Paediatric Orthopaedic Diagnosis

Asking the Right Questions

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Benjamin Joseph
Aster Medcity
Kochi
Kerala
India

James Robb
Orthopaedic
University of St Andrews
Scotland
UK

Randall T. Loder
James Whitcomb Hospital
Indianapolis, IN
USA

Ian Torode
Victorian Paediatric Orthopaedic Centre
Melbourne
Australia

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In gratitude to our wives, Susan, Christine, Frith, and Lyn, who forfeited many hours while we worked to complete this book.

Benjamin Joseph, Randall Loder,
James Robb, Ian Torode

Foreword

This new work by four very experienced paediatric orthopaedic surgeons utilizes a novel approach designed to enable the clinician to make an accurate diagnosis of quite a variety of musculoskeletal conditions which affect children. While the presentation is new, the conceptual basis goes back to the beginning of organized medicine. The authors begin each chapter with a series of questions which the practitioner should ask the patient or parent. This, of course, represents the taking of a history. How often have each of us had to relearn the dictum that the most important diagnostic study of all is a careful history. I believe that it was William Osler who said, "Listen to your patient and he will tell you the diagnosis."

The next step in the search for a diagnosis is the physical examination. The authors put this in modern parlance with categories, "look, feel, and move." Within each category both common and unusual findings are considered. Thus, the reader is encouraged to avoid quickly jumping to studies and tests before performing a thoughtful and thorough physical examination of the patient.

Next the authors move through appropriate studies, radiographs, laboratory exams, and the like. These considered, the physician is now ready to construct a differential diagnosis. Especially helpful are the charts which are constructed to further organize the previously presented information. Finally appropriate references are cited to allow the clinician to broaden his or her knowledge.

The chosen topics encompass most of the common presentations to a clinician both in the general setting and a specialty setting. Students and trainees will find the book especially helpful as they seek to acquire diagnostic skills.

The authors have deliberately avoided describing treatment of the various conditions. This will be welcomed by practitioners in the primary care arena. Their need is one of recognition, understanding of the appropriate approach to making a diagnosis. Other resources are available when needed to guide treatment.

In summary this book will be a valuable teaching aid in a variety of clinical settings. Most common conditions are covered by this authoritative and easily understood presentation, and many emerging practitioners will keep it handy.

TX, USA — Tony Herring, MD

Foreword

Paediatric orthopaedics is at crossroads. It is perhaps the last general orthopaedic specialty that covers a spectrum of well-recognized and more subtle musculoskeletal disorders that may or may not be difficult to diagnose. Moreover, there is a variation in age-specific conditions. This book addresses the question of the strategy for the approach to diagnosis in the various age groups according to the presenting symptom or disorder. Most common disorders, as they present, are addressed and then dissected in question form to provide a route to the correct diagnosis. This is the overwhelming success of the book. There is an old saying that "every patient is trying to tell the doctor something if only they would listen." Modern-day paediatric orthopaedic diagnosis is often in the hands of paramedical or nursing staff, and frequently the primary disorder presents in the emergency department rather than to primary or secondary care. This book provides a ready handbook to structured investigation and clinical assessment which is now frequently overlooked, in current practice, in preference for a generic nonspecific imaging protocol. It will be invaluable as a tool, not only for frontline staff but also as a prompt for clinical teaching in the clinic and a reference for residents in training. The major contribution, however, is in the overall clinical safety of its approach to diagnosis and its avoidance of inappropriate overinvestigation in the various age groups, with common or rarer disorders.

Southampton, UK Prof. N.M.P. Clarke, ChM, DM, FRCS, FRCS Ed

Preface

The clinician's role in reaching a diagnosis is, in many ways, similar to a detective tracking down the culprit in a crime. Both the clinician and the detective ask questions. The more pertinent the questions become, the closer the disease or culprit is to being identified.

When confronted with a child with a limp, the first question that crops up is, "*is it a painful or painless limp?*" The answer to this question clearly narrows down the possibilities. If the child has no pain, the second question could be "*when did the child start limping*?" If the child has been limping right from the time he or she started walking, the possible diagnoses come down to a handful of conditions that can easily be differentiated by clinical examination and one or two simple investigations. Thus, the emphasis of this book is to assist the reader to formulate rational questions to facilitate the diagnosis of the cause of orthopaedic problems occurring in children of different ages.

The book has been divided into sections based on the usual age at presentation: the newborn, the infant, the toddler and the preschool child, the child in the school-going age, and the adolescent. It is possible that a child with a congenital anomaly may present for the first time in the school-going age; in which case, the reader would have to look under the newborn section to get the necessary information.

An attempt has been made to include all the common conditions that are encountered in clinical practice; some of the rarer conditions have also been included in several chapters to make this book wider in its scope. However, it is not intended to present a comprehensive, exhaustive discussion of each topic.

It is hoped that this book will be of benefit to fellows and junior consultants in children's orthopaedics, neonatology, and paediatrics in their respective busy practices.

Kochi, India — Benjamin Joseph
Indianapolis, IN, USA — Randall T. Loder
Edinburgh, UK — James Robb
Melbourne, Australia — Ian Torode

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Part I

The Newborn

The Deformed Hip

1

Randall T. Loder

1.1 Introduction

The spectrum of malformations of the hip in the newborn is wide and ranges from a minor degree of acetabular dysplasia to major deficiency like proximal focal femoral deficiency. Developmental dysplasia of the hip (DDH) is the commonest of hip anomalies; early diagnosis and treatment often results in a satisfactory outcome, while a delay may make treatment more complicated and the outcome less predictable. Some abnormalities of the hip may not be evident on cursory examination since the hip is a deep-seated joint unlike the knee where minor deformities are easily recognized.

The important questions that need to be answered while trying to make a diagnosis of the nature and cause of congenital anomalies of the hip in the newborn are listed below.

1.2 Questions to Establish a Diagnosis

- **Was the baby premature or full term?**
- **Is the baby the first born?**
- **Is there a history of breech presentation?**
- **Is there a family history of hip dysplasia?**
- **Is there a history of reduced fetal movements during pregnancy?**
- **Is there a history of maternal illness or substance abuse?**
- **Are the joints of the limbs hypermobile or are they stiff?**
- **Are there other musculoskeletal deformities?**

Was the baby premature or full term?

Hip dysplasia is more frequent in full-term infants and it is infrequent in premature babies (Loder and Skopelja 2011).

Is the baby the first born?

Developmental dysplasia of the hip is more common in first-born babies.

Is there a history of breech presentation?

There is a strong association between developmental dysplasia of the hip and breech presentation.

Is there a family history of hip dysplasia?

A positive family history may be present in developmental dysplasia of the hip.

Is there a history of reduced fetal movements during pregnancy?

Reduced fetal movement during pregnancy (akinesia) is often reported in arthrogryposis.

Is there a history of maternal illness or substance abuse?

Ingestion of drugs or maternal illness early in pregnancy can predispose the infant to significant congenital malformations and syndromes.

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Are the joints of the limbs hypermobile or are they stiff?

Stiff joints are encountered in arthrogryposis while hypermobile joints are seen in Larsen syndrome; in both these conditions, the hip may be dislocated.

Are there other musculoskeletal deformities?

Metatarsus adductus and congenital muscular torticollis are often associated with hip dysplasia. Symmetrical joint contractures and dislocations are often associated with arthrogryposis, Larsen syndrome, and myelomeningocele. Infantile scoliosis is often associated with the windswept hip deformity (Green and Griffin 1982).

Is there pseudoparalysis or pain with hip examination?

Neonatal proximal femoral physeal fractures can occur in difficult deliveries.

1.3 Physical Examination

1.3.1 Look

Observe if the normal groin crease is present or lost. Note if the gluteal creases are symmetric. Look for deformities of the hip in the sagittal, coronal, and transverse planes. Observe if one limb is short with shortening of the femoral segment (Galeazzi sign). Note if the leg is also short. Look for spontaneous movement of the lower limbs and note if movements are reduced or absent.

1.3.2 Feel

Palpate the femoral triangle and see if the normal resistance offered by the femoral head is present. Palpate the femoral pulse and note if the vascular sign of Narath is positive. Palpate the trochanter and see if the normal relationship to the anterior superior iliac spine is maintained.

1.3.3 Move

Abduct both hips with the hips and knees held in 90° of flexion, and observe if there is any limitation of abduction (normally in the new born, the thighs may almost touch the couch). Check the ranges of internal and external rotation of the hips.

1.3.4 Special Tests

Tests for Neonatal Hip Instability: The Barlow and Ortolani Tests

The Ortolani test indicates a reduction of a dislocated hip. The Barlow test indicates a dislocation of a reduced hip. These tests should be performed with the baby relaxed and warm and without a diaper (Fig. 1.1).

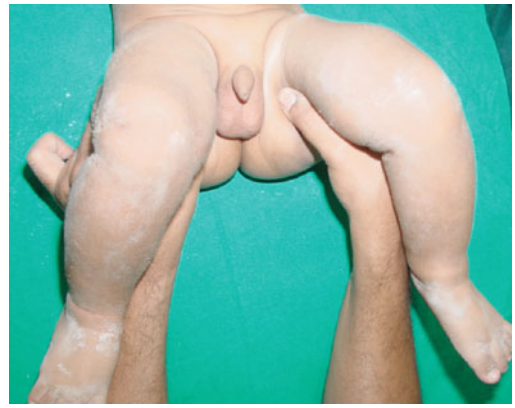
The Ortolani test is performed with the hip flexed and adducted, and then with the examiner's fingers gently resting on the greater trochanter and slight traction, the hip is gently abducted. Specifically for the left hip, the right hand gently holds the left thigh with the middle/ring finger resting on the greater trochanter and the thumb on the medial thigh. The left hand stabilizes the opposite hip. Then with gentle abduction and slight traction, the examiner will feel a clunk as the femoral head reduces into the acetabulum. The opposite positioning of the hands is used on the right hip. Each hip is tested individually.

The Barlow test is performed with the hip flexed and abducted; similar positioning of the hand is used as in the Ortolani test. Then the thigh is gently moved into an adducted position with gentle posterior pressure. The examiner will feel the femoral head exit the posterior aspect of the acetabulum (Barlow 1963).

1.4 Investigations to Confirm the Diagnosis

1.4.1 Radiography

Plain radiographs of the pelvis are not indicated in the newborn; the hip is highly cartilaginous and thus difficult to interpret. Between birth and age 4 months, ultrasound is the imaging method



Thigh of the infant is held with the thumb on the medial aspect of the thigh and the index and middle fingers on the trochanter

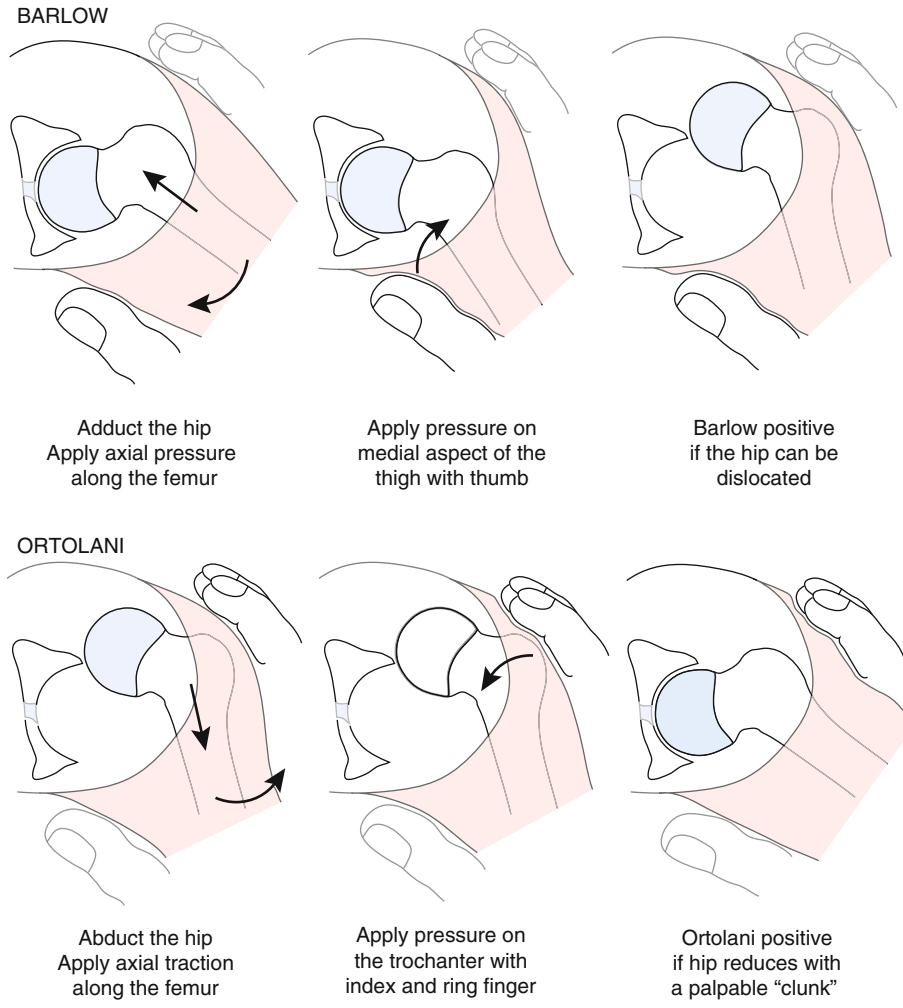


Fig. 1.1 Diagram of the technique of performing the Ortolani and Barlow tests

of choice. After 4 months of age, plain radiographs are used. DDH can range from simple acetabular dysplasia to frank high-riding dislocation. Tönnis describes 4 classes of hip dysplasia. Grade 0 is a normal hip; grade 1 is where the ossification center of the capital femoral epiphysis is medial to Perkin's line; grade 2 the ossification center is lateral to Perkin's line but below the superolateral margin of the acetabulum; grade 3 the ossification center is at the level of the superolateral margin of the acetabulum; and grade 4 the ossification center is above the superolateral margin of the acetabulum (Tönnis et al. 1987).

