, often those with curves greater than 50

degrees are treated surgically to prevent respiratory compromise. Since younger patients still have substantial thoracic growth left, surgical strategies often involve placement of some sort of growing rod to allow for support and continued growth of the spine and chest wall. Once the patient reaches adolescence, the spine may be definitively fused with posterior spinal fusion. If scoliosis develops during adolescence, surgical treatment is usually performed for curves greater than 40 degrees, for progressive curves, or when lung function deteriorates.

Unique to spinal muscular atrophy patients, now that Nusinersen treatment is available, is to perform a laminectomy at a single level in the lumbar spine to allow access for future intrathecal administration of the medication. Anterior release is rarely necessary in spinal muscular atrophy patients.

Treatment of the hip dysplasia in spinal muscular atrophy patients remains controversial. Many advocate for observation, as dislocations tend to be asymptomatic and the recurrence rate after open reduction is higher than for idiopathic hip dislocations. If pain is present, the decision may be made to treat the dislocation surgically, with soft tissue releases and femoral/pelvic osteotomies.

For soft tissue contractures of the lower extremity, optimal treatment also remains controversial. If the patient is ambulatory, knee flexion contractures may be addressed with hamstring lengthening, and foot and ankle deformities addressed with tendon lengthening procedures. However, many soft tissue contractures are left alone and observed, especially in non-ambulatory patients.

Life expectancy in spinal muscular atrophy varies depending on the subtype. Spinal muscular atrophy type 1 patients usually die by 2 years of age. Type 2 patients may be expected to live to young adulthood. Type 3 patients have approximately normal life expectancy. Improvements in respiratory care and support have served to increase the life expectancy of these patients.

Nusinersen leads to improved muscle function while retarding disease progression. Long-term outcomes of the medication wait to be seen, as the medication has only been approved for use in spinal muscular atrophy patients within the last 4 years.


## RISK FACTORS AND PREVENTION

Because spinal muscular atrophy is inherited in an autosomal recessive manner, if there is no known family history of the disease, parents may be unaware of their carrier status. Accordingly, spinal muscular atrophy was added to the Recommended Uniform Screening Panel for newborns in 2018. (This panel is a list of disorders that the federal government recommends for states to screen as part of their newborn screening programs.)


## KEY TERMS

Spinal Muscular Atrophy, Survival motor neuron, Werdnig-Hoffman disease, Nusinersen, Zogensma