Sanjeev Sabharwal
Christopher A. Iobst
Editors

Pediatric Lower Limb Deformities

Principles and Techniques of Management

Second Edition

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ISBN 978-3-031-55766-8 ISBN 978-3-031-55767-5 (eBook)
https://doi.org/10.1007/978-3-031-55767-5

This Springer imprint is published by the registered company Springer Nature Switzerland AG
The registered company address is: Gewerbestrasse 11, 6330 Cham, Switzerland

Preface

Welcome to the second edition of *Pediatric Lower Limb Deformities: Principles and Techniques of Management*. In line with the goals of the previous edition, this updated textbook comprehensively covers all aspects of pediatric lower limb deformity from the hip to the toes. The author of each chapter was carefully chosen as an international expert for the assigned topic and was asked to provide current, state-of-the-art management principles for the reader to use in their day-to-day clinical practice. This up-to-date treatise is presented in a format that not only provides valuable fundamental information for the young surgeon but also covers nuances, including practical tips and tricks that will be relevant to the advanced limb deformity surgeon as well.

There are two special features of this second edition that are worth highlighting. First, besides updating the content of each chapter, ten entirely new chapters have been added including those highlighting Patient Reported Outcome Measures (PROMs) in limb lengthening and deformity correction; how to set up a limb deformity practice; pin site care and management of bone defects. The second update involves supplementing the content of each of the 40 chapters with an invited commentary from another authority in the field. In this way, the reader gets the benefit of at least two different perspectives on the topic as well as additional pearls and insights from experts in the field.

Finally, on reviewing the Table of Contents for the list of contributors, you will notice that this unique textbook has a decidedly international flavor. In order to make this work relevant to a broad audience of limb reconstruction surgeons and trainees, we have made a concerted effort to include perspectives from surgeons practicing in regions with varying degrees of resource availability and unique clinical pathology. Thus, we purposely invited contributions from a diverse group of professionals who were not only content experts in the assigned topic but also practiced in different clinical settings. Our goal was to tap into the incredible pediatric limb reconstruction work being done around the globe and to share this amazing repository of resources with you, the reader. We both have thoroughly enjoyed preparing this second edition and have learned an enormous amount from our colleagues and hope that you will too.

Oakland, CA, USA — Sanjeev Sabharwal
Columbus, OH, USA — Christopher A. Iobst

Acknowledgments

Acknowledgement by Sanjeev Sabharwal, MD, MPH (Director, Limb Lengthening and Complex Reconstruction Center, UCSF Benioff Children's Hospital, Oakland, CA, USA)
This second edition would not have been possible without the professional relationships and friendships that were developed over the years with members of the Limb Lengthening and Reconstruction Society (LLRS), Pediatric Orthopaedic Society of North America (POSNA), and Paediatric Orthopaedic Society of India (POSI). I am thankful to the residents, staff, and faculty of the Department of Orthopaedics at the University of California, San Francisco, who have taught me the value of patience and persistence. I have been lucky to have the institutional and academic infrastructure and an awesome team, here at UCSF Benioff Children's Hospital in Oakland, California.

The constant support of my dear friend and co-editor, Chris Iobst and the Springer publishing team, especially Kristopher Spring was vital in making this second edition a reality.

I am extremely grateful to my parents, grandparents, mentors, and students for helping me recognize the importance of integrity and hard work. Thanks to my dearest wife, Ranjit, who for the past 35 years took care of essentially everything so that I could pursue an academic career in pediatric orthopedics and limb reconstruction. I appreciate our three lovely children, Samir, Simran, and Sabhyta and their growing families for keeping me honest and grounded.

Acknowledgement by Christopher A. Iobst, MD (Director, Center for Limb Lengthening and Reconstruction, Nationwide Children's Hospital, Columbus, OH, USA)
I would like to reiterate Sanjeev's message of appreciation and gratitude to the entire international limb lengthening and reconstruction community. I am eternally indebted to the generosity and kindness of my mentors and countless colleagues who have guided and educated me along the way. I am constantly impressed by the spirit of kindness and willingness to help displayed throughout the entire limb reconstruction field. This second edition is an example of that collaborative power and sharing of knowledge. We hope it will continue to provide valuable reference and education to surgeons both young and old.

Personally, I must thank the Center for Limb Lengthening and Reconstruction team at Nationwide Children's Hospital for their hard work and dedication to taking care of patients with limb deformities. We have been able to make our dream of establishing a Center that provides comprehensive care for patients and their families become a reality. As we recently celebrated our eighth birthday, I would especially like to thank Danielle, Ashley, Cheri, and Jessica who have been critical, invaluable team members from the start.

Finally, I would like to thank my family. My parents, for their lifelong support and for teaching me the value of hard work and education. I could never have been able to follow this path without them. To my wife, Sybil, for providing the peaceful atmosphere, understanding and love necessary to complete the time-consuming process of editing a reference textbook.

Contents

Part II Related Concepts and Management Options

Part III Underlying Conditions

Part IV Congenital and Developmental Disorders

Part V Sequelae and Complications

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Part I

General Principles and Techniques

Etiology of Lower Limb Deformity

1

Allyson Zakrzewski and Viral V. Jain

Etiologies of lower extremity deformity include conditions that result in limb length inequality, angular deformity, and/or asymmetric girth. There are many different conditions that can result in deformity in children. Despite some overlap, categorizing them can help simplify diagnosis and treatment. Pediatric limb deformities can be classified into four main etiologic groups: (1) underlying conditions, (2) congenital, (3) developmental, and (4) acquired. Table 1.1 demonstrates the wide variety of etiologies that contributed to limb length inequality, deformity, and asymmetric girth.

Table 1.1 General categorization of lower limb deformity in children

Underlying conditions with examples	
Metabolic	Rickets
	Renal osteodystrophy
	Endocrinopathies (hypothyroidism, growth hormone deficiency)
Genetic conditions	Osteogenesis imperfecta
	Neurofibromatosis
	Skeletal dysplasia's
	Arthrogryposis
	Larsen Syndrome
Tumor and tumor-related conditions	Benign tumors
	Malignant tumors
	Fibrous dysplasia
	Ollier's syndrome
	Multiple hereditary exostosis
Inflammatory conditions	Juvenile idiopathic arthritis
	Hemophilic arthropathy
Congenital	
Limb deficiencies	Congenital femoral deficiency (proximal focal femoral deficiency)
	Congenital fibular deficiency (fibular hemimelia)
	Congenital tibial deficiency (tibial hemimelia)
Tibial bowing	Posteromedial bowing
	Anteromedial bowing
	Anterolateral bowing (congenital pseudarthrosis)
Others	Coxa vara
	Congenital knee dislocation
	Congenital patella dislocation
	Hemihypertrophy
Developmental	
Genu varum	Physiologic varus
	Blount's disease—Infantile, adolescent
	Focal fibrocartilaginous dysplasia
Genu valgum	Physiologic valgus
	Idiopathic genu valgum
Acquired	
Residual deformity	Slipped capital femoral epiphysis
	Legg-calve perthes disease
	Developmental dysplasia of the hip
	Avascular necrosis
Post-traumatic	Physeal arrest from fracture involving the growth plate
	Fracture malunion
	Cozen's phenomenon
	Femoral overgrowth
Post-infectious	Physeal arrest from osteomyelitis
	Physeal arrest from septic arthritis
	Physeal bar equivalent
	Meningococcal septicemia
Iatrogenic	Repetitive and aggressive physeal fracture reduction
	Contracture manipulation
	Surgery at or around the physis
	Physeal injury following ligamentous reconstruction

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S. Sabharwal, C. A. Iobst (eds.), *Pediatric Lower Limb Deformities*, https://doi.org/10.1007/978-3-031-55767-5_1

Underlying Medical Conditions

Metabolic Disorders

Endocrinopathies and metabolic disorders can affect normal regulatory signals necessary for chondrocyte maturation and new bone formation. Disorders such as hypothyroidism and growth hormone deficiency have been shown to weaken the physis and can result in conditions such as slipped capital femoral epiphysis (SCFE) [1]. Compressive forces across these weakened growth plates can lead to the development of angular deformities.