1.4.2 Ultrasound

Ultrasound of the hip is very useful to assist in the diagnosis of hip dysplasia as well as monitoring treatment. There are two methods (Weintroub and Grill 2000) commonly used: the static method of Graf (1984) and the dynamic method of Harcke (1995, 2005).

The method of Graf assesses the morphology of the hip using angular measurement (α and β angles) and the percentage of femoral head coverage (Fig. 1.2). The dynamic method of Harcke assesses the stability of the hip with the Ortolani and Barlow maneuvers.

1.5 Differential Diagnosis

1.5.1 Developmental Dysplasia of the Hip

Neonatal Hip Instability
This is diagnosed by a positive Barlow sign or feeling some subluxation without complete dislocation. A complete dislocation may or may not be reducible with the Ortolani maneuver. Dynamic and static ultrasound will confirm the diagnosis.

Acetabular Dysplasia with or Without Instability
This is simply a shallow acetabulum. If there is no instability, no abnormality will be felt upon the Barlow maneuver. It can be diagnosed only with either ultrasound (where the α angle will be less than normal) or by a plain radiograph (where the acetabular index will be higher than normal) (Tönnis 1976).

1.5.2 Teratogenic Hip Dislocation

Hip Dislocation in Arthrogryposis
The hip is frequently involved in children with arthrogryposis (Bernstein 2002; Bevan et al. 2007) and can range from a mild contracture to complete dislocation. Physical examination often demonstrates a flexion and adduction contracture of the hip. When there is a dislocation, it is usually very rigid and high riding such that the Ortolani sign is negative; ultrasound will clearly demonstrate the dislocation in the newborn; plain radiography will also demonstrate the dislocation (Fig. 1.3).

Hip Dislocation in Spina Bifida
Children with spina bifida often either are born with a hip dislocation or develop it later in life (Broughton et al. 1993). They are often associated with flexion and adduction contractures. Those born with a hip dislocation have the teratologic type. In children with higher levels of spina bifida, the hip adductors and flexors are strong while the hip abductors and extensors are weak (e.g. L3 and L4); this can lead to a dislocation (Fig. 1.4).

Hip Dislocation in Skeletal Dysplasias and Generalized Syndromes
One form of skeletal dysplasia that is associated with congenital hip and knee dislocation is spondyloepiphyseal dysplasia with congenital joint

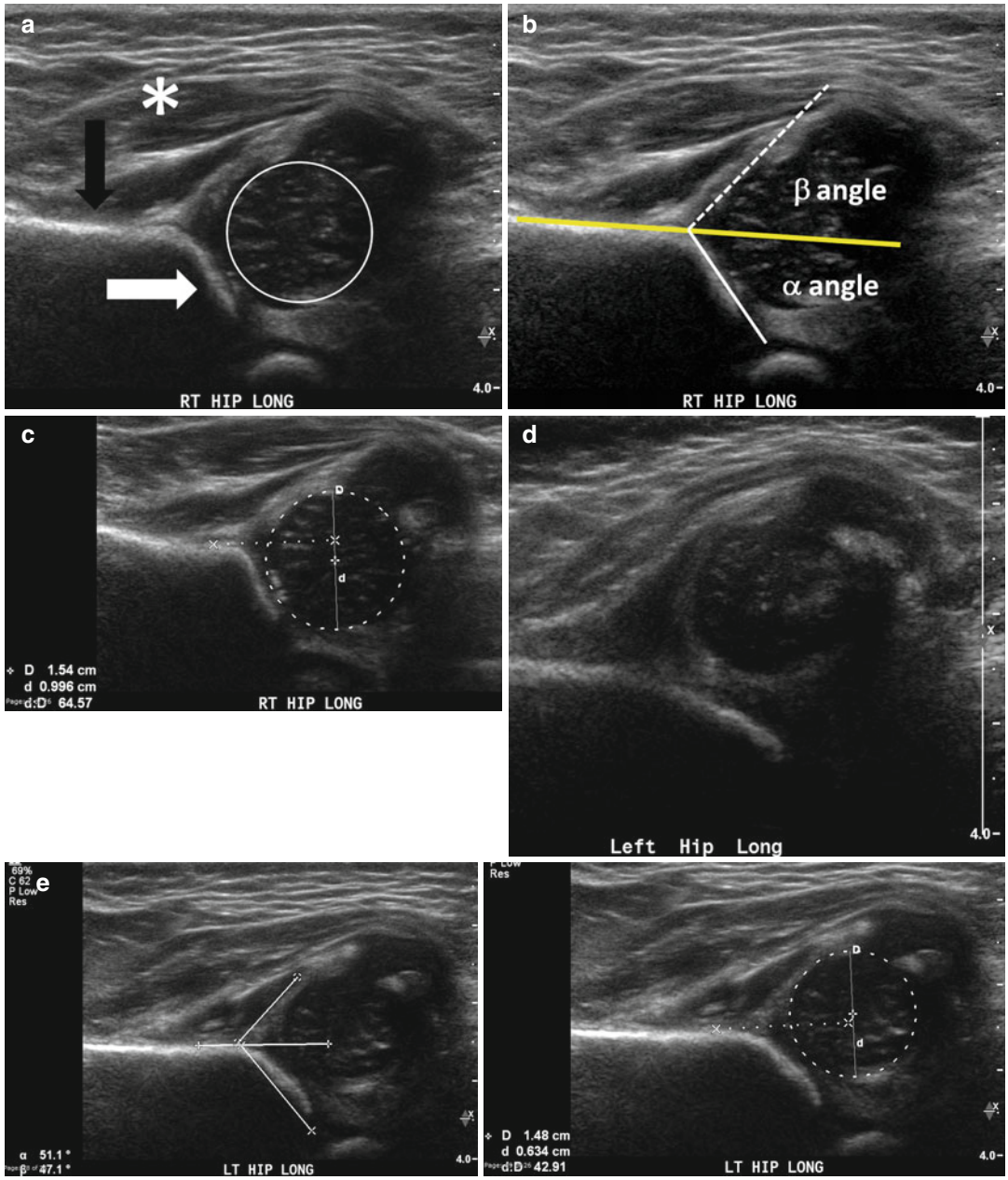


Fig. 1.2 Hip ultrasounds in DDH. (**a**) A representative longitudinal ultrasound image of a normal neonatal hip. The ilium is marked by the *solid black arrow*, the bony acetabular roof by the *white arrow*, the abductor muscles by the *white asterisk*, and the femoral head by the *white circle*. (**b**) Measurement of the alpha (α) and beta (β) angles on ultrasound establish the Graf class. The baseline is first drawn and is the line along the ilium as it intersects the bony and cartilaginous portions of the acetabulum (line 1, *solid yellow line*). Line 2 (*solid white line*) is from the acetabular edge to the triradiate cartilage; line 3 (*dashed white line*) is from the acetabular edge to the lateral cartilaginous roof. The α angle is the angle between the baseline line 2; the β angle is the angle between the baseline line 3. A normal α angle is > 60°. (**c**) The percentage of femoral coverage is calculated as (d/D) × 100. D is the entire diameter of the femoral head and d is the portion covered by the bony acetabular roof. In this example, the percentage of femoral coverage is 65%. (**d**) A complete hip dislocation; note there is no contact between the femoral head and the acetabulum. (**e**) Mild acetabular dysplasia in an infant born breech. The α Graf angle is 51 (**a**) degrees and the femoral head coverage is 42.9 % (**b**)

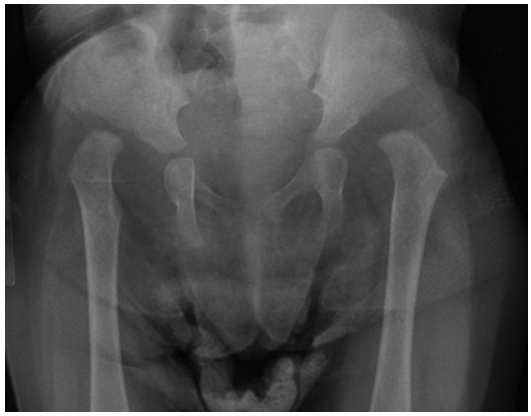


Fig. 1.3 Bilateral teratogenic hip dislocations in an infant with arthrogryposis

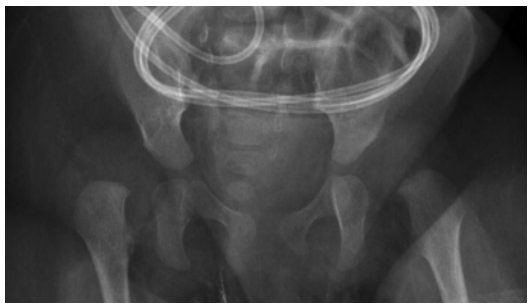


Fig. 1.4 Complete dislocation of the right hip and marked subluxation of the left hip in an infant with high lumbar myelomeningocele

dislocation (Hermanns et al. 2008). In addition to the dislocations, short stature and progressive kyphosis are characteristic features of the condition. The dysplasia is caused by a CHST3 mutation. Larsen syndrome (Larsen et al. 1950) is a constellation of multiple joint dislocations (hip, elbow, knee), severe foot deformities (equinovarus or equinovalgus), and dysmorphic facies. It can often be diagnosed prenatally with ultrasound (Rochelson et al. 1993; Tongsong et al. 2000). Congenital dislocation of the knee commonly occurs in association with Larsen syndrome (Houston et al. 1981; Laville et al. 1994; Rønningen and Bjerkreim 1978; Steel and Kohl 1972).

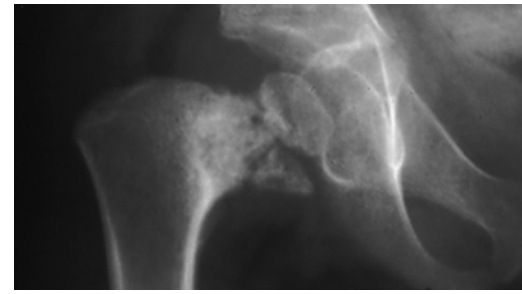


Fig. 1.5 The radiograph of a child with congenital coxa vara. Note the inferior metaphyseal triangular fragment

1.5.3 Congenital Coxa Vara

Congenital coxa vara is defined as a neck-shaft angle of the proximal femur less than normal. It is also associated with a classic inferior metaphyseal triangular fragment (Beals 1998; Weinstein et al. 1984). It is frequently unilateral and often associated with a short femur. Physical examination will not demonstrate any instability, but there will be decreased internal rotation of the hip, and in the severe case with a significantly short femur, there will be a positive Galeazzi sign. It is often not diagnosed until the infant begins to walk, at which point a Trendelenburg limp is noted. Radiographs demonstrate the reduced neck-shaft angle, a more vertical proximal femoral physis, inferior displacement of the epiphysis, and the triangular inferior metaphyseal fragment (Fig. 1.5).

1.5.4 Proximal Focal Femoral Deficiency

Proximal femoral focal deficiency (PFFD) is part of the wide spectrum of congenital femoral deformity which can range from congenital coxa vara and congenital short femur to almost complete absence of the femur (Gillespie 1998; Lange et al. 1978; Panting and Williams 1978; Pappas 1983). There are numerous different

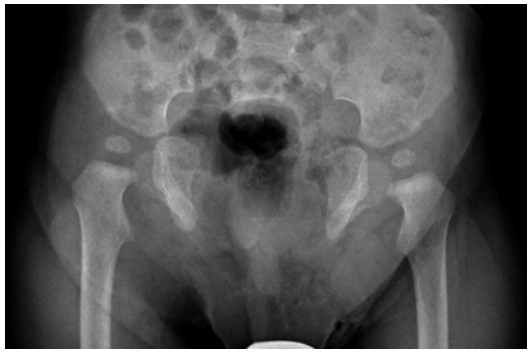


Fig. 1.6 The pelvis radiograph of a child with bladder exstrophy. Note the symphyseal widening

classification systems. Clinically the thigh is shorter than the opposite side (unless bilateral) and with the hip and knee in flexion. There is often associated external rotation of the thigh. Ortolani and Barlow tests are negative. Plain radiographs will confirm the diagnosis.

1.5.5 Rare Conditions

The Hip in Exstrophy of the Bladder
Due to widening of the symphysis in children with bladder/cloacal exstrophy, the hips are externally rotated and acetabular retroversion (Fig. 1.6). This will lead to an increased external rotation gait when the child becomes of walking age (Sponseller et al. 1995).

Hip Dysplasia Associated with Abduction Contracture of the Contralateral Hip
Hip dysplasia can be seen in children with an abduction contracture. The dysplastic hip is the one opposite the side of the contracture. Physical examination will demonstrate asymmetric gluteal folds and an apparent leg length discrepancy (Green and Griffin 1982).

Neonatal Proximal Femoral Physeal Fracture
A new born can sustain such a fracture during birth; the limb will be held in a position of external rotation and pseudoparalysis will be present (Lindseth et al. 1971; Ogden et al. 1984; Theodorou et al. 1982). Radiographs will demonstrate proximal and lateral migration of the proximal femoral metaphysis, mimicking a dislocation. Ultrasonography will demonstrate the presence of the proximal femoral epiphysis in the acetabulum.

Septic Conditions
Septic arthritis of the hip in the infant with or without concomitant osteomyelitis and iliopsoas abscess will result in a position of flexion and often abduction of the involved hip. The infant will be very irritable but may or may not be febrile. The hip will demonstrate marked irritability with passive motion, and a pseudoparalysis is often present (Frank et al. 2005; Karmazyn et al. 2007; Robben 2004; Samora and Klingele 2013). Appropriate imaging using ultrasound and MRI will point to the diagnosis.

1.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Table 1.1.

Table 1.1 Establishing the diagnosis of the cause of a malformed hip in the newborn

History						
First-born girl child Delivered at full term (these features may not be always present)	Sex, order of birth not relevant	Sex, order of birth not relevant	Sex, order of birth not relevant	Sex, order of birth not relevant	Sex, order of birth not relevant	Sex, order of birth not relevant
Positive family history may be present	Positive family history not present	Positive family history usually not present	Positive family history may be present	Positive family history often present	Positive family history seldom present	Positive family history seldom present
History of breech delivery may be present	–	–	–	–	–	–
Normal fetal movement	Reduced fetal movement in utero (akinesia) often present	Reduced fetal movement in utero may be present	Normal fetal movement	Normal fetal movement	Normal fetal movement	Normal fetal movement
–	–	–	–	–	–	History of maternal illness or substance abuse may be present occasionally
Physical examination						
Barlow and/or Ortolani test positive	Barlow and Ortolani tests will usually be negative	Barlow and/or Ortolani test may be positive	Barlow and Ortolani test positive	Barlow and Ortolani test will usually be negative	Barlow and Ortolani test negative	Barlow/Ortolani test negative
Limitation of abduction of the hip may be present	Limitation of abduction of the hip will be present	Limitation of abduction may be present	Excessive range of hip motion in all planes	Limitation of abduction may be present	Limitation of abduction and exaggeration of adduction present	Abduction may be normal or reduced
Generalized ligament laxity may be present	Ligament laxity will not be present	Ligament laxity not present	Marked ligament laxity present	Ligament laxity not present	No ligament laxity	No ligament laxity
Spontaneous movement of the lower limbs will be normal indicating that there is no paralysis	Spontaneous movement of the lower limbs will be reduced	Spontaneous movement of the lower limbs may be reduced (hip flexion may be retained)	Spontaneous movement of the lower limbs normal	Spontaneous movement of the lower limbs normal	Spontaneous movements of the lower limbs normal	Spontaneous movements of the lower limbs present

Passive internal rotation of the hip may be greater than external rotation suggesting that the femur is anteverted	Passive rotations of the hip will be reduced	Passive rotations of the hips may be normal	Passive rotations of the hip exaggerated	Passive rotations of the hip may be reduced	Passive external rotation of the hip exaggerated and internal rotation reduced	Rotations of the hip may be reduced
No deformities of knee	Flexion or hyperextension deformity of the knee may be present	Flexion deformity of the knee may be present or there may be no deformity at the knee	Hyperextension deformity of the knee may be present	Hyperextension deformity of the knee may be present	No knee deformity	No knee deformity
Metatarsus adductus may be present	Equinovarus or convex pes valgus deformities may be present	Equinus/equinovarus/ convex pes valgus may be present	Equinovarus deformity may be present	Equinovarus deformity may occasionally be present	No foot deformity	Foot deformity may or may not be present
Knee will be stable	Knee may be dislocated but instability will not be demonstrable	Knee will be stable	Knee often unstable	Knee may be unstable	Knee stable	Knee deformity may be present
				Short stature and altered body proportions may be evident		Very short thigh segment
Working diagnosis: Developmental dysplasia of the hip (DDH)	Working diagnosis: Teratologic hip dislocation in arthrogryposis	Working diagnosis: Paralytic hip dislocation in spina bifida	Working diagnosis: Hip dislocation in Larsen syndrome	Working diagnosis: Teratologic hip dislocation in skeletal dysplasia	Working diagnosis: Coxa vara	Working diagnosis: Proximal focal femoral deficiency
Investigations						
Ultrasound: Acetabular dysplasia Reduced alpha angle and increased beta angle	Plain radiograph: Hip dislocation	Plain radiograph: Spina bifida Hip dislocation	Plain radiograph: Hip dislocation Abnormal ossification of calcaneum and distal humerus	Plain radiograph: Hip dislocation Epiphyseal dysplasia Platyspondyly	Plain radiograph: Reduced neck-shaft angle	Plain radiograph: Proximal focal femoral deficiency
Diagnosis						
DDH	Hip dislocation in arthrogryposis	Paralytic hip dislocation in spina bifida	Hip dislocation in Larsen syndrome	Hip dislocation in spondyloepiphyseal dysplasia	Congenital coxa vara	PFFD

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The Deformed Femur

2

Randall T. Loder

2.1 Introduction

Malformation of the femur may be an isolated anomaly, or it may be associated with abnormalities involving the hip, knee, or the rest of the skeleton. Congenital anomalies of the femur include shortening, failure of development of the proximal or distal ends of the femur, bowing, or duplication; some of these may occur together.

2.2 Questions to Establish a Diagnosis

- **Is there a history of a viral illness or substance abuse in the mother during pregnancy?**
- **Is there a family history of similar deformities?**
- **Is the deformity unilateral or bilateral?**
- **Is the femur fully formed or is there a failure of formation of either the proximal or distal part of the femur?**
- **Is there a deficiency of the bones of the leg?**
- **Are other limbs also deformed, and if so, are the deformities symmetrical?**

Is there a history of a viral illness or substance abuse in the mother during pregnancy?

Ingestion of drugs or other exposures early in the pregnancy can predispose the infant to significant congenital malformations.

Is there a family history of similar deformities?

This is important when assessing for a skeletal dysplasia, although a new mutation may also be involved without any family history.

Is the deformity unilateral or bilateral?

Bilateral femoral deformities are more likely in a skeletal dysplasia (e.g., campomelic dwarfism). Femoral deficiency syndromes can be either unilateral or bilateral.

Is the femur fully formed or is there a failure of formation of either the proximal or distal part of the femur?

Failure of formation of the proximal femur (proximal focal femoral deficiency or PFFD) and failure of formation of the distal end of the femur (distal focal femoral deficiency or DFFD) are associated with severe shortening of the limb. In PFFD the hip is unstable, while in DFFD the knee is unstable.

Is there a deficiency of the bones of the leg?

Aplasia of a bone of the leg may be seen in association with PFFD and also with a bifid femur.

Are other limbs also deformed, and if so, are the deformities symmetrical?

Bowing of the femur may be associated with bowing of other long bones in campomelic dysplasia and osteogenesis imperfecta. Bowing in campomelic dysplasia is usually mild and symmetric, while in osteogenesis the bowing may be severe and asymmetric.

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2.3 Physical Examination

2.3.1 Look

Note what the visible deformity is; note if there is shortening or bowing. If the thigh appears bowed, note the direction and site of bowing. Observe if there is a skin dimple over the site of bowing. Note if the deformities are bilateral and symmetric. Observe if there are deformities in the legs and upper limbs. Look for facial dysmorphism.

2.3.2 Feel

Palpate the femur through its entire length; specifically note if the femoral head and the femoral condyles are palpable. Note the site and extent of bowing if present. Confirm if there is any bony outgrowth on the femoral shaft (duplication). Note if there is any tenderness over the femur.

2.3.3 Move

Perform the Barlow and Ortolani maneuvers to check for hip instability. Check for instability of the knee. Note if there is pain on moving the hip and knee. Check the passive range of hip, knee, and ankle motion.

2.4 Investigations to Confirm the Diagnosis

Plain Radiography
Observe for bowing deformities (e.g., osteogenesis imperfecta, campomelic dwarfism) and pseudarthrosis (proximal femoral deficiency). If there is a concern for a skeletal dysplasia, a skeletal survey must be done (see Chap. 23).

Ultrasound
In the very young child, this can define if there is a cartilaginous femoral head or a pseudarthrosis from a proximal femoral focal deficiency (Grissom and Harcke 1994; Kayser et al. 2005).

Genetic Evaluation
There are over 40 genetic syndromes with femoral bowing (Alanay et al. 2007). Consultation with a geneticist is essential to identify the syndrome.

2.5 Differential Diagnosis

2.5.1 Shortening

Congenital Short Femur
This is the mildest deformity in the spectrum of femoral hypoplasia/aplasia. It simply is a shorter femur compared to the contralateral side (Gillespie 1998; Gillespie and Torode 1983; Pappas 1983) and is frequently associated with knee laxity and genu valgus in the older child (Gillespie and Torode 1983; Sanpera et al. 1995). It is also a concomitant deformity in children with fibular hemimelia/hypoplasia.

2.5.2 Failure of Development

Proximal Focal Femoral Deficiency
Proximal femoral focal deficiency (PFFD) is part of the wide spectrum of congenital femoral deformity which can range from congenital coxa vara, congenital short femur, to almost complete absence of the femur (Gillespie 1998; Lange et al. 1978; Panting and Williams 1978; Pappas 1983). It can be diagnosed on prenatal ultrasound (Gonçalves et al. 1996). There are numerous different classification systems (Aitken 1969; Gillespie and Torode 1983; Pappas 1983; Sanpera et al. 1995). Clinically the thigh is shorter than the opposite side (unless bilateral) and with the hip and knee in flexion (Fig. 2.1). There is often associated external rotation of the thigh. Ortolani and Barlow tests are negative. Plain radiographs will confirm the diagnosis (Fig. 2.2a–e).

Distal Femoral Focal Deficiency
This is an extremely rare condition, and the distal femoral epiphysis may be present with a short distal femoral segment and present with a pseud-

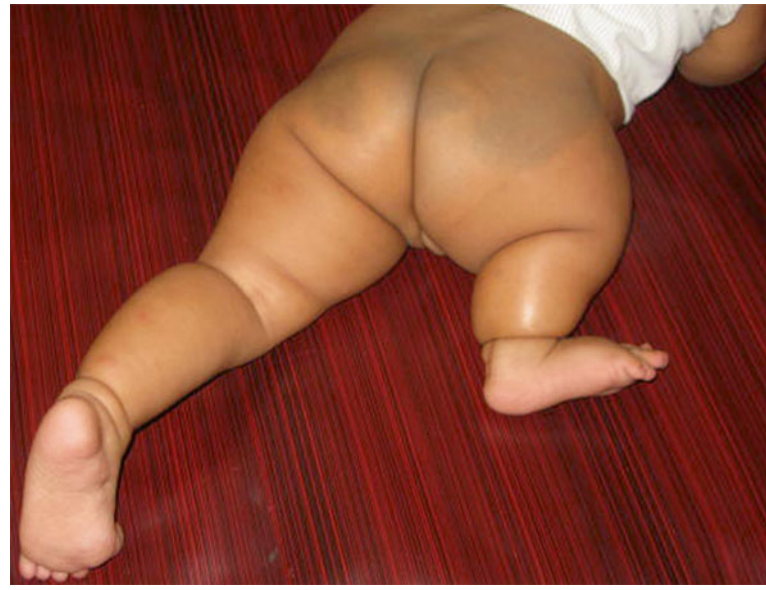


Fig. 2.1 A clinical photograph of a child with proximal femoral focal deficiency. In this severe case, the ankle in the involved extremity is opposite the level of the knee in the normal extremity

arthrosis between the distal femur and the more proximal shaft or completely absent distal femoral physis and epiphysis (Gilsanz 1983; Taylor et al. 2009) (Fig. 2.3a–c).

2.5.3 Bowing Without Shortening

Osteogenesis Imperfecta
Children with osteogenesis imperfecta may be born with bowing of the femur, and often the bowing may be demonstrated prenatally on the ultrasound scan. Osteogenesis imperfecta is the third most common cause for prenatal femoral bowing (Alanay et al. 2007). The bowing is most commonly in the proximal third and most frequently the bowing is anterolateral (Fig. 2.4).

2.5.4 Bowing with Shortening

Campomelic Dysplasia (Synonyms: Camptomelic Dysplasia, CMPD)
Campomelic dysplasia may be diagnosed prenatally at 13 weeks of gestation by bowing of the femora. At birth the femurs are short and bowed. The mild or moderate bowing of the femur is in the anterolateral direction at the proximal third of the bone (Fig. 2.5). The tibia is also bowed anterolaterally in the distal third. Clubfeet, hypoplastic fibulae, developmental dysplasia of the hips, cleft palate, abnormal genitalia in the male, and tracheomalacia may be present. Another radiographic feature is absent ossification of the pedicles of thoracic vertebrae. Upper cervical instability may be present. Campomelia is the most common of over 40 distinct entities that present with femoral bowing at birth or prenatally (Alanay et al. 2007).