Rickets

Rickets is a clinical manifestation of defective mineralization of the physis secondary to disruption of calcium/phosphate homeostasis. This can be related to genetic as well as nutritional causes. Hypophosphatemic rickets is a Vitamin D resistant form that is caused by the inability of renal tubules to absorb phosphate. While this can be inherited in an autosomal dominant and autosomal recessive fashion, the most common form has an X-linked dominant inheritance. This group of disorders results from a mutation in the phosphate-regulating endopeptidase X-linked gene (PHEX), which is expressed in osteocytes. Mutation in this gene leads to increased levels of fibroblast growth factor 23 (FGF 23), leading to decreased renal phosphate absorption and suppression of (25)-OH-1α hydroxylase activity [2]. Vitamin D-dependent rickets is less common and is related to mutations of (25)-OH-1α hydroxylase.

Nutritional causes are important to consider as well (Fig. 1.1). Risk factors that predispose children to rickets and Vitamin D deficiency include poor nutritional intake, premature birth, dark skin, living in higher latitudes with limited sun exposure, obesity, and malabsorption syndromes (celiac disease, pancreatic insufficiency, and biliary obstruction). Exclusive breast-feeding particularly in non-white children has also been associated with the development of nutritional rickets [3]. The different forms of rickets have similar skeletal manifestations often presenting with lower extremity angular deformity and decreased longitudinal growth with radiographic findings including widened irregular physis and cupped or flared metaphysis.

Renal Osteodystrophy

Renal osteodystrophy occurs secondary to end-stage renal disease and results from the kidney's inability to produce adequate amounts of 1, 25 dihydroxyvitamin D3 as well as phosphate retention. The elevated phosphate levels lead to hyperparathyroidism, which can result in brown tumors and subperiosteal erosions. In children, renal osteodystrophy can lead to growth retardation as well as angular deformities. In addition to the development of valgus or varus lower extremity alignment (typically at the knee but also at the ankle), children with renal osteodystrophy are also at risk for the development of SCFE. The severity of skeletal deformity does not correlate with the degree of control of the renal disease [4].

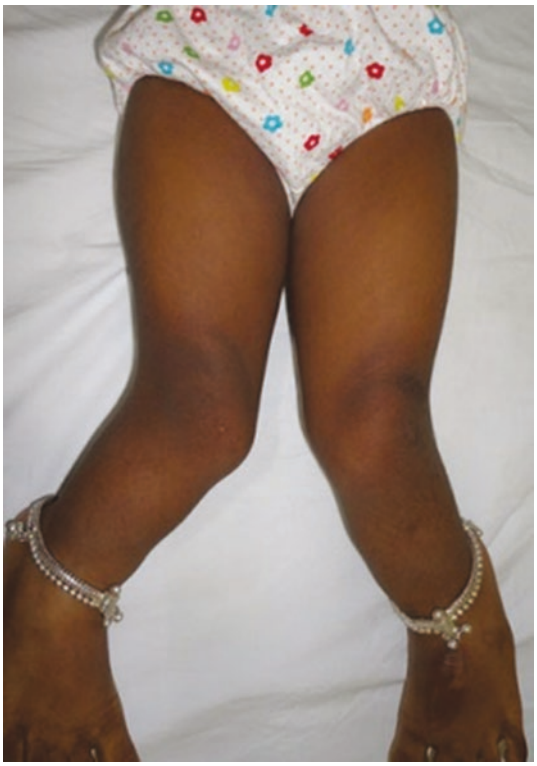

Fig. 1.1 Genu valgum in a 7-year-old female secondary to nutritional rickets

Genetic Disorders

Osteogenesis Imperfecta

Osteogenesis imperfecta is broadly used to describe a group of genetic disorders that result in increased bone fragility and low bone mass. The most common mutations are in COL1A1 and COL1A2, which encode alpha chains of type I collagen. Clinical presentation varies but patients frequently present to an orthopedic surgeon with lower extremity fragility fractures. The underlying disorder results in poor remodeling potential during healing, and multiple fractures can result in deformity of long bones, which can be in a single plane or multiple planes. Additionally, these children typically have short stature and coxa vara. Bisphosphonates are often used for treatment to increase bone mineral density with the goal of reducing overall fracture rates [5].

Neurofibromatosis

Neurofibromatosis (NF) is a group of genetic disorders involving products of all three germ lines: neuroectoderm, mesoderm, and endoderm. The most orthopedically relevant disorder in this group is Neurofibromatosis Type 1, which previously had been known as von Recklinghausen disease. This is an autosomal dominant disorder with a prevalence rate of 1:4000. It occurs equally in all ethnic groups and 50% are a result of new genetic mutations. The clinical presentation itself is variable with some patients having subclinical to severe manifestations. The associated gene is located on chromosome 17 (17q) and affects neurofibromin, which acts as a tumor suppressor gene [6].

Diagnosis of neurofibromatosis is made by several distinguishing clinical features (Table 1.2). Orthopedic clinical manifestations include tibial dysplasia and hemihypertrophy, which can lead to limb length discrepancies and limb deformities. Both are discussed in greater detail later in the chapter. In addition to lower extremity deformity, spinal deformities are commonly seen in children with neurofibromatosis. Spinal deformities remain the most common orthopedic manifestation with rates of incidence reported around 30% [6]. All patients with neurofibromatosis should be screened for scoliosis during orthopedic evaluation.

Table 1.2 Diagnostic criteria of neurofibromatosis

Diagnostic Criteria of Neurofibromatosis Type 1 (> 2 criteria for diagnosis)
Six or more café au lait spots measuring at least 15 mm in adults and 5 mm in prepubertal individuals (Fig. 1.2)
Two or more neurofibromas of any type or one plexiform neurofibroma
Freckling in the axillary or inguinal region
Optic Glioma
Two or more Lisch nodules (iris Hamartomas)
A distinctive bony lesion (sphenoid wing dysplasia, thinning of long bone cortex, anterolateral bowing of the tibia, or pseudoarthrosis of a long bone)
A first-degree relative with NF1

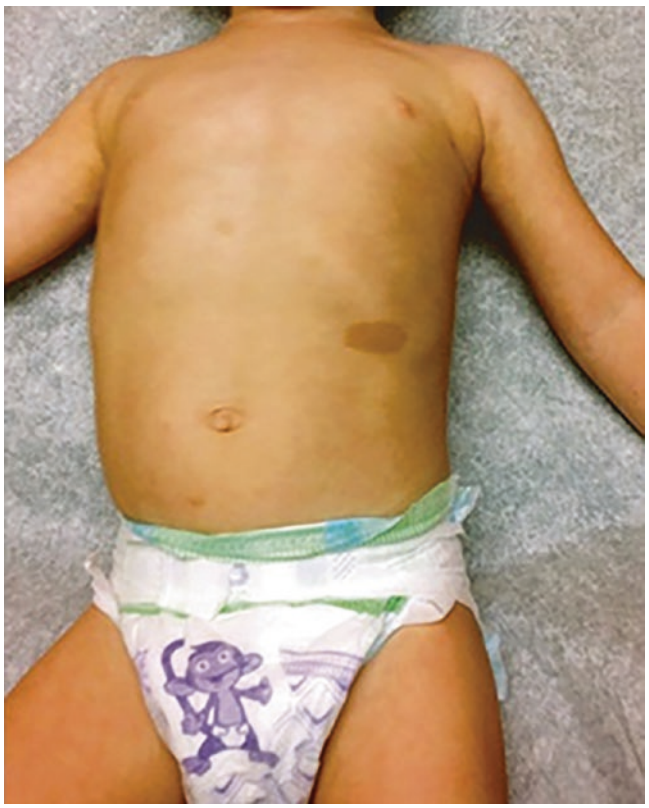

Fig. 1.2 Three-year-old female with neurofibromatosis type 1 (NF1). Café au lait spots are seen on the abdomen

Neuromuscular Conditions

Cerebral Palsy

Cerebral palsy is a static encephalopathy that results secondary to an injury to the premature brain. This results in cognitive and musculoskeletal manifestations. From an orthopedic perspective, the presentation of children with cerebral palsy is varied and is based on the presence, distribution, and severity of underlying spasticity. Tone imbalances can lead to the development of deformities of the lower extremity including rotational and angular deformities along with joint contractures.