2.5.5 Duplication

Bifid Femur
This is usually a duplication of the distal end of the femur, frequently associated with tibial aplasia (Kotakemori and Ito 1978; Aalami-Harandi and Zahir 1976; Küsswetter et al. 1976)

2.5.6 Rare Conditions

Kyphomelic Dysplasia
This is an autosomal recessive syndrome with the most striking feature being angulation of the femora which are also short and broad with flared

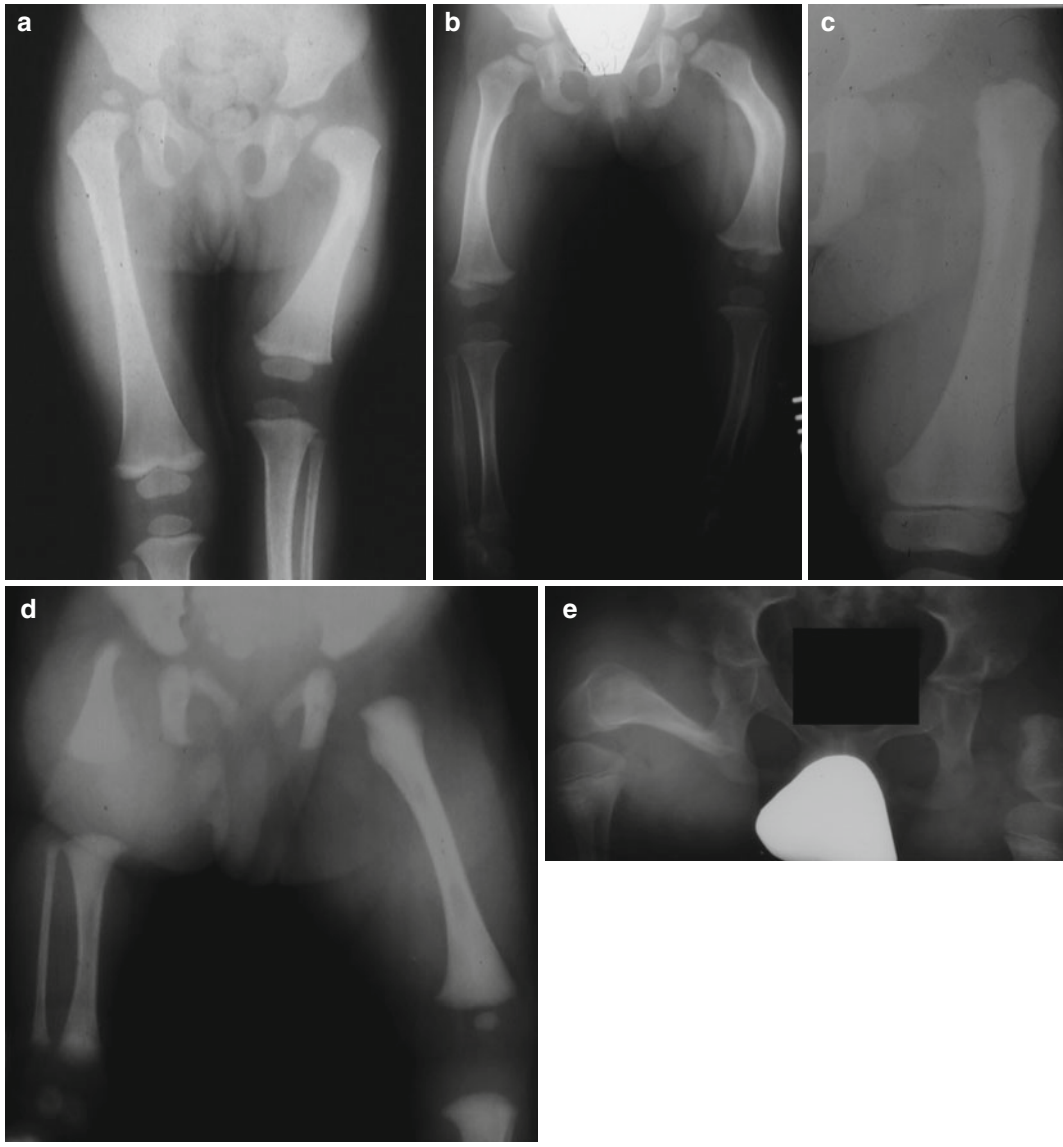


Fig. 2.2 Variations in the radiographic appearance of children with proximal femoral focal deficiency (PFFD): (**a**) A congenital short femur with minimal bowing. (**b**) A congenital short femur with moderate bowing. (**c**) A mild PFFD with pseudarthrosis of the femoral neck and intertrochanteric region. (**d**) A moderate PFFD, with no hip joint but with some distal femoral remnant. (**e**) Severe bilateral PFFD, with essentially no proximal femur bilaterally

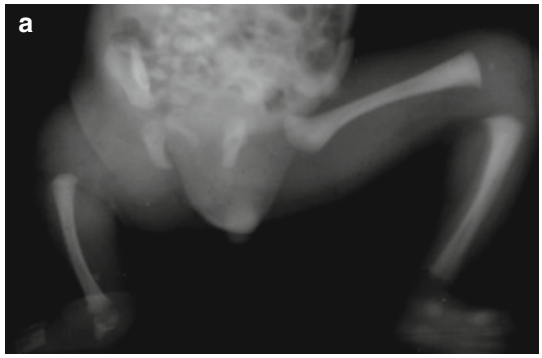
metaphyses (Cormier-Daire et al. 2004; Turnpenny et al. 1990); often there is bowing of the radius and tibia.

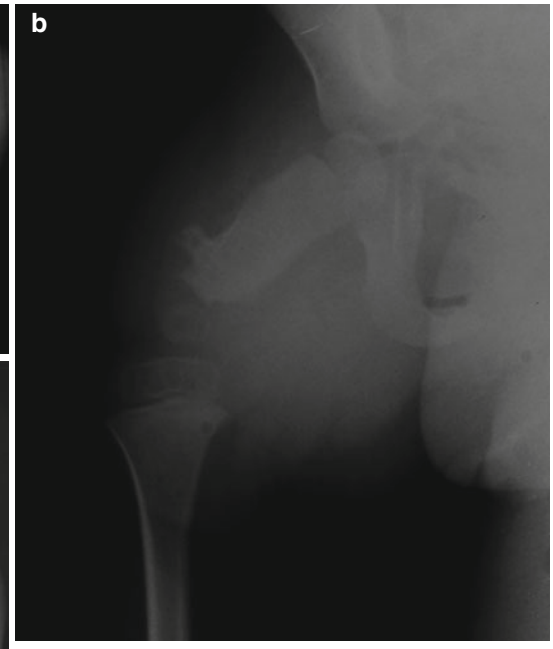
Stüve-Wiedemann Syndrome

This is an autosomal recessive syndrome comprising of long bone bowing, scoliosis, and respiratory compromise (Cormier-Daire et al. 2004; Hassan et al. 2010). Although usually lethal in infancy, some children survive. The femora demonstrate coxa vara and bowing with knee hyperextension.

2.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Table 2.1.





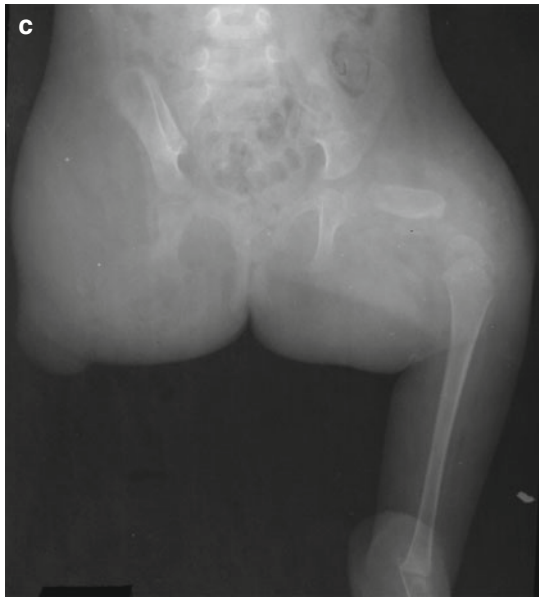


Fig. 2.3 Radiograph of the pelvis and lower limbs in a new born infant; the femur is not visualized on the right side, and the fibula is also absent on the same side (**a**). Over a period of time, the proximal femur has ossified, and now the nature and extent of distal focal femoral deficiency is evident (**b**). Another infant with distal focal femoral deficiency on the left and amelia on the right side (**c**)

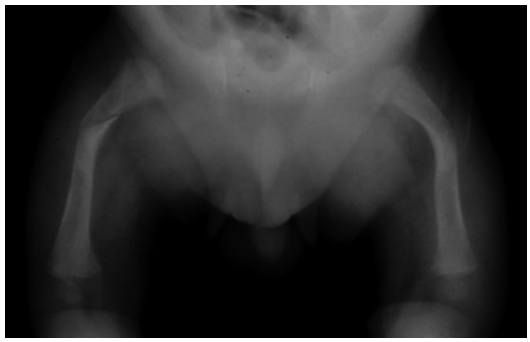


Fig. 2.4 Anterolateral bowing in a child with osteogenesis imperfecta

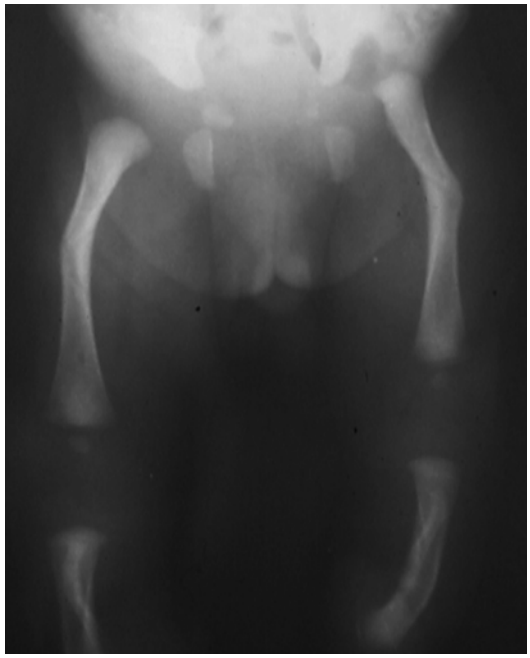


Fig. 2.5 Femoral bowing in a child with campomelic dysplasia

Table 2.1 Establishing the diagnosis of nature of abnormality in a newborn with a deformed femur

History					
–	History of maternal illness or substance abuse during pregnancy may be present	History of maternal illness or substance abuse during pregnancy may be present	–	–	History of maternal illness or substance abuse during pregnancy may be present
–	–	–	Positive family history may be present	Positive family history may be present	–
Physical examination					
Short femur	Very short femur	Very short femur	Normal sized femur in relation to body	Short femur	Femur may be of normal length or short
Unilateral involvement	May be unilateral or bilateral	May be unilateral or bilateral	Bilateral	Bilateral	Unilateral or bilateral
Femur not bowed	Femur not bowed	Femur not bowed distal end not palpable	Femur bowed anteriorly or anterolaterally Severity of bowing varies from mild to severe	Femur bowed anterolaterally in proximal third Bowing usually mild Skin dimple may be present over the bow	Femur bifid in the middle or distal third The duplicated femur may be on the medial or lateral side
Hip stable	Hip unstable	Hip stable	Hip stable	Hip may be unstable	Hip stable
Knee stable	Knee may be unstable	Knee very unstable	Knee stable	Knee stable	Knee stable
Tibia normal	Tibia may be normal	Tibia may be normal	Tibia may be bowed (bowing often anteriorly)	Tibia bowed anterolaterally at distal third	Tibia may be absent
Fibula normal	Fibula may be absent	Fibula may be absent	Fibula may be bowed (if tibia is bowed)	Fibula hypoplastic	Fibula may be absent
Foot normal	Foot normal	Foot normal	Foot normal	Clubfoot may be present	Foot may be deformed
			Blue sclera and ligament laxity may be present		
–	–	–	Upper limbs may be bowed	Upper limbs may be bowed	–
Working diagnosis: Congenital short femur	Working diagnosis: Proximal focal femoral deficiency	Working diagnosis: Distal focal femoral deficiency	Working diagnosis: Osteogenesis imperfecta	Working diagnosis: Campomelic dwarfism	Working diagnosis: Bifid femur
Investigations					
Plain radiograph will show a well formed femur Full-length films will be needed to demonstrate limb length inequality	Plain radiograph will show failure of normal formation of the proximal femur	Plain radiograph will show failure of normal formation of the distal femur	Plain radiograph will show generalized osteopenia Evidence of previous or fresh fractures and bowing of long bones of the upper and lower limbs	Plain radiograph will show bowing of the femur and tibia Hip dislocation is present and will be seen on the radiograph of the pelvis	Plain radiograph will show duplication of the femur with or without aplasia of the fibula or tibia
Diagnosis					
Congenital short femur	Proximal focal femoral deficiency	Distal focal femoral deficiency	Osteogenesis imperfecta	Campomelic dysplasia	Bifid femur (femoral duplication)

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The Deformed Knee

3

Randall T. Loder

3.1 Introduction

The deformed knee at birth indicates significant anatomic abnormalities. The deformity may be isolated or may be part of a more generalized syndrome. The questions that need to be answered to arrive at a diagnosis are listed below.

3.2 Questions to Establish a Diagnosis

- **Is the knee in extension, hyperextension, or flexion?**
- **Is there an associated rotational deformity at the knee?**
- **Is there webbing of the popliteal fossa?**
- **Is the deformity unilateral or bilateral and symmetrical?**
- **Is there a deformity of the foot with an abnormal number of toes?**
- **Is there a history of viral illness during pregnancy?**
- **Is there a family history of similar deformities?**
- **Was the delivery difficult?**
- **What is the range of motion of the knee?**

Is the knee in extension, hyperextension, or flexion?

Congenital knee dislocations are typically a hyperextension deformity. Knee flexion deformities are more commonly seen in neuromuscular conditions (arthrogryposis, myelomeningocele) or with congenital anomalies like tibial hemimelia and congenital dislocation of the patella. In arthrogryposis the knee may be stiff in extension without any hyperextension.

Is there an associated rotational deformity at the knee?

Rotational deformities, usually external, are often present in congenital dislocation of the patella. Multiplanar deformities may be seen in Larsen syndrome.

Is there webbing of the popliteal fossa?

This would indicate the pterygium syndrome.

Is the deformity unilateral or bilateral and symmetrical?

Bilateral knee deformities are more likely seen in arthrogryposis, syndromes, and hemimelias, while a transphyseal fracture is nearly always unilateral.

Is there a deformity of the foot with an abnormal number of toes and a short leg?

Tibial hemimelia often presents with a knee flexion contracture and associated clubfoot, often with extra toes, and will demonstrate varying degrees of tibial shortening.

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Is there a history of viral illness during pregnancy?

Exposures early in the pregnancy can predispose the infant to significant congenital malformations and/or syndromes; the exposures include viral illnesses (Martínez-Frías et al. 2001) and chemical or physical exposures (Coelho et al. 2000; Hall 1996). Abnormalities seen on prenatal ultrasonography likely indicate a syndromic or dysplastic issue.

Is there a family history of similar deformities?

This is important when assessing for a syndrome, although if a new mutation is involved, the family history would be negative.

Was the delivery difficult?

This might indicate an acute distal femoral transphyseal fracture.

What is the range of motion of the knee?

Congenital hyperextension deformity typically will demonstrate a good passive flexion arc of motion. Congenital dislocation of the knee may demonstrate a short arc of motion in hyperextension and can often not even be brought to neutral extension. No or only a few degrees of passive motion is typically seen in arthrogryposis, myelomeningocele, and pterygium syndrome deformities. Total loss of motion from the deformed position suggests that there is intra-articular or extra-articular ankylosis of the knee.

3.3 Physical Examination

3.3.1 Look

Identify whether the deformity is a flexion or extension deformity and whether there is a rotational component as well (Fig. 3.1). Observe if the deformity is unilateral or bilateral and symmetrical. Note if there are dimples or puckering over the front of the knee and if there is webbing behind the knee. Note if there are deformities of the hips and feet. Look for facial dysmorphism.

3.3.2 Feel

Palpate the front of the knee and if the patella is palpable determine if it is in the normal position and if it is mobile. Feel the hamstring tendons and see if they are tight if a flexion deformity is present. See if the quadriceps is tight if a hyperextension deformity is present.

3.3.3 Move

Note the passive range of motion of the knee. Also note the range of hip motion as developmental hip dysplasia is much more frequent in newborn babies with congenital knee dislocation.

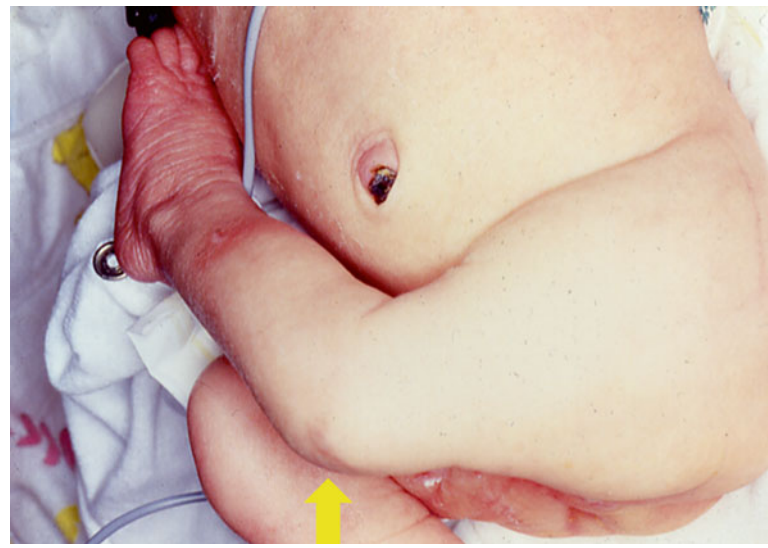

Fig. 3.1 Hyperextension deformity of the knee in a child with a genetic syndrome. Note the prominent posterior femoral condyles (*yellow arrow*)

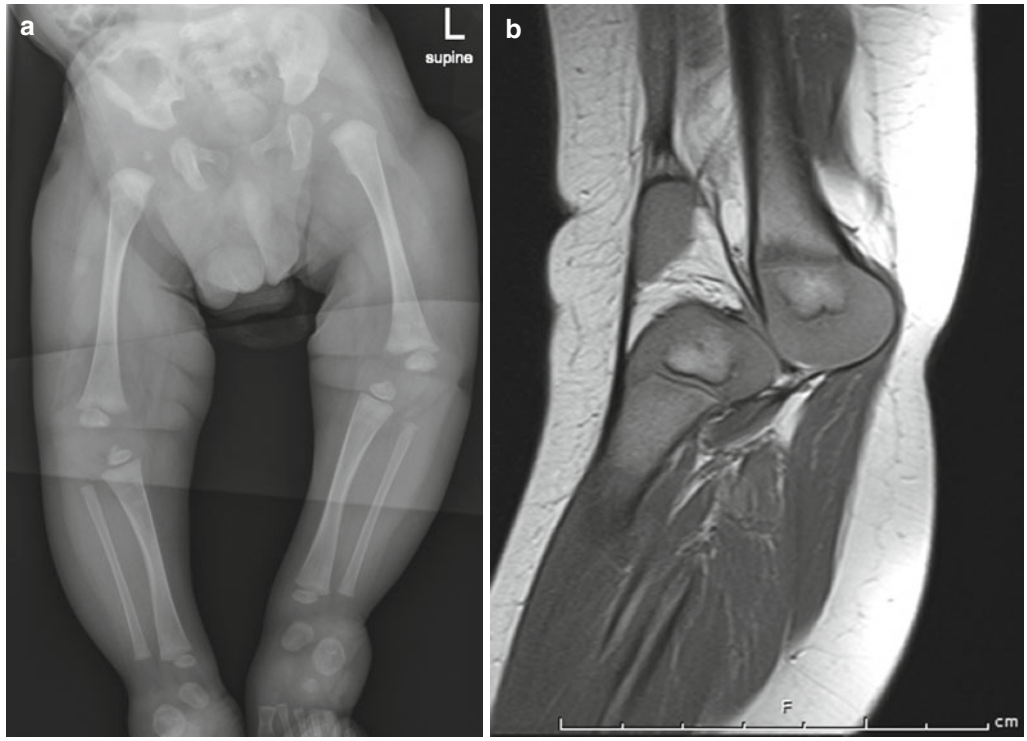

Fig. 3.2 (**a**) An AP radiograph of the lower extremities of a child with Larsen syndrome and bilateral congenital knee dislocations. Note the anterior displacement of the tibia relative to the femur along with rotational deformity, especially the left knee. (**b**) A sagittal MRI of the knee demonstrating anterior and proximal displacement of the tibia on the femur

Check if the ranges of movement of the other extremities are normal.

3.4 Investigations to Confirm the Diagnosis

Plain Radiography

This is typically the first step and will demonstrate any gross abnormality of the tibiofemoral relationship. It is important that true anteroposterior and lateral radiographs are obtained; appropriate positioning for these views may not be easy as the popliteal crease may be absent and bony landmarks may not be easily palpable especially in arthrogryposis with a knee dislocation.

A congenital knee dislocation will demonstrate the tibia anterior to the femur, occasionally with proximal migration as well (Fig. 3.2). Simple hyperextension deformity will not demonstrate anterior subluxation of the tibia relative to the femur. Radiographs of the leg will assist in the diagnosis of tibial hemimelia (Fig. 3.3). Posterior subluxation of the tibia relative to the femur may be seen in severe knee flexion contractures associated with arthrogryposis, pterygium syndrome, and tibial hemimelia.

Ultrasound

This can be very useful to determine the anatomy between the cartilaginous distal femoral and proximal tibial epiphyses. If the tibia cannot be visualized on plain radiographs, an ultrasound scan can determine if there is a cartilaginous proximal tibial remnant and differentiate the various types of tibial hemimelia (Fig. 3.3c). Hip ultrasound may be needed to properly ascertain hip status in the hyperextended knee.

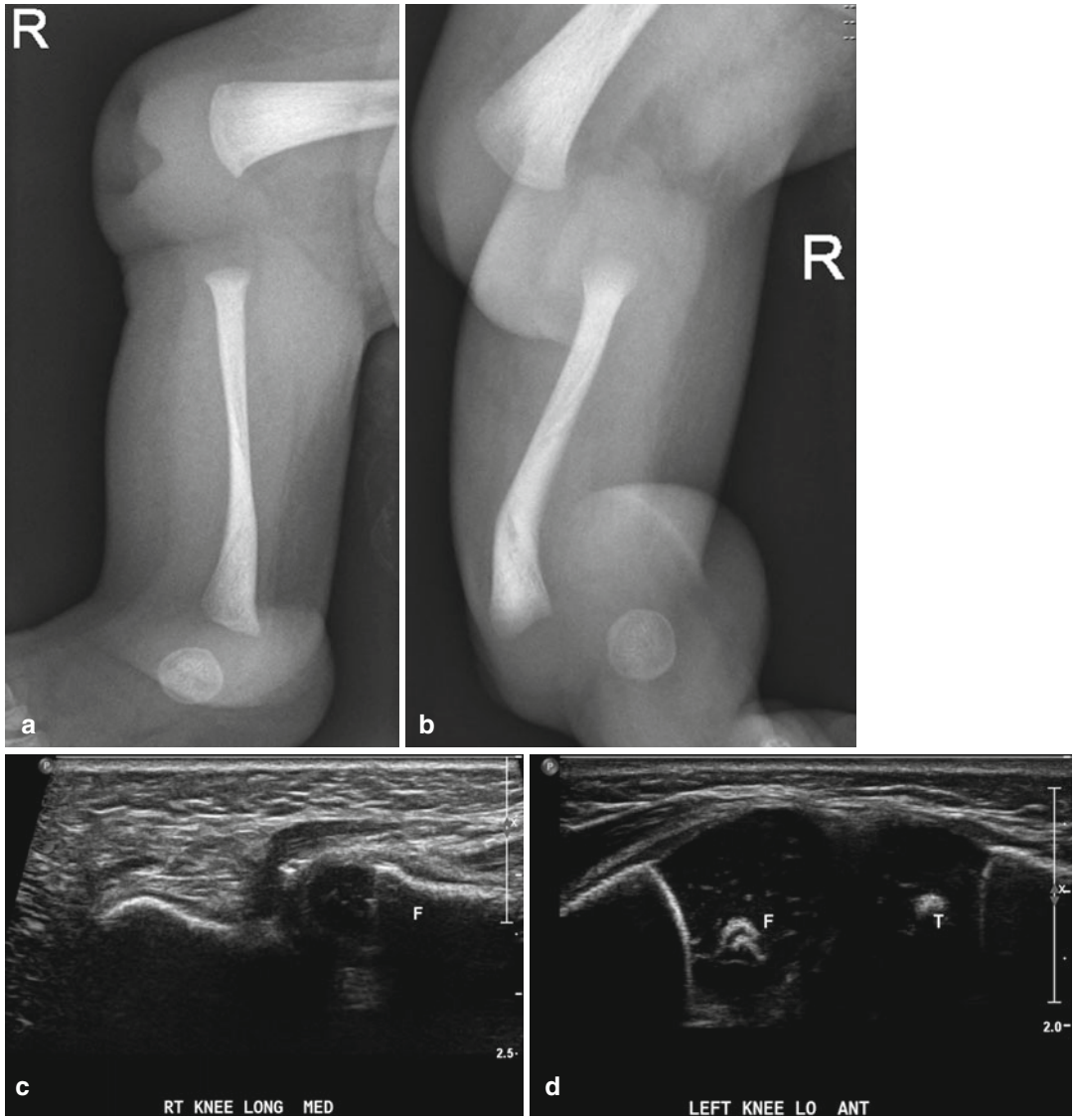

Fig. 3.3 (**a**, **b**) AP and lateral radiographs of the knee of a child with complete tibial hemimelia (Jones type 1a). (**c**, **d**) An ultrasound demonstrating no cartilaginous proximal tibial epiphysis on the right (**c**) but with one present on the left (**d**)

MRI

This is not commonly needed but will give detailed information regarding the femoral-tibial relationships as well as the absence or presence and location of the patella, status of the cruciate ligaments, and quadriceps and hamstring muscles and their insertions.

Genetic Evaluation

Genetic studies are frequently necessary to assist in the diagnosis of a syndrome.