Charcot Marie Tooth

Charcot Maire Tooth disease (CMT) is the most common hereditary motor and sensory neuropathy. There are many described subtypes with the most common subtype being a duplication of the peripheral myelin protein 22 gene (PMP 22) on chromosome 17, which is inherited in an autosomal dominant fashion. Demyelination of peripheral nerves and dorsal root ganglia is progressive and leads to muscle atrophy as well as loss of proprioception and deep tendon reflexes. Weakness in the peroneus brevis, tibialis anterior, as well as intrinsic muscles of the foot results in the most common presentation of a cavovarus foot with claw toes (Fig. 1.3). Other musculoskeletal manifestations include calf atrophy, scoliosis, hip dysplasia, as well as upper extremity intrinsic wasting, resulting in weak pinch and grasp [7].

Arthrogryposis and Related Syndromes

Arthrogryposis multiplex congenital is a heterogeneous group of conditions that present as nonprogressive contractures involving at least two joints at more than two different areas of the body at the time of birth. While not well understood, arthrogryposis is thought to be a genetically mediated syndrome that is related to decreased fetal movement during in-utero development. Hips are commonly involved with typical flexion and abduction contractures with hip

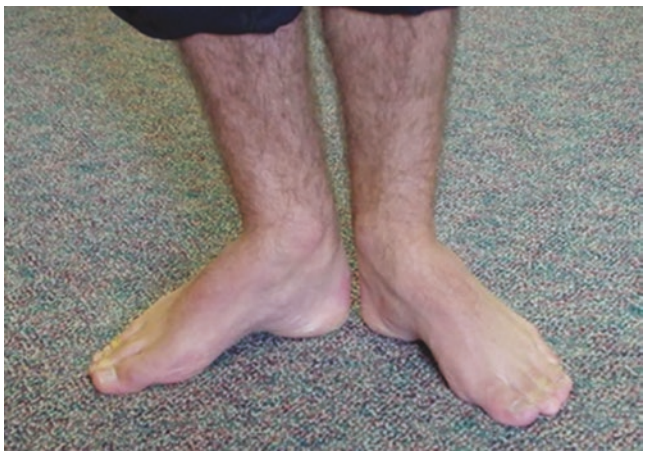

Fig. 1.3 Seventeen-year-old patient with Charcot Marie Tooth Disease (CMT). Cavus deformity of the foot is seen bilaterally

dislocation noted in 15–30% of cases. Involvement of the knees can be with either flexion or extension contractures, although flexion contractures are more common. Foot deformities are frequent with 80–90% of children with arthrogryposis having a clubfoot or congenital vertical talus [8].

Larsen syndrome is a rare disorder that is characterized by multiple joint dislocations that are noted at birth. Mutation has been noted in the FLNB gene, which encodes for filamin B, a cytoskeletal binder that helps chondrocytes differentiate and proliferate [9]. Dislocations of the hips, knees, and cervical spine abnormalities are commonly seen. Clubfoot deformities are rigid and can be difficult to treat [10].

Multiple Pterygium syndrome is a spectrum of disorders, resulting in skin webbing of joints, commonly the elbows and knees. Most cases involve a mutation in the CHRNG gene, which encodes for the gamma subunit of the fetal acetylcholine receptor protein important in neuromuscular signaling. As the adult acetylcholine receptor gene predominates after 33 weeks gestation, joint contractures without concomitant muscle weakness are the typical presentation [11].

Skeletal Dysplasia

Osteochondral dysplasias are disorders of growth and development of cartilage and bone. There are numerous conditions included in the category of skeletal dysplasias with most variations resulting in short stature and angular deformities of the lower extremity. Relevant skeletal dysplasia, underlying genetic findings, lower extremity findings, as well as other orthopedic findings are shown in Table 1.3.

Bone Tumors

Benign bone tumors and tumor-like lesions can result in lower extremity deformity. This is related to pathologic fracture, proximity of the tumor to the physis, or as a result of treatment. Therapeutic curettage or injection can also result in damage to the adjacent growth plate. Additionally, radiation therapy can inhibit the growth of the physis by altering chondroblast activity [14]. The overall risk is related to the amount of radiation delivered, the size of the field, and the patient's overall growth potential. Malignant tumors frequently require wide resection often including the nearby physis, which can lead to an increasing limb length discrepancy with growth.

Polyostotic diseases can result in severe limb deformities. Multiple hereditary exostosis (MHE) is a condition that results secondary to the loss of function of EXT1 and EXT2 proteins, resulting in multiple osteochondromas. These patients typically have short stature and can develop valgus alignment of the hip, knee, and ankle. Fibrous dysplasia is a developmental abnormality that is caused by a mutation in the GS-α protein that leads to failure of the production of normal lamellar bone. While any bone can be involved, the proximal femur is the most common and leads to the characteristic Shepard's crook deformity. Additionally, progressive bowing deformities of the long bones are seen with fibrous dysplasia, and 60% of patients with polyostotic fibrous dysplasia have a reported limb length discrepancy. Ollier's disease is a form of multiple enchondromatosis, which produces islands of cartilage in the diaphyseal and metaphyseal regions of long bones. These lesions often encroach on the physis and can result in inhibition of growth. Typically, this involves one limb more than the other and, as such, can lead to both angular deformities and significant limb length discrepancy [15].

Inflammatory Conditions

Juvenile idiopathic arthritis is a group of conditions that is characterized by chronic joint inflammation of unknown etiology that lasts longer than 6 weeks in a child under the age of 16. This was previously referred to as juvenile rheumatoid arthritis but few children were found to be rheumatoid factor positive. Etiology is multifactorial and likely due to several environmental and genetic factors [16]. Contractures can develop from chronic inflammation and may need to be released. Overgrowth has been reported and is associated with younger age at the time of presentation [16].

Table 1.3 Relevant skeletal dysplasia's and orthopedic manifestations [12, 13]

Condition	Typical inheritance	Involved protein	Common lower extremity deformities	Other orthopedic manifestations
Achondroplasia	Autosomal dominant	Gain of function FGFR3	Rhizomelic dwarfism Genu varum	Lumbar stenosis Thoracolumbar kyphosis Foramen magnum stenosis Trident hand
Pseudoachondroplasia	Autosomal dominant	COMP	Hip dysplasia Genu valgum Genu varum Wind-swept deformity	Cervical instability Kyphoscoliosis Platyspondyly
Hypochondroplasia	Autosomal dominant	FGFR3	Mild short stature Rhizomelic dwarfism Genu varum	Spinal stenosis
Diastrophic Dysplasia	Autosomal recessive	SLC6A2 (sulfate transporter)	Joint contractures Genu valgum Patella dislocation Equinovarus feet	Hitchhiker's thumbs Cervical kyphosis Thoracic kyphosis
Kniest Syndrome	Autosomal dominant	COL2A1	Large stiff joints Equinovarus foot	Odontoid hypoplasia Kyphoscoliosis
Spondyloepiphyseal dysplasia congenita	Autosomal dominant	COL2A1	Short stature Coxa vara Genu valgum Equinovarus foot	C1–C2 instability Scoliosis Platyspondyly
Spondyloepiphyseal dysplasia tarda	X linked	COL2A1	Short stature	Platyspondyly
Multiple epiphyseal Dysplasia	Autosomal dominant	COMP, type IX collagen	Coxa vara Valgus deformity Joint contractures	Normal spine
MPS Type 1 (hurler)	Autosomal recessive	α-l-Iduronidase	Hip dysplasia Genu valgum	Kyphoscoliosis Carpal tunnel syndrome Gibbus deformity
MPS Type IV (Morquio)	Autosomal recessive	Keratin sulfate	Ligamentous laxity Genu valgum Progressive acetabular dysplasia	Odontoid hypoplasia C1–C2 instability Platyspondyly Carpal tunnel

Hemophilic arthropathy results in repeated intra-articular bleeding, leading to synovitis, joint contractures, and joint destruction. Recurrent hemarthrosis can result in lasting changes to chondrocyte activity and cartilage matrix integrity, leading to abnormalities in epiphyseal growth. Asymmetrical growth can result leading to angular deformities. Limb Length discrepancy can also result in related hyperemia [17].

Congenital Etiology

Hemihypertrophy and Hemiatrophy

Hemihypertrophy and hemiatophy are conditions that result in abnormal growth of the affected extremity. These conditions can be idiopathic or related to an underlying syndrome but, in general, lead to significant differences in limb girth and limb length equality. Figure 1.4 demonstrates the limb length discrepancy and asymmetrical girth seen in idiopathic hemihypertrophy. Overgrowth and undergrowth syndromes are detailed in Table 1.4.

Both syndromic and idiopathic hemihypertrophy have been associated with embryonal tumors, such as Wilm's tumor, adrenal cell carcinoma, and hepatoblastoma [18]. Genetic evaluation is important to identify subgroups of patients in which this risk is increased. Beckwith–Wiedemann syndrome has an incidence of embryonal tumors of 8.8%, and serial abdominal exams and ultrasounds are recommended every 3–4 months in this population until age 7 [19].

Congenital Femoral Deficiency

Congenital femoral deficiency is a spectrum of disorders that involve abnormalities in the femur. Proximal focal femoral deficiency (PFFD) is a distinct subset in which the defect is within the primary ossification center of the proximal femur. Variable deficiencies are noted with the mildest form, resulting in a short femur to more severe forms in which the proxi-

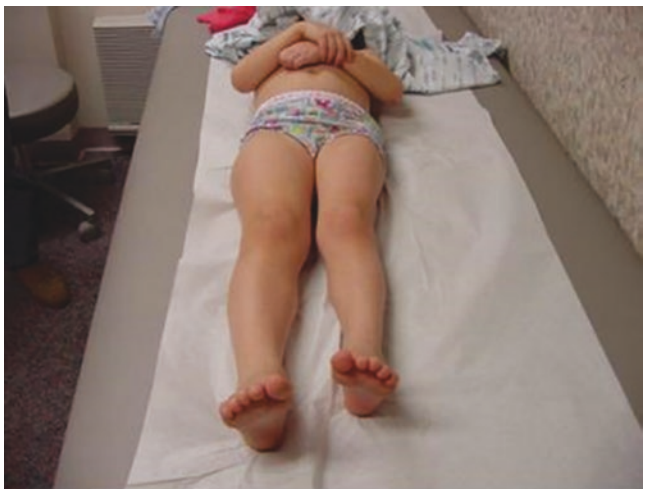

Fig. 1.4 Limb length discrepancy and asymmetrical girth in idiopathic hemihypertrophy

Table 1.4 Common overgrowth and undergrowth syndromes [20]

Syndrome	Genetic association	Common clinical features
Common overgrowth syndrome		
Beckwith–Wiedemann syndrome	Mutation of 11p15 leading to overactivity of the IGF-2 gene	Macroglossia, macrosomia, midline abdominal wall defects, neonatal hypoglycemia, and hemihypertrophy
Proteus syndrome	Somatic activating mutation in AKT1 kinase	Progressive macrodactyly, hemihypertrophy, thickening of the skin, lipomas, subcutaneous tumors, epidermal nevus, and macrocephaly
Neurofibromatosis type 1	Mutation of the NF1 gene located on chromosome 17 results in disruption of Ras signaling controlled by neurofibromin	Café-au-lait spots, cutaneous neurofibromas, axillary freckling, optic glioma, Lisch nodules, vertebral scalloping, tibial dysplasia, and plexiform neurofibroma
Klippel–Trenaunay syndrome	In some patients—angiogenic factor VG5Q, translocation (5:11, 8:14)	Nevus Flammeus (port-wine stain), venous and lymphatic malformations, soft tissue and bony hypertrophy
Common undergrowth syndrome		
Syndrome	Genetic association	Common clinical features
Russel–silver Syndrome	Loss of methylation on chromosome 11p15 (30–60%) Maternal uniparental disomy on chromosome 7 (10% of patients)	Short stature, small triangular faces, disproportionately normal head circumference, and clinodactyly
Cutis marmorata telangiectatica congenita	Unknown	Generalized or localized reticulated cutaneous vascular mottling, macrocephaly, glaucoma, hemiatrophy, vascular anomalies

mal femur is absent and the distal femoral shaft creates a pseudo articulation at the hip. Patients with PFFD present with a short thigh with the hip in a flexed, abducted, and externally rotated position (Fig. 1.5). Additionally, knee contractures are common, and this condition is also associated with ACL deficiency.

In most cases, the underlying genetic link is unknown although there is a form associated with abnormal facies (femoral hypoplasia—unusual facies syndrome), which is inherited in an autosomal dominant fashion [21].

Congenital Fibular Deficiency

This condition, also known as fibular hemimelia, consists of shortening or absence of the fibula along with variable involvement of the lateral bones of the lower extremity. This is the most common long-bone deficiency with an incidence between 7.4 and 20 cases per million live births [22]. The exact etiology is unknown and cases are typically sporadic. Graham et al. suggested that exogenous vascular or mechanical disruption of limb bud function on the apical ectodermal ridge may lead to fibular hemimelia [23]. Angiographic abnormalities have been identified as well [24].

Typically, patients with fibular hemimelia present with a short tibia, anteromedial bowing, and marked equinovalgus deformity of the ankle. Additional abnormalities associated with the condition are hypoplastic lateral femoral condyle, cruciate ligament deficiency, coxa vara, ball and socket ankle joint, tarsal coalitions, and absent lateral rays of the foot. Femoral deficiencies are often seen in conjunction with fibular hemimelia. While the magnitude of femoral shortening is variable, it often contributes to the overall limb shortening. In addition to limb length discrepancy, valgus alignment of the lower extremity often develops from the dysplastic lateral condyle, seen in 93% of children with fibular hemimelia [25].

Congenital Tibia Deficiency

Congenital tibia deficiency (Tibia Hemimelia) is a partial to complete absence of the tibia at birth. It is one of the rare congenital lower limb deformities. While most cases have an unknown etiology, there are some associated syndromes that have autosomal dominant inheritance patterns. Four different autosomal dominant syndromes have been identified and include Warner Syndrome (Tibia Hemimelia-foot polydactly–tripahlangeal thumb syndrome), Tibia hemimeliala–micromelia–trigonobrachycephaly syndrome, Tibial hemimelia diplopodia syndrome, and Tibial hemimelia split hand and foot syndrome. In syndromic forms of tibia deficiency, defects in the sonic hedgehog pathway have been identified [26, 27]. Inherited forms of tibial hemimelia are typically bilateral and are associated with other congenital anomalies. In a review, 79% of patients had associated abnormalities of the hip, hand, or spine occurring alone or in various combinations [28]. Unlike other longitudinal deficiencies, tibial hemimelia is associated with visceral involvement typically of the cardiac,