3.5 Differential Diagnosis

3.5.1 Flexion Deformity

Neuromuscular Causes

Arthrogryposis

Flexion knee deformities are very common in children with arthrogryposis (Bernstein 2002; Bevan et al. 2007; Lampasi et al. 2012; Sarwark et al. 1990). The deformity is usually very rigid

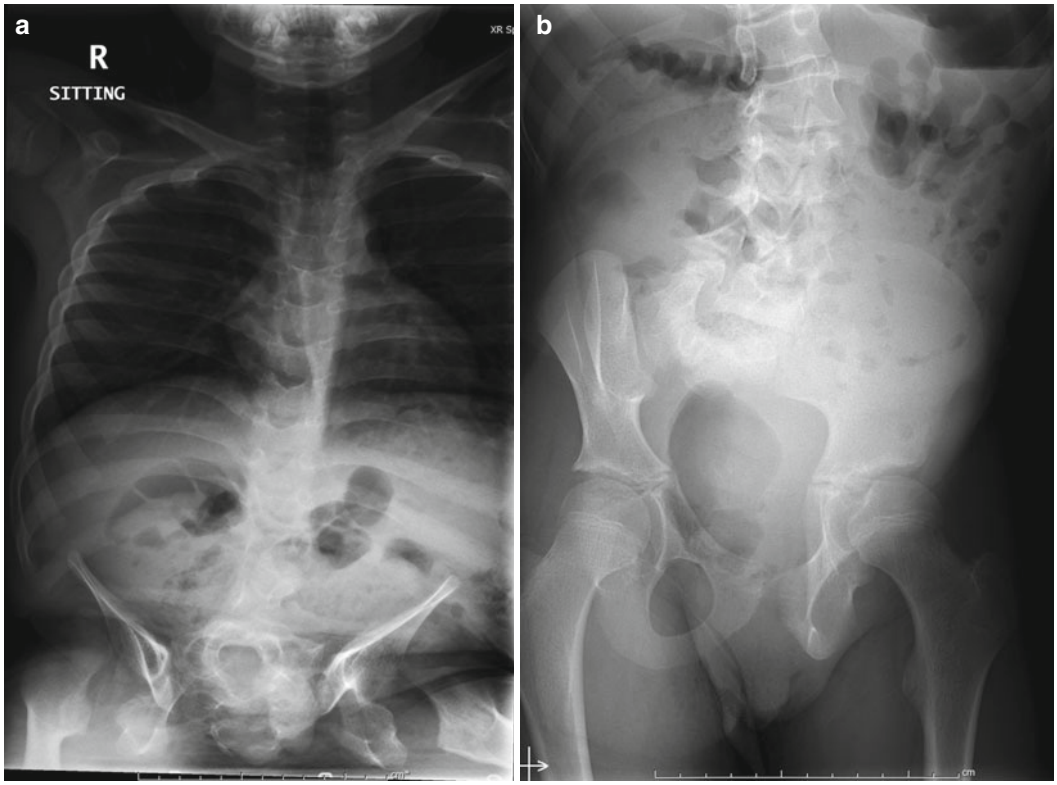

Fig. 3.4 (**a**, **b**) Caudal regression syndrome; complete absence of the last four lumbar vertebrae, sacrum, and coccyx (**a**) and partial absence of the distal sacrum (**b**)

with minimal passive motion; active motion is usually not present. There are often anterior skin dimples over the knee, and the skin is smooth with few flexion creases. Pterygium deformity may be present in the most severe cases.

Spina Bifida

Flexion deformity of the knee is not uncommon in spina bifida, especially higher level lesions. The diagnosis of spina bifida is usually evident after inspection and physical examination of the spine. The deformity is often rigid with a small arc of passive motion; active knee extension is usually absent.

Sacral Agenesis (Caudal Regression Syndrome)

Sacral agenesis is a congenital absence of the sacrum, either partial or complete; caudal regression is a more severe involvement up to complete absence of the lumbar spine, sacrum, and coccyx with a synchondrosis between the two iliac wings (Adra et al. 1994; Banta and Hichols 1969; Boulas 2009; Guidera et al. 1991; Merello et al. 2006; Phillips et al. 1982). The child is often born to a diabetic mother (Banta and Hichols 1969; Blumel et al. 1959; Passarge and Lenz 1966; Perrot et al. 1987; Phillips et al. 1982; Renshaw 1978). The knee deformity appears similar to that in spina bifida; however, clinical examination of the spine demonstrates no open dysraphism. The flexion contracture can be associated with a pterygium syndrome (Banta and Hichols 1969; Phillips et al. 1982). In more involved cases, the lumbar spine is shorter than normal or completely absent in the most severe case (Banta and Hichols 1969; Phillips et al. 1982). Plain radiographs usually confirm the diagnosis of sacral agenesis (Fig. 3.4).

Congenital Dislocation of the Patella

Congenital dislocation of the patella may occur as an isolated deformity or in concert with some other syndrome, such as the nail-patella syndrome (Ghanem et al. 2000). The patella may be dislocated at birth or become dislocated

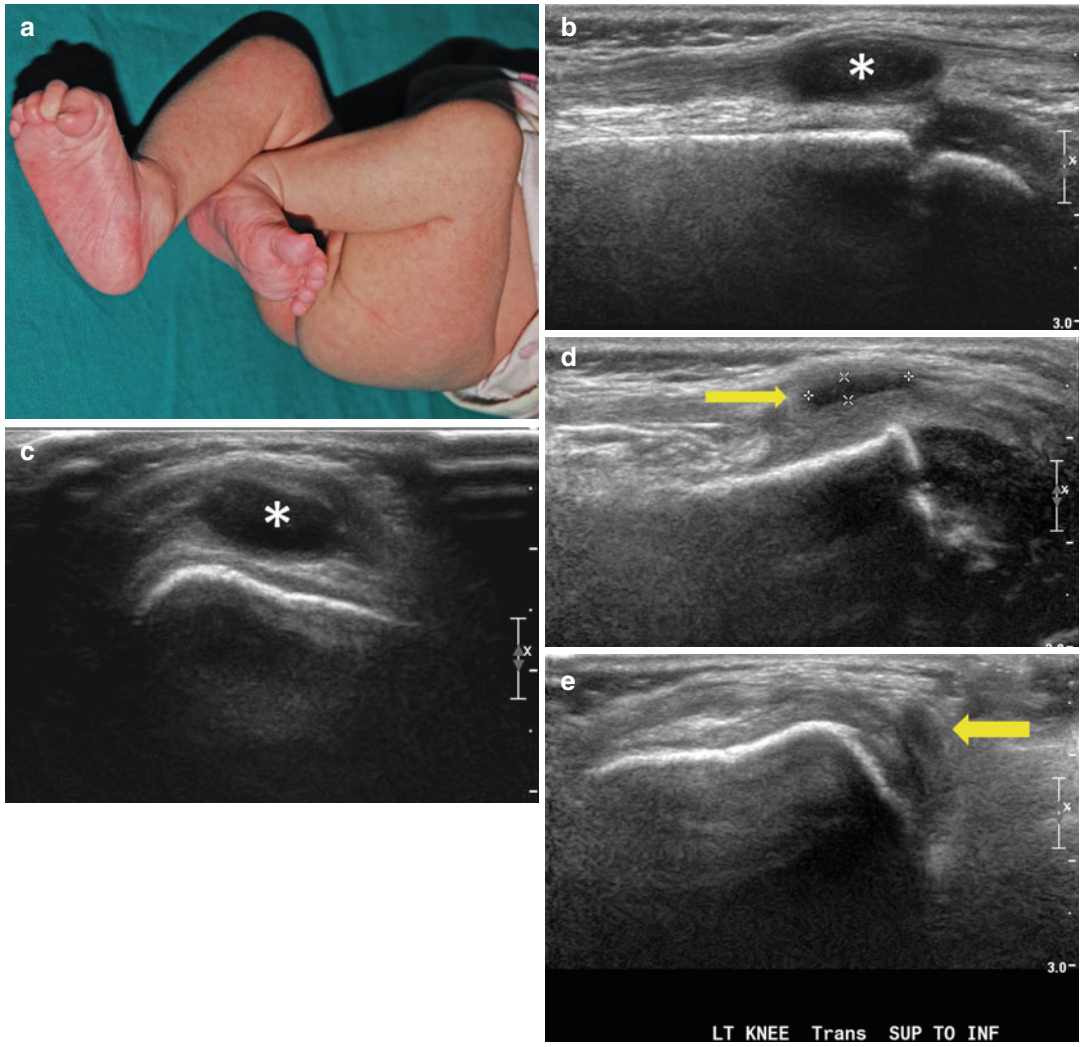

Fig. 3.5 Congenital dislocation of the left patella (**a**); the flexion deformity of the knee and external rotation of the leg are seen. The sagittal (**b**) and transverse (**c**) ultrasound scans of the right knee demonstrate a normal size patella (*asterisk*) in the normal position. The ultrasound scans of the left knee (**d**, **e**) demonstrate a hypoplastic patella (*yellow arrow*) lateral to the femur

within the first year or two of life due to a hypoplastic patella and/or femoral trochlear groove (Ghanem et al. 2000; Jones et al. 1976; Stanisavljevic et al. 1976). The hallmark feature of either type is limited active extension of the knee, genu valgum, and external tibial rotation with secondary lateral tibial tubercle position (Fig. 3.5). Congenital dislocation of the patella usually presents within the first year or two of life as a delay or increasing difficulty in walking. This difficulty in walking is due to the knee flexion contracture, extensor weakness, and external tibial rotation (Langenskiöld and Ritsilä 1992).

Congenital Popliteal Pterygium Syndrome/ Multiple Pterygium Syndrome

This is the very characteristic affecting the face (cleft lip/palate), limbs, and genitalia (Froster-Iskenius 1990; Herold et al. 1986; Parikh et al. 2004). The most pertinent extremity feature is the large popliteal pterygium (Fig. 3.6), often extending from the ischium to the calcaneus. There is minimal knee motion. The foot is often in fixed equinus.

3.5.2 Extension Deformity

Isolated Congenital Dislocation of the Knee
Congenital dislocation of the knee may occur as an isolated deformity or in association with some other syndrome, such as Larsen syndrome, myelomeningocele, or arthrogryposis (Ko et al. 1999) (Fig. 3.7). The deformity ranges from a mild hyperextension deformity which rapidly responds to simple stretching to frank, complete dislocation with proximal migration of the tibia relative to the distal femur. The child frequently has other anomalies of the musculoskeletal system, typically hip dysplasia and foot deformity (Ooishi et al. 1993). Absence of facial dysmorphism excludes Larsen syndrome; children with arthrogryposis typically have symmetrical contractures/deformities in all four extremities and severe foot deformities (e.g., talipes equinovarus or congenital vertical talus) (Bernstein 2002; Bevan et al. 2007; Hall 1997).

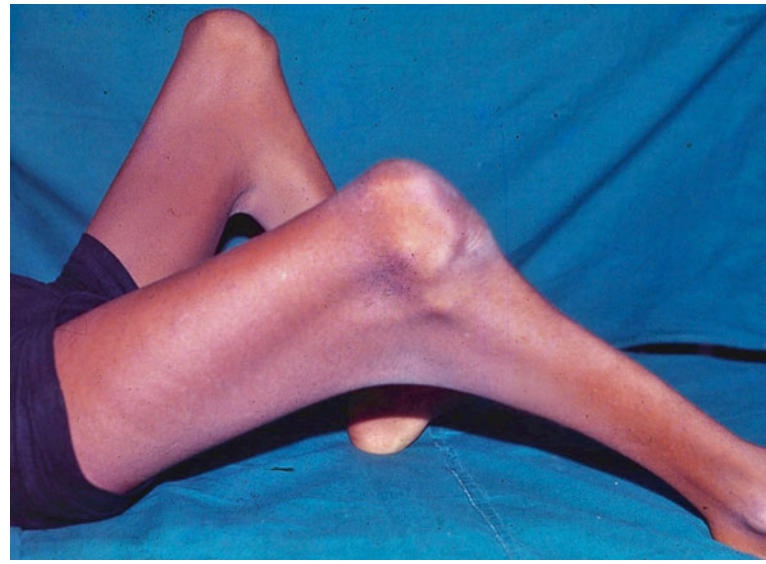

Fig. 3.6 Popliteal pterygium in an older child

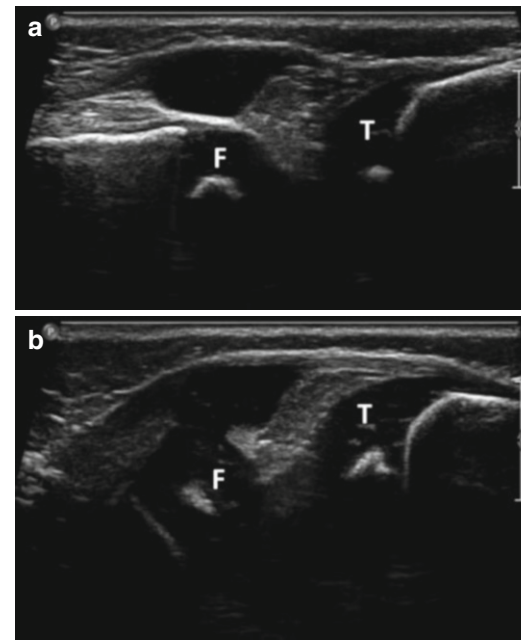

Fig. 3.7 Hyperextension deformity of the knee in a child with high lumbar level myelomeningocele; an ultrasound scan in extension demonstrates the hyperextension deformity but without any dislocation or proximal displacement of the proximal tibial epiphysis on the distal femoral epiphysis (**a**). An ultrasound scan in flexion (**b**) demonstrates that the deformity is easily correctible. *F* distal femoral epiphysis, *T* proximal tibial epiphysis