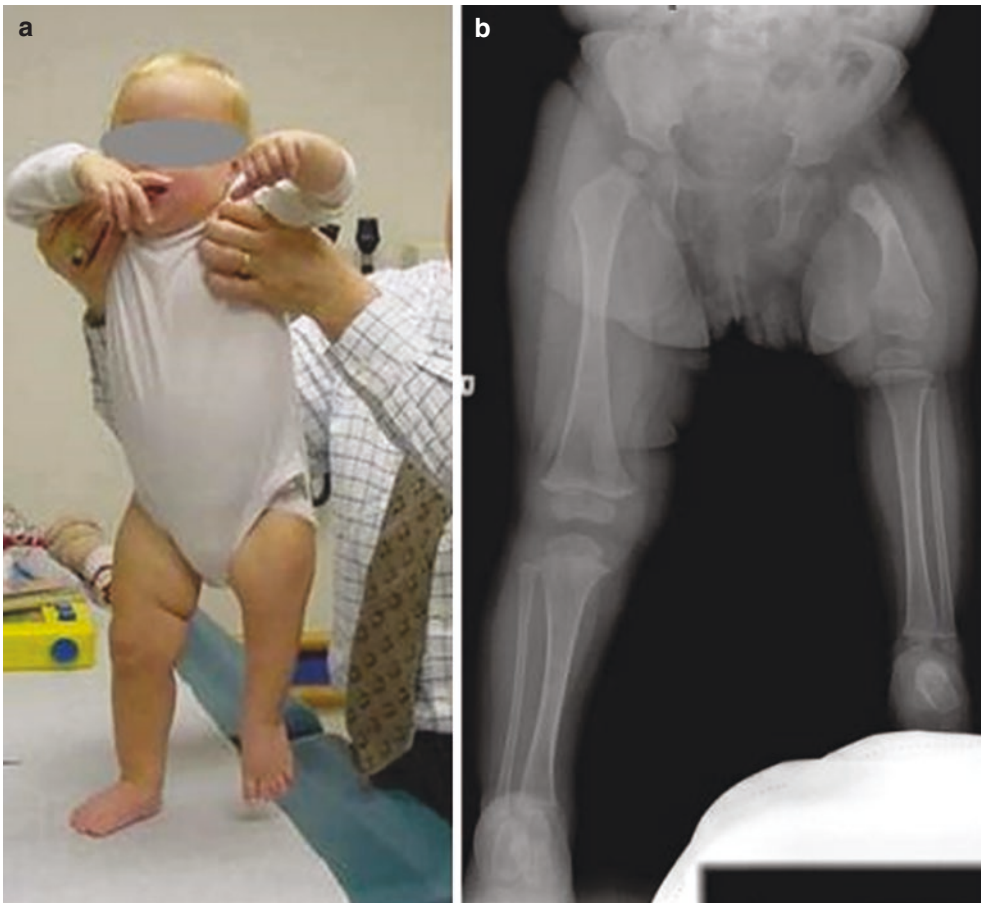

Fig. 1.5 (**a** and **b**) Clinical and radiographic findings of proximal focal femoral deficiency. Eighteen-month-old male with proximal focal femoral deficiency. Note the short femur and absence of the proximal femur on the left side

gastrointestinal, and genitourinary systems [28]. Presentation typically includes anterolateral bowing of the tibia, flexion contracture of the knee, and rigid equinovarus and supination deformity of the foot. Most children will have hamstring function, but presence of quadricep function is variable and related to the degree of absence of tibia.

Congenital Knee Dislocation

Congenital knee dislocation is the most severe form of congenital knee hyperextension. The overall incidence of congenital knee dislocation has an incidence of 1 per 100,000 [29]. It is readily identifiable at birth as there is marked hyperextension at the knee with the foot typically pointing toward the infant's mouth or shoulder (Fig. 1.6). This condition is thought to be secondary to abnormal fetal positioning. Once in an abnormal position, limited movement related to an underlying neuromuscular condition or hyperlaxity results in persistent hyperextension, which leads to quadriceps atrophy and fibrosis. Others have postulated that the absence of the cruciate ligaments is a key factor in the development of congenital knee dislocation [30].

Bilateral congenital knee dislocation is almost always syndromic in nature and has been associated with hyperlaxity syndromes (such as Larsen Syndrome, Beal's Syndrome, and Ehlers–Danlos syndrome) as well as underlying neuromuscular conditions (such as arthrogryposis, myelomeningocele). It is also important to assess for additional orthopedic manifestations including ipsilateral hip dislocation and club foot that are commonly seen in infants with congenital knee dislocation [30, 31].

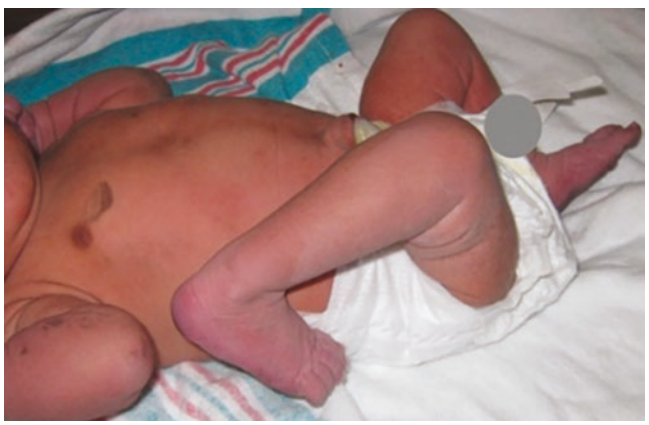

Fig. 1.6 Clinical presentation of congenital knee dislocation. Newborn with congenital knee dislocation. Initially treated with serial casting and eventually required quadricep release

Congenital Patella Dislocation

This is a relatively rare condition that presents with a laterally displaced and hypoplastic patella. This is present at birth and has a spectrum of severity. The most severe forms result in knee flexion contracture and are diagnosed in infancy while milder forms may not present until the child is older when functional problems related to quadricep weakness are noted. These children are typically present with genu valgum with significant external rotation of the tibia noted on exam. The patella is small and is often difficult to palpate in its lateral location [32].

Congenital patella dislocation is thought to be secondary to an embryologic cause. Normally, the quadriceps rotates from a lateral to an anterior position during fetal development. In children with a congenital patella dislocation, the quadriceps is short and found to be more lateral and a thickened iliotibial band is often present. This may prevent the normal rotation of the quadriceps during development and result in a dislocated patella at birth. Others have reported a possible hereditary link. Congenital patella dislocation has been associated with a variety of conditions including diastrophic dysplasia, arthrogryposis, Rubinstein-Taybi Syndrome, nail-patella syndrome, and Ellis van Creveld Syndrome [32, 33].

Coxa Vara

Congenital coxa vara is a developmental abnormality of the proximal femur that is characterized by a cartilaginous defect in the femoral neck. This leads to an abnormal decrease in the femoral neck-shaft angle, shortening of the femoral neck, relative overgrowth of the greater trochanter, and shortening of the affected limb. Overall the incidence is rare with 1 in 25,000 live births being affected [34]. While not all patients have progression, worsening of coxa vara is thought to occur secondary to excessive biomechanical stress on the abnormally positioned proximal femoral physis.

Presentation typically occurs after walking age and even into adolescence. Children with unilateral involvement typically present with easy fatigability or aching pain in the gluteal musculature, a Trendelenburg gait pattern, or mild limb length discrepancy. Bilateral involvement typically presents with a waddling gait with or without fatigue or muscular pain. Bilateral cases are associated with underlying conditions or skeletal dysplasias including Cleidocranial dysostosis, spondylometaphyseal dysplasia, and metaphyseal dysostosis (Jansen type). Coxa vara is thought to have a genetic component, as well as it can occur in families and twins [35].

In addition to congenital coxa vara, other causes include coxa vara secondary to congenital femoral deficiency and acquired coxa vara. These are considered separate entities as different radiographic and clinical findings are seen. Some causes of acquired coxa vara include slipped capital femoral epiphysis, sequelae of avascular necrosis from Legg–Calve–Perthes Disease, femoral neck fracture or traumatic dislocation, and septic necrosis. Additionally, coxa vara can be seen in underlying bone disorders including osteogenesis imperfecta, fibrous dysplasia, osteopetrosis, and renal osteodystrophy.

Congenital Bowing of the Tibia

Anterolateral bowing of the tibia is a condition seen at birth with an apical prominence of the lateral tibia. It is most frequently associated with tibial dysplasia or congenital pseudarthrosis. The pseudarthrosis is typically not present at birth but develops due to underlying disease process and deformation, resulting in a fracture that often progresses to a nonunion. Overall, this is a rare condition with an incidence of 1: 140,000—190,000. Anterolateral bowing and congenital pseudarthrosis have been associated with underlying conditions. Neurofibromatosis Type 1 is frequently associated with both conditions. While 5.7% of patients with neurofibromatosis have this tibial deformity, 55% of cases of anterolateral bowing and pseudarthrosis are associated with neurofibromatosis [36]. Additionally, fibrous dysplasia can be seen in 15% of children with anterolateral tibial bowing [36]. While rare, amniotic band syndrome can result in local constriction leading to the development of pseudarthrosis [37].

Posteromedial bowing of the tibia is a deformity that is present at birth and is typically seen in combination with calcaneovalgus foot, although they can occur independently (Fig. 1.7). The underlying cause is thought to be secondary to intrauterine malposition [38]. Typically, there is spontaneous but incomplete correction of bowing within the first 4 years of life. The most common sequelae of posteromedial bowing is a limb length discrepancy, and children should be followed until skeletal maturity [38, 39].

Anteromedial bowing is most commonly seen in association with congenital fibular deficiency as noted in the previous section.

Developmental

Physiologic Genu Varum

Most newborns are born with 10–15 degrees of varus angulation of the lower extremities. As the child stands and walks, this deformity can appear more prominent. Internal tibial torsion, which is also a common finding in this age group, can also exacerbate the deformity. Parental concern over the

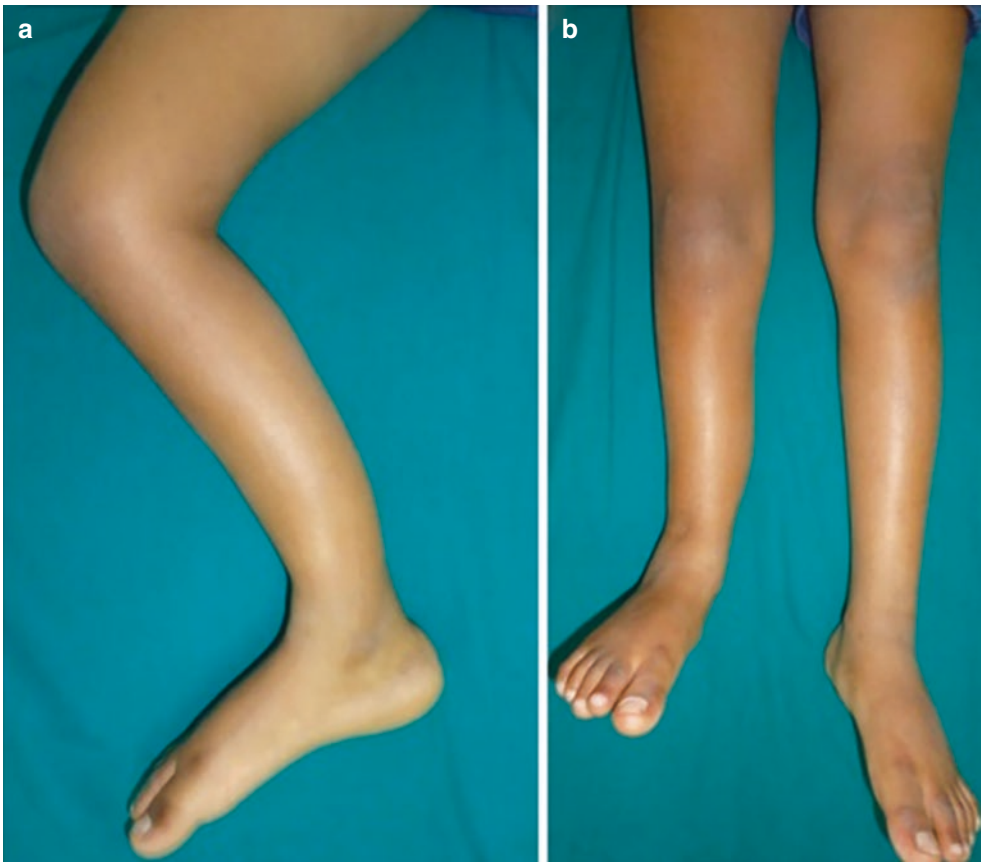

Fig. 1.7 (**a** and **b**) Posteromedial bowing. Nine-year old with right-sided posteromedial bowing and limb shortening. No significant calcaneus deformity noted

appearance of the lower extremities as the child begins to stand and walk is what typically prompts evaluation. Cases are often bilateral and are noted to improve with time. Spontaneous resolution is common by 18–24 months. Typically, alignment through childhood progresses to maximum valgus between ages 3 and 4 before gradual correction to adult alignment by the age of 7 years. While usually, this condition self-corrects, if varus alignment is persistent past 24 months or if significant clinical progression is noted, radiographs should be obtained to evaluate for underlying disorders [40].

Blount's Disease

Tibia vara, often referred to as Blount's disease, is characterized by abrupt varus deformity of the proximal tibia. In Blount's description in 1937, he described an irregular physeal line of the proximal medial tibia and a wedge-shaped epiphysis with medial metaphyseal beaking [41]. In contrast to physiologic genu varum, this is a progressive deformity. Two main forms exist and are classified based on time of onset—infantile (<4 years old) and adolescent (>10 years old).

While there may be a positive family history, infantile Blount's disease is considered a developmental disorder as radiographic features are typically not seen in children younger than 1 year (Fig. 1.8). Typically, children with Blount's disease are early walkers and are often obese. Finite element modeling has shown that compressive forces sufficient to retard physeal growth can be produced on the medial tibial plateau in a 2-year-old in the 90th percentile for body weight and with a 20-degree deformity [42]. Additionally, higher BMIs have been associated with greater overall varus alignment in children with infantile tibia vara [43].

Adolescent Blount's disease is a distinct form of tibia vara that presents later in childhood. The underlying etiology of adolescent Blount's disease is multifactorial and is not fully understood [44]. It is thought that progressive varus deformity results from repetitive trauma to the posteromedial proximal tibial physis due to increased body weight. While some have postulated that residual static varus alignment influences the development of adolescent Blount's disease, Davids et al. demonstrated that increased thigh girth and the resulting "fat-thigh gait" has been shown to lead to dynamic loading of the medial proximal tibial physis that can lead to compressive forces capable of inhibiting physeal growth [45]. The role of Vitamin D deficiency in the development of Blount's Disease continues to be investigated. Race has also been shown to be a factor, with adolescent Blount's disease being more common in African American populations, although it is seen in all races and ethnicities [44].

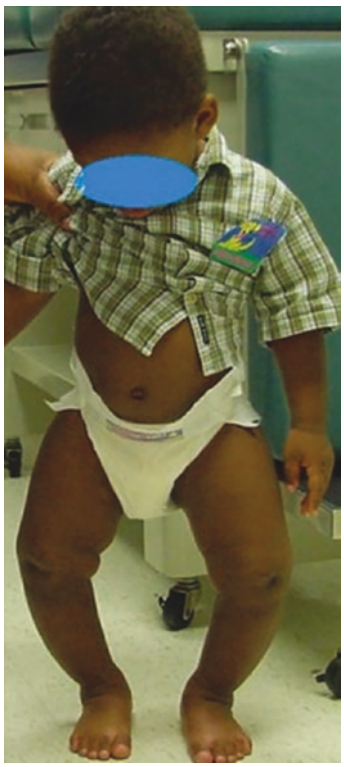

Fig. 1.8 Infantile blounts. Four-year-old male with Infantile Blount's disease. This is more common in African American populations

Proximal Focal Fibrocartilaginous Dysplasia

This is a rare idiopathic benign condition that can cause tibia vara. This typically presents as an abrupt varus deformity distal to the joint line of the knee and corresponds to cortical sclerosis at the metaphyseal–diaphyseal junction of the proximal tibia (at the insertion of the pes anserine). Abnormal fibrocartilage has been identified at the tendon insertion and the concept of this acting as a fibrous tether has been proposed. This deformity has the potential to correct spontaneously even with large varus deformities [46].

Genu Valgum

Physiologic valgus first becomes apparent around age 2 and reaches a maximum between ages 3 and 4. The valgus alignment of the lower extremity typically decreases over the next couple of years until adult alignment (5–7 degrees of valgus) is achieved at around age 7. Worsening genu valgum after age 7 is often pathologic. Common causes of genu valgum are shown in Table 1.5. In addition to underlying causes or injuries to the physis of the distal femur or proximal tibia, genu valgum can be idiopathic in nature. A recent study showed that 71% of children with idiopathic genu valgum were obese, which was significantly higher than in the normal population [47].