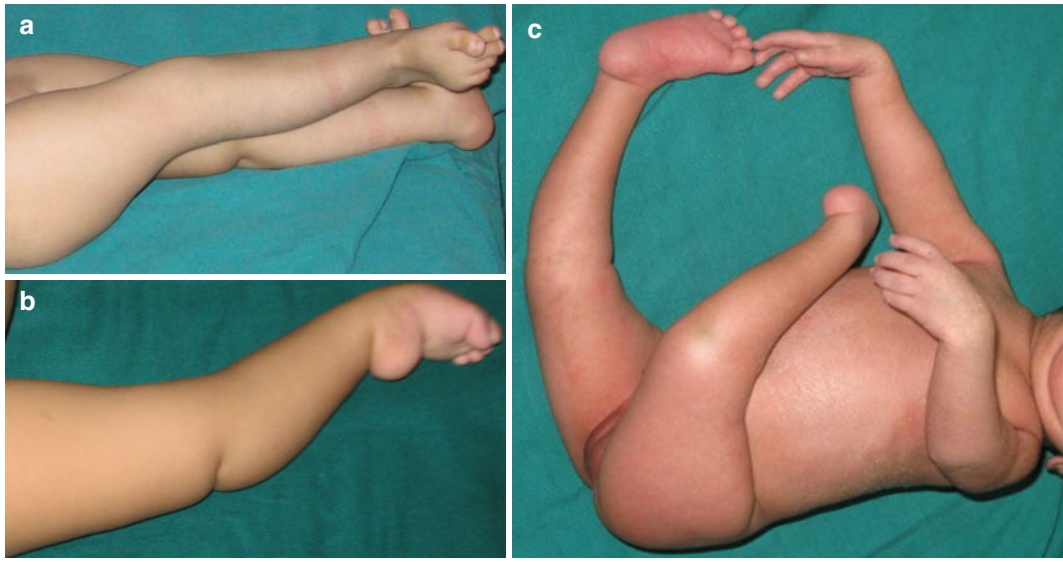

Fig. 3.8 Quadriceps contracture in arthrogryposis may vary in severity; the knee may be in extension (**a**), mild hyperextension (**b**), or severe hyperextension as seen on the left lower limb of this infant (**c**)

Congenital Dislocation of the Knee Associated with Arthrogryposis

Hyperextension with or without knee dislocation may be seen in arthrogryposis (Bernstein 2002; Sarwark et al. 1990) (Fig. 3.8a–c). Knee dislocation is rare in arthrogrypotic extension contractures of the knee (Bevan et al. 2007; Lampasi et al. 2012) although proximal and lateral migration of the patella is commonly seen (Borowski et al. 2008).

Congenital Dislocation of the Knee in Larsen Syndrome

Larsen syndrome (Larsen et al. 1950) is a constellation of multiple joint dislocations (hip, elbow, knee), severe foot deformities (equinovarus or equinovalgus), and dysmorphic facies. It is due to a mutation in the filamin B protein (Zhang et al. 2006). It can often be diagnosed prenatally with ultrasound (Rochelson et al. 1993; Tongsong et al. 2000). Congenital dislocation of the knee commonly occurs in association with Larsen syndrome (Houston et al. 1981; Laville et al. 1994; Rønningen and Bjerkreim 1978; Steel and Kohl 1972) (Fig. 3.2).

Congenital Quadriceps Contracture without Dislocation

This is extremely rare in the absence of other syndromes. The child will have a hyperextended knee and/or limited flexion (Chiu et al. 1974; Gammie et al. 1963; Gunn 1964; Nozawa et al. 2004; Özdemir et al. 2006). Historically they were associated with injections into the thigh muscles (Chiu et al. 1974), although there are often other etiologies (Williams 1968).

3.5.3 Rare Conditions

Distal Femoral Epiphyseal Separation (Birth Trauma)

This is an extremely rare condition (Jain and Bielski 2001; Krosin and Lincoln 2009; Mangurten et al. 2005). The infants are often large for gestational age and/or have associated joint contractures (e.g., arthrogryposis, spina bifida). The knee is swollen with pseudoparalysis. Radiographs will demonstrate that the femur and tibia are not aligned; the diagnosis can be especially difficult if the distal femoral epiphysis has not yet ossified; in this case, ultrasound can be quite helpful (Brown and Eustace 1997).

Congenital Aplasia of the Patella and Quadriceps

This is quite rare (Oner et al. 2013) and needs to be differentiated from the nail-patella syndrome which in addition to patellar hypoplasia/aplasia has associated elbow abnormalities, iliac horns,

and renal and ophthalmologic (glaucoma) involvement (Sweeney et al. 2003). It can also be associated with other syndromes, such as the Meier-Gorlin (Bongers et al. 2001) and genitopatellar (Cormier-Daire et al. 2000; Reardon 2002).

Congenital Ankylosis of the Knee

This is a rare condition and is usually a flexion deformity with absolutely no active or passive motion of the knee (Madadi et al. 2010; Yaniv et al. 2004). Imaging demonstrates a complete cartilaginous or osseous coalition.

3.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Tables 3.1 and 3.2.

Table 3.1 Establishing the diagnosis of a flexed knee at birth

History				
History of akinesia (diminished fetal movement in utero)	Born with swelling or open defect in the lumbosacral region	History of maternal diabetes may be present	Nothing contributory in antenatal history	History of akinesia (diminished fetal movement in utero) may be present
Physical examination				
Flexion deformity of knee	Flexion deformity of knee	Flexion deformity of knee	Flexion deformity of knee + External rotation deformity of the leg	Flexion deformity of knee + Webbing in popliteal region
Other knee usually symmetrically involved	Other knee often (not always) symmetrically involved	Other knee often (not always) symmetrically involved	Other knee occasionally involved	Other knee often (not always) involved
Other regions: Hips, feet, wrists, elbows, and shoulders may be deformed	Other regions: Hips and feet often deformed but upper limbs spared	Other regions: Hips and feet often deformed but upper limbs spared Anorectal anomaly may be present	Other regions: No other region affected	Other regions: Cubital web and webbing of the neck may be present Cleft lip/palate, syndactyly, bifid scrotum in males, and hypoplastic labia in girls may be present
Quadriceps weakness present	Quadriceps weakness present	Quadriceps weakness present	Quadriceps ineffective (since quadriceps mechanism no longer anterior to axis of knee) Muscle not actually weak	Quadriceps often weak
No spinal deformity at birth	Defect in posterior elements of lumbar or lumbosacral spine Spinal deformity may be present at birth	Sacrum not palpable and the two posterior iliac spines almost touching each other Spinal deformity may be present at birth	No spinal deformity	Spinal deformity occasional
No sensory loss	Sensory loss present	Sensory loss may not be present	No sensory loss	No sensory loss

(continued)

Table 3.1 (continued)

Muscle weakness affecting upper and lower limbs	Muscle weakness affecting the lower limbs	Muscle weakness affecting the lower limbs	No other muscle weakness	Muscle weakness not a feature
Working diagnosis: Arthrogryposis	Working diagnosis: Spina bifida	Working diagnosis: Sacral agenesis	Working diagnosis: Congenital dislocation of patella	Working diagnosis: Popliteal pterygium
Investigations				
–	Plain radiograph of lumbosacral spine	Plain radiographs of pelvis and lumbosacral spine	Radiograph not useful as patella not ossified	–
–	–	–	Ultrasound may demonstrate lateral position of the patella	–
Diagnosis				
Arthrogryposis	Spina bifida	Sacral agenesis	Congenital dislocation of the patella	Popliteal pterygium/ multiple pterygium syndrome

Table 3.2 Establishing the diagnosis of an extended or hyperextended knee at birth

History			
History of akinesia (diminished fetal movement in utero) may be present	Antenatal history not contributory	History of akinesia (diminished fetal movement in utero)	Antenatal history not contributory
Physical examination			
Extension deformity of the knee	Hyperextension deformity of the knee	Hyperextension deformity of knee	Hyperextension deformity of knee
Other knee usually symmetrically involved	Other knee may be involved	Other knee usually symmetrically involved	Other knee usually symmetrically involved
Other regions: Hips, feet, wrists, elbows, and shoulders may be deformed	Other regions: DDH may be present	Other regions: Hips, feet, wrists, elbows, and shoulders may be deformed	Other regions: Hips may be dislocated Feet may be deformed
Stiff joints	Other joints are normal	Stiff joints	Hypermobile joints
Face normal	Face normal	Face normal	Facial dysmorphism
Working diagnosis: Congenital quadriceps contracture without knee dislocation	Working diagnosis: Isolated congenital dislocation of the knee	Working diagnosis: Congenital knee dislocation associated with arthrogryposis	Working diagnosis: Congenital knee dislocation associated with Larsen syndrome
Investigations			
Plain lateral radiograph of the knee: No dislocation	Plain lateral radiograph of the knee: Dislocation	Plain lateral radiograph of the knee: Dislocation	Plain lateral radiograph of the knee: Dislocation Lateral radiograph of foot may show additional ossific center of calcaneum Lateral radiograph of the elbow may show dysplastic lower end of humerus
Diagnosis			
Congenital quadriceps contracture (including arthrogryposis)	Isolated congenital dislocation of the knee	Congenital dislocation of the knee in arthrogryposis	Congenital dislocation of the knee in Larsen syndrome

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The Bowed Tibia

4

Benjamin Joseph

4.1 Introduction

Deformities of the tibia may be diagnosed on prenatal ultrasound scans (Begam et al. 2011), and it is important that the orthopedic surgeon is familiar with the spectrum of conditions that may manifest as a bowed tibia. While a prenatal diagnosis may be possible, in the majority of instances, a definitive diagnosis of the bowed tibia may only be possible once the child is born. Before examining the child, it is useful to list questions that need to be answered during the examination as the answers to these questions will help in establishing at least a working diagnosis, if not a definite diagnosis.

4.2 Questions to Establish a Diagnosis

- **In which plane is the deformity?**
- **What is the site of the deformity?**
- **Are there associated deformities of the foot?**
- **Are there other anomalies of the foot?**
- **Are both legs deformed symmetrically?**
- **If the deformity is unilateral, is there shortening of the leg or thigh of the affected side?**
- **Are there other extraskeletal clinical features that may aid the diagnosis?**

In which plane is the deformity?

The plane of the deformity can often help in making a diagnosis in children who are born with a deformed tibia. The tibia may be bowed anteromedially (Fig. 4.1a, b), anterolaterally, or posteromedially (Fig. 4.1c, d).

What is the site of the deformity?

Bowing of the tibia is most frequently seen at the junction of the middle and lower third of the bone when the underlying problem is a congenital anomaly.

Are there associated deformities of the foot?

The common deformities of the foot that may be associated with a bowed tibia at birth are equinovalgus and calcaneovalgus (Fig. 4.2a); the former is associated with an anteromedial bow and the latter with a posteromedial bow. Less frequently an equinus or equinovarus deformity may be noted (Figs. 4.2b) and in some instances the foot may not be deformed.

Are there other anomalies of the foot?

There may be a loss of lateral rays of the foot in association with anteromedial bowing of the tibia (Fig. 4.3). This is characteristically seen in fibular aplasia. Absence of lateral rays may not be seen in children with symmetrical anteromedial bow deformities (Fig. 4.4).

Are both legs deformed symmetrically?

If both the lower limbs have identical deformities, it is possible that the child may have a

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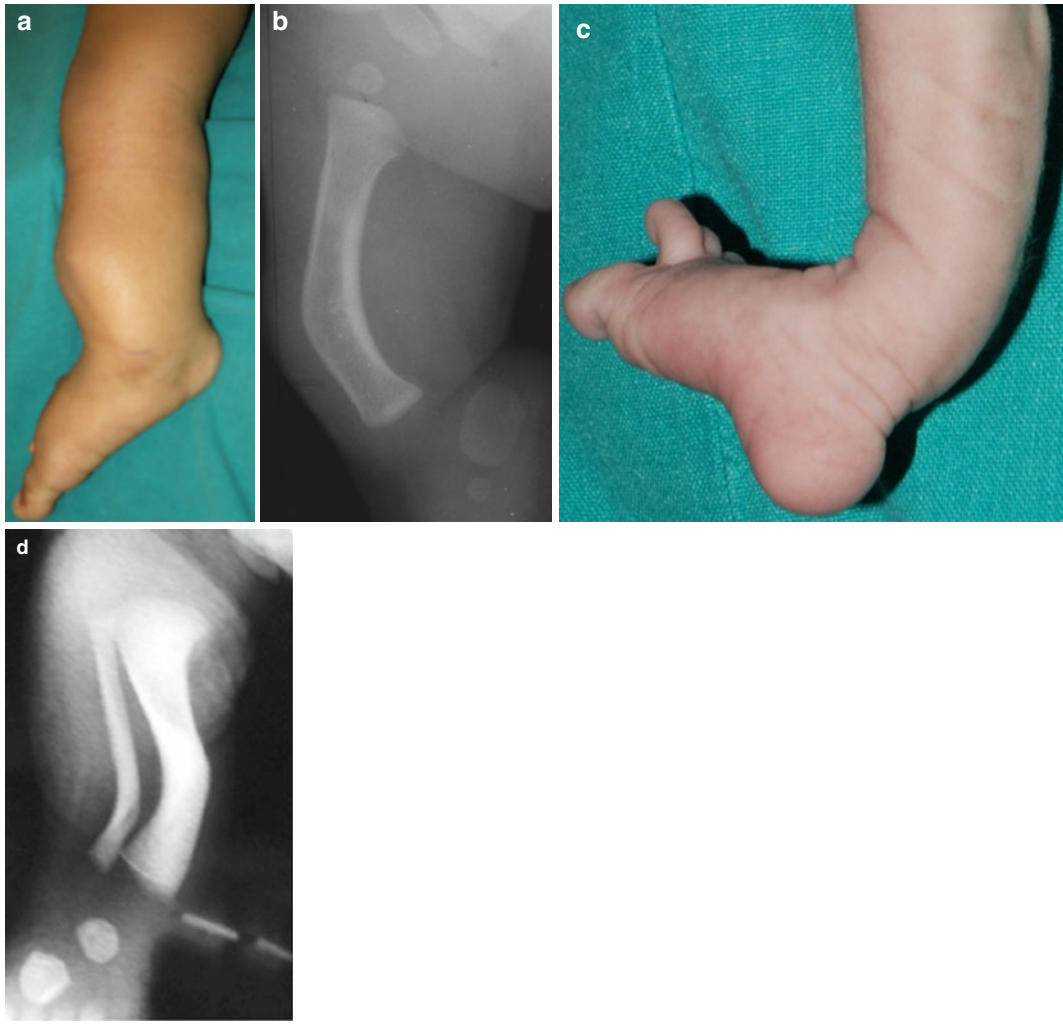

Fig. 4.1 Anteromedial bowing in fibular hemimelia (**a**, **b**) and congenital posteromedial bowing (**c**, **d**)

form of mesomelic skeletal dysplasia (Fig. 4.4). A careful assessment of body proportions may confirm the mesomelic pattern with shortening of the leg segment.