Table 1.5 Common causes of genu valgum

Unilateral presentation	Bilateral presentation
Post traumatic physeal injury	Physiologic
Proximal tibial metaphyseal fracture (Cozen's phenomenon)	Rickets
Osteochondroma	Idiopathic
Focal Fibrocartilaginous dysplasia	Pseudoachondroplasia
Fibrous Dysplasia	Morquio's (MPS IV)
Encondromatosis	Chondroectodermal dysplasia (Ellis van Creveld)
Fibular hemimelia	Multiple Hereditary exostosis
	Spondyloeiphyseal dysplasia

Acquired

Residual Hip Deformity

Outcomes of pediatric hip disorders (such as developmental hip dysplasia, SCFE, and Perthes Disease) are largely dependent on the congruency of the hip joint and avoidance of complications including avascular necrosis. It is important to consider the effect that these disorders, and their treatment, have on lower extremity limb length and alignment. Complete physeal arrest, either from the condition itself or as a result of treatment, can lead to limb length discrepancy. Children may present with an abductor lurch secondary to the corresponding greater trochanteric "overgrowth." Partial arrest can lead to the development of coxa vara or coxa valga. Additionally, studies evaluating coronal lower extremity alignment after Legg Calve Perthes Disease and SCFE have shown the development of valgus lower extremity alignment over time [48, 49].

Physeal Fractures

Injuries that are juxtaphyseal or ones that involve the physis can cause direct injury to the chondrocytes and interrupt normal growth of the extremity. This can lead to complete arrest with no further growth or partial arrest which can lead to angular deformity or slowing of growth. The risk of development of a physeal arrest is related to the Salter-Harris Classification, mechanism of injury, and degree of initial displacement. Reduction should be carefully attempted as repeated reductions may increase the risk of premature physeal closure [50]. Significant clinical deformity or limb length discrepancy can develop and is based on the age of the patient and the amount of growth remaining.

Physeal injuries of the distal femur are the most clinically relevant, given the magnitude of potential growth and the undulating nature of the physis. This undulation leaves the physis vulnerable to arrest as a fracture line can potentially cross multiple zones of the physis encouraging the potential of bony bar formation.

Salter-Harris Classification and initial displacement of the fracture correlate with complications [51]. The incidence of physeal arrest has been shown to be as high as 50% [51, 52]. Leg length discrepancies or angular deformity can develop and often necessitate additional intervention.

Femoral neck fractures occur typically as part of high-energy polytrauma. While avascular necrosis remains a significant and devastating complication of these fractures, premature physeal arrest can occur in up to 22% of cases [53]. This can be related to the injury at the time of fracture or secondary to implant placement. Limb length discrepancy tends to be minimal given the robust nature of the distal femoral physis. The angular deformity can occur from partial arrest resulting in coxa valga or coxa vara.

Distal tibia fractures have an overall rate of premature physeal closure of 13% with a reported range between 0.2% and 42%. Salter Harris IV fractures are most likely to develop arrest followed by Salter-Harris II fractures [54]. Mechanism of injury also plays a role as supination-external rotation injuries have a premature arrest rate of 35% in comparison to the 54% seen in children with pronation abduction-type injuries [55]. An important criterion for the development of physeal arrest in these injuries is displacement following reduction [53, 54]. Angular deformity is more common in pronation external rotation injuries [56].

Nonphyseal Fractures

Fractures of the lower extremities can result in angular deformity and limb length discrepancy even if the growth plate is not directly involved. Fracture malunion can result in long bone deformity. In younger children, this may remodel over time but as the child ages and approaches skeletal maturity, little remodeling will occur.

Femoral overgrowth is a well-documented phenomenon after femoral shaft fracture. This is related to increased vascular perfusion of the femoral physis during fracture healing. Typically, this measures about 1 cm and rarely exceeds 2 cm [57]. Femoral overgrowth has been reported, irrespective of the treatment methods. Given the anticipation of overgrowth, 1 cm of shortening is acceptable in young children when treating femoral shaft fractures with hip spica casting.

Progressive valgus angulation has been documented after a nondisplaced or minimally displaced fracture of the proximal tibia metaphysis. Known as Cozen's phenomenon, after healing of the fracture, a valgus deformity develops, which can be alarming in appearance (Fig. 1.9). There are many theories in regard to the mechanism of the phenomenon including soft tissue interposition, stimulation of the medial physis or tethering of the lateral physis by the intact fibula. The reported incidence of Cozen's Phenomenon varies between 50 and 90% with multiple studies noting the resolution of this deformity with time with few having clinically significant genu valgum requiring intervention [58, 59].

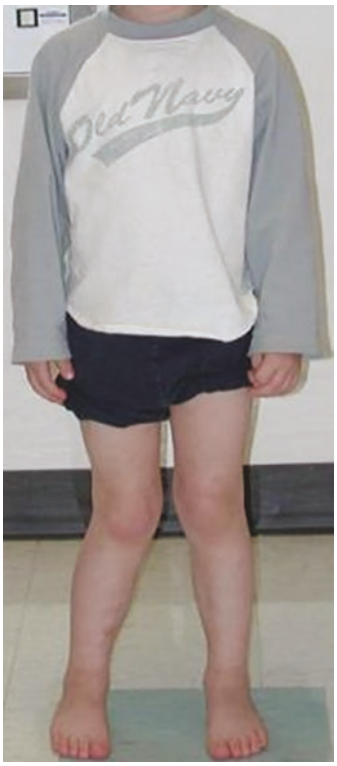

Fig. 1.9 Cozen's phenomenon after proximal tibia metaphyseal fracture. Six-year-old male, 1-year-post-proximal tibia metaphyseal fracture that developed genu valgum. He was observed serially and noted to completely remodel the valgus deformity

Post-Infectious

Both osteomyelitis and septic arthritis can lead to growth arrest or disturbance. Chondrocytes can be damaged by the infection itself or by the surgical debridement required to treat it, resulting in a bony bar formation. Even with the infection being successfully treated, the clinical effect may not be apparent for several years.

The infectious process can result in physeal cell death, which can produce similar tethering properties without osseous bar formation. Physeal bar equivalent has been reported early in life after infection near the distal femur physis [60]. Additionally, meningococcal septicemia can lead to a vascular occlusive process, resulting in damage to multiple growth plates and ultimately may present years later as limb length discrepancy or angular deformity [61].

Iatrogenic

Physeal damage leading to growth arrest can be from iatrogenic causes. Reduction of physeal fractures should be done carefully as repeated or overaggressive reduction attempts can lead to physeal damage and arrest. Additionally, manipulation after partial healing can also lead to physeal injury. Closed manipulation of a lower extremity contracture should be done carefully as physeal separations can occur with excessive force.

Surgical procedures near the physis also have the potential to cause iatrogenic injury to the physis, and careful dissection is important to avoid injury to the perichondral ring. Additionally, transphyseal hardware, improperly placed intramedullary nails, or instrumentation for guided growth can result in premature physeal growth arrest.

Ligament reconstruction around the knee often requires drilling tunnels that are near or across the distal femoral or proximal tibial physis. Both the cross-sectional area of the drilled tunnel and the angulation of the tunnel are thought to contribute to the development of both angular deformity and limb length discrepancy after anterior cruciate ligament (ACL) reconstruction with animal studies demonstrating that disruption of 7% of cross-sectional area leads to growth disturbance [62]. Growth abnormalities have been seen with both transphyseal and all epiphyseal ACL reconstruction. The most common deformities are genu valgum, recurvatum, and overgrowth. Overgrowth is seen in 62% of cases of limb length discrepancy after ACL reconstruction [63]. Growth disturbances have also been reported after Medial patellofemoral ligament (MPFL) reconstruction secondary to variability in tunnel placement [64].

Summary

The purpose of this chapter is to introduce the vast array of etiologies of lower limb deformities in the pediatric population. These deformities can be classified into underlying, congenital, developmental, or acquired conditions. The remaining chapters in this book will further discuss the diagnosis, classification, and management of specific disorders.

Commentary

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Limb deformities in children may result from innumerable causes, and the severity and functional impairment of a deformity can vary widely between patients. Drs. Zakrzewski and Jain have done a commendable job broadly categorizing these myriad causes into congenital, acquired, developmental, and those associated with underlying conditions. As they aptly say, "despite some overlap, categorizing them can help simplify diagnosis and treatment." Identifying the etiology of a limb deformity may uncover associated orthopedic and non-orthopedic problems. In appropriate patients, genetic counseling is an important consideration; a genetic counselor can help decide if and what testing may be germane and can provide useful information about future family planning [65, 66]. Testing may confirm or refute a diagnosis, guide treatment, and help access services and research trials. Advanced imaging such as MRI may provide useful information such as the presence of unossified cartilage and define neurovascular and soft tissue anatomy. The accumulated information from a thorough evaluation and work-up allows the family and clinical team to understand the natural history and prognosis of the deformity and develop a comprehensive treatment plan, and in some cases a "life plan."

Treatment options for pediatric limb deformities are almost as varied as the deformities themselves. They depend on patient, family, and surgeon/clinical team factors. These factors include, but are not limited to diagnosis, prognosis, comorbidities, cultural, religious, social, family economics, surgeon/clinical team experience, and medical care access. After a diagnosis is established, treatment goals need to be clearly defined and a personalized reconstructive plan created. Regardless of the particular cause, treatment goals may include any or several of the following: prevent deformity progression, decrease disability, decrease pain, improve function, correct deformity, stabilize joints, normalize limb alignment, equalize limb lengths, and prevent deformity recurrence.

The orthopedic surgeon must be part of the comprehensive treatment team. Prior, during, and after orthopedic interventions, associated conditions, comorbidities, and abnormalities may need to be treated or optimized by the appropriate specialist. Examples include metabolic (e.g., rickets), hematologic (e.g., TAR syndrome), and structural (e.g., Arnold-Chiari) abnormalities. The surgical treatment plan must include pre-operative, intra-operative, and post-operative considerations for associated problems. Examples include screening for spinal stenosis, taking precautions for malignant hyperthermia and latex allergies, and monitoring platelet counts. Understanding potential problems, obstacles, and complications for a particular deformity allow the surgeon and treatment team to optimize their reconstructive plan and minimize sub-optimal outcomes [67].

The reconstructive plan for limb deformity may include operative and/or non-operative options. Non-operative treatment for limb length discrepancies and deformities includes the use of shoe lifts (for lower extremity), orthoses, and prostheses depending on the etiology, amount, and underlying shape and function of the extremity [68]. Surgical options depend on the defined goals of treatment and include combinations

of acute and gradual correction with internal and external fixation in single or multiple stages. The reconstructive plan may evolve over time based on outcomes, response to previous treatments, changes in goals, and advances in medical and surgical treatments.

Limb deformities in pediatric patients may progress with time and future growth. Complete and partial growth arrest results in axial and angular deformities. Progressive limb length discrepancy, for most conditions, can be accurately predicted which helps guide the reconstructive plan [69, 70]. Predicted discrepancies above a certain threshold may alter a surgeon's treatment plan (e.g., prosthetic fitting–with or without amputation–in lieu of multiple staged lengthenings, or combining contralateral shortening by growth arrest with ipsilateral lengthening. Patients should be followed through skeletal maturity with discrepancy calculations repeated throughout growth. If the predicted discrepancy or reconstructive complexity exceeds the surgeon's capabilities, experience, or comfort level, they have the opportunity to refer.

Treatment options for pediatric limb deformities continually evolve and mature. Advances in orthotics, alongside prosthetics design and manufacturing, have improved their use and functionality. Surgical advances have come from modifications of existing procedures, advent of new reconstructive procedures, introduction of new hardware and software, and repurposing existing hardware in innovative ways. Examples of these advances include percutaneous osteotomy and fixation, cross-union technique for congenital pseudarthrosis of the tibia [71], femoral shortening with posterior knee soft-tissue lengthening and capsulotomy for arthrogryposis [72, 73], computer-dependent hexapod fixators that can accommodate more complex deformities, and extra-medullary placement of lengthening rods in younger and smaller patients [74, 75].

Generalizations for limb deformity surgeons:

- Understand the pathoanatomy
- Formulate an individualized reconstructive plan
- Optimize patients prior to surgery
- Plan to obtain and then maintain deformity correction
- Consider how a deformity may change over time
- Anticipate and prevent complications but be prepared to treat them
- Remember that limb function is more important than limb length

References

1. Loder RT, Wittenberg B, DeSilva G. Slipped capital femoral epiphysis associated with endocrine disorders. J Pediatr Orthop. 1995;15(3):349–56.
2. Bitzan M, Goodyer PR. Hypophosphatemic rickets. Pediatr Clin N Am. 2019;66(1):179–207.
3. Misra M, Pacaud D, Petryk A, et al. Vitamin D deficiency in children and its management: review of current knowledge and recommendations. Pediatrics. 2008;122(2):398–417.
4. Barrett IR, Papadimitriou DG. Skeletal disorders in children with renal failure. J Pediatr Orthop. 1996;16(2):264–72.
5. Dwan K, Phillipi CA, Steiner RD, et al. Bisphosphonate therapy for osteogenesis imperfecta. Cochrane Database Syst Rev. 2016;10(10):Cd005088.
6. Crawford AH, Schorry EK. Neurofibromatosis update. J Pediatr Orthop. 2006;26(3):413–23.
7. Jani-Acsadi A, Ounpuu S, Pierz K, et al. Pediatric Charcot-Marie-Tooth disease. Pediatr Clin N Am. 2015;62(3):767–86.
8. van Bosse HJP, Pontén E, Wada A, et al. Treatment of the lower extremity contracture/deformities. J Pediatr Orthop. 2017;37(Suppl 1):S16–s23.
9. Dobbs MB, Boehm S, Grange DK, et al. Case report: congenital knee dislocation in a patient with larsen syndrome and a novel filamin B mutation. Clin Orthop Relat Res. 2008;466(6):1503–9.
10. Larsen LJ, Schottstaedt ER, Bost FC. Multiple congenital dislocations associated with characteristic facial abnormality. J Pediatr. 1950;37(4):574–81.
11. Carrera-García L, Natera-de Benito D, Dieterich K, et al. CHRNG-related nonlethal multiple pterygium syndrome: muscle imaging pattern and clinical, histopathological, and molecular genetic findings. Am J Med Genet A. 2019;179(6):915–26.
12. Baitner AC, Maurer SG, Gruen MB, et al. The genetic basis of the osteochondrodysplasias. J Pediatr Orthop. 2000;20(5):594–605.
13. White KK, Sousa T. Mucopolysaccharide disorders in orthopaedic surgery. J Am Acad Orthop Surg. 2013;21(1):12–22.
14. Goldwein JW. Effects of radiation therapy on skeletal growth in childhood. Clin Orthop Relat Res. 1991;262:101–7.
15. Reif TJ, Matthias J, Fragomen AT, et al. Limb length discrepancy and angular deformity due to benign bone tumors and tumor-like lesions. J Am Acad Orthop Surg Glob Res Rev. 2021;5(3):e00214.
16. Bovid KM, Moore MD. Juvenile idiopathic arthritis for the pediatric orthopedic surgeon. Orthop Clin North Am. 2019;50(4):471–88.
17. Rodriguez-Merchan EC. Prevention of the musculoskeletal complications of hemophilia. Adv Prev Med. 2012;2012:201271.
18. Dempsey-Robertson M, Wilkes D, Stall A, et al. Incidence of abdominal tumors in syndromic and idiopathic hemihypertrophy/isolated hemihyperplasia. J Pediatr Orthop. 2012;32(3):322–6.
19. Brioude F, Kalish JM, Mussa A, et al. Expert consensus document: clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. Nat Rev Endocrinol. 2018;14(4):229–49.
20. Ballock RT, Wiesner GL, Myers MT, et al. Hemihypertrophy. Concepts and controversies. J Bone Joint Surg Am. 1997;79(11):1731–8.
21. Burn J, Winter RM, Baraitser M, et al. The femoral hypoplasia-unusual facies syndrome. J Med Genet. 1984;21(5):331–40.
22. Crawford DA, Tompkins BJ, Baird GO, et al. The long-term function of the knee in patients with fibular hemimelia and anterior cruciate ligament deficiency. J Bone Joint Surg Br. 2012;94(3):328–33.
23. Graham JM Jr. Limb anomalies as a consequence of spatially-restricting uterine environments. Prog Clin Biol Res. 1983;110(Pt A):413–22.