If the deformity is unilateral, is there shortening of the leg or thigh of the affected side?

Shortening of the thigh and the leg are characteristically seen in children with a major congenital reduction defect such as fibular hemimelia (Fig. 4.5a). Occasionally the shortening of the thigh segment can be severe suggesting that there is an underlying deficiency of the femur in addition to the fibular deficiency. In children with identical bilateral deformities and shortening, the mesomelic segment alone is short and this results in altered body proportions (Fig. 4.5b).

Are there other extraskeletal clinical features that may aid the diagnosis?

Presence of café au lait spots suggests that the child has neurofibromatosis (NF-1) that is frequently associated with anterolateral bowing. These spots may not always be present on the extremities, and so the torso must be examined

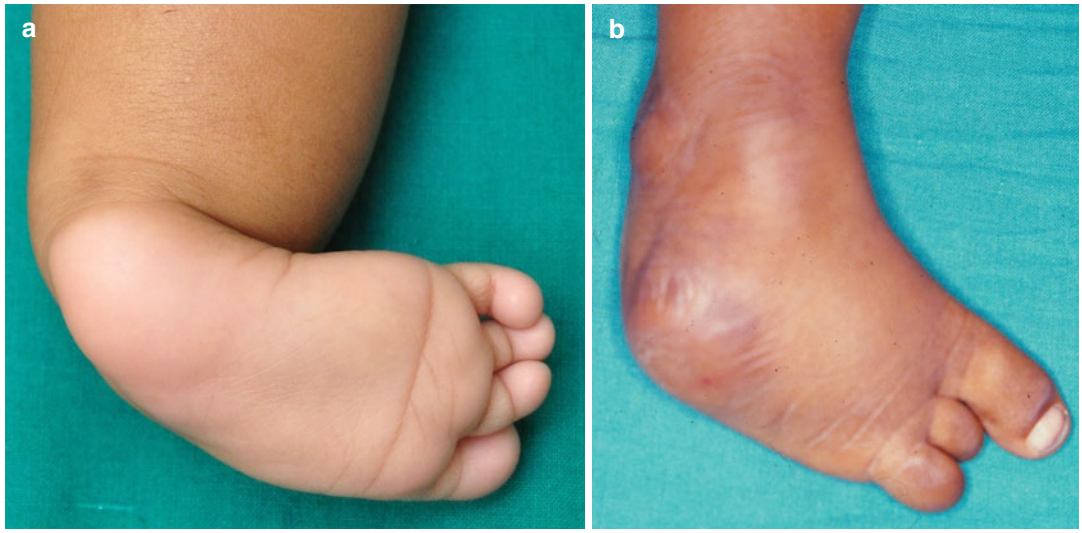

Fig. 4.2 Tibial bowing associated with calcaneovalgus (**a**), equinovarus (**b**)

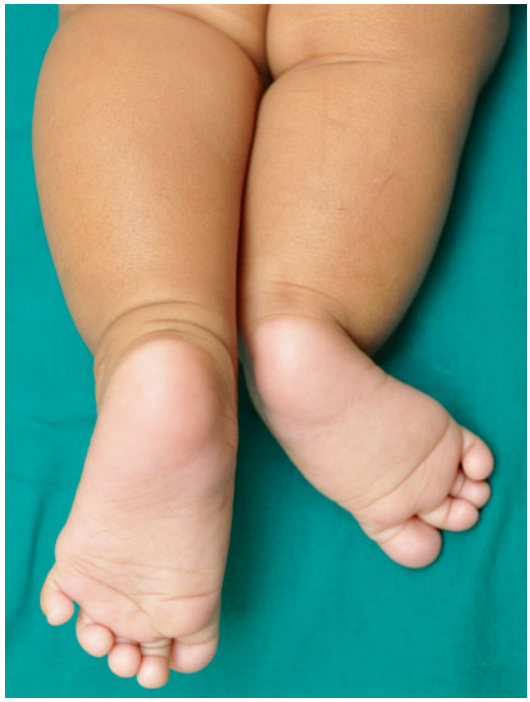

Fig. 4.3 Loss of rays seen in association with anteromedial bowing in children with fibular hemimelia

after disrobing the baby (Fig. 4.6). A blue sclera may indicate that the child has a form of osteogenesis imperfecta.

4.3 Investigations to Confirm the Diagnosis

4.3.1 Plain Radiographs

The diagnosis can be made from plain radiographs of the affected limbs.

Radiological Features of Fibular Hemimelia

- Fibula: May be totally or partially absent.
- Tibia: Bowed anteromedially; the severity of the bow varies from very mild to severe.

  The tibia is shorter than normal.
- Tarsal bones: Often massive talocalcaneal coalition is present (the coalition may only become evident on plain radiographs in the older child).
- Rays: Often lateral rays missing; the number of missing rays varies.

Radiological Features of Pre-pseudarthrotic Anterolateral Bowing of the Tibia

- Tibia: Bowed anterolaterally most frequently at the junction of the lower and middle third of the leg and less frequently in the middle third or the distal third.

  The medullary cavity at the site of the bow may be partially or totally obliterated.

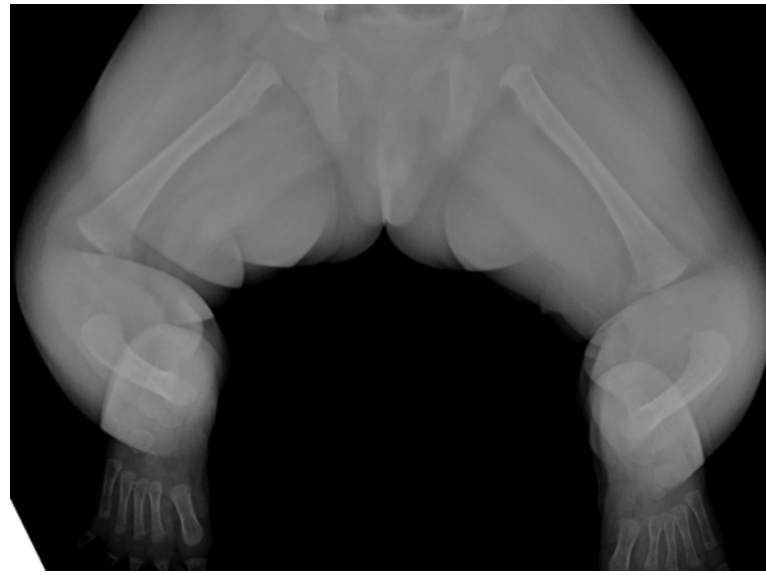

Fig. 4.4 Radiograph of the lower limbs of an infant with a form of mesomelic dysplasia with bilateral fibular hemimelia. The deformities of the legs and feet are symmetrical and there are no missing rays of the feet

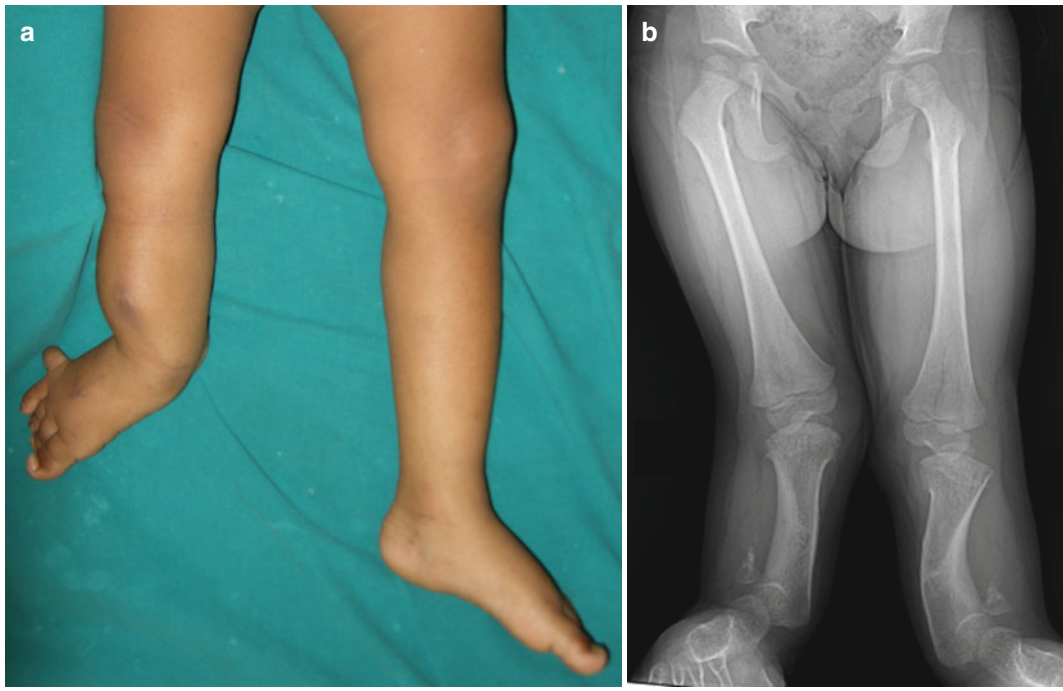

Fig. 4.5 Significant shortening of the left leg is present in a child with fibular hemimelia (**a**); the femur is minimally shorter on the affected side. The mesomelic segments of the lower limbs are short in this child with mesomelic dysplasia with tibial bowing and fibular hemimelia (**b**)

The bone may be narrowed at the site of the bow. Very rarely, in the newborn there may be a fracture at the site of the bow.

- Fibula: The fibula may be normal or bowed and show changes similar to the tibia.
- Foot: The foot is normal at birth.

Radiological Features of Congenital Posteromedial Bowing of the Tibia

- Tibia: Bowed posteromedially at the junction of the middle and lower third.

  Medullary cavity not obliterated but may be narrow.

  The tibia is shorter than the opposite tibia and the degree of shortening appears to be related to the severity of bowing.
- Fibula: May be bowed.
- Foot: Calcaneovalgus deformity. The tarsals and the rays are normal.

4.3.2 Ultrasound

In children with major reduction defects such as fibular hemimelia, it is important to exclude visceral anomalies, and an abdominal ultrasound scan and echocardiography are useful for this purpose.

4.4 Differential Diagnosis

4.4.1 Fibular Hemimelia

Partial or complete agenesis of the fibula may be associated with bowing of the tibia. The severity of bowing varies from negligible bowing that can barely be discerned to bowing of almost 90°. The direction of bowing is anterior or anteromedial. A variable number of postaxial rays may be missing and the foot may be deformed; equinovalgus deformity is the most frequent deformity, and equinus and equinovarus are rarer. Plain radiographs are usually sufficient to determine if the fibular deficiency is partial or complete. The number of rays of the foot that are present appears to have a bearing on the outcome. If there are three or more rays, the chances of being able to salvage the foot seem higher than if there are fewer rays (Coventry and Johnson 1952; Achterman and Kalamchi 1979; Stanitski and Stanitski 2003).

4.4.2 Congenital Pseudarthrosis of the Tibia

Anterolateral bowing of the tibia is the precursor to congenital pseudarthrosis of the tibia (Fig. 4.6a). The bowed tibia is prone to fracture and the fracture usually occurs after the child starts walking (Grill et al. 2000; Joseph and Mathew 2000; Joseph et al. 2003). Rarely the fracture may occur in the infant (McKeown and Frazer 1961) (Fig. 4.6b). Treatment is particularly difficult when the fracture occurs very early in life. A large proportion of children with congenital pseudarthrosis of the tibia have neurofibromatosis type-I (NF1); café au lait spots on the skin are present at birth point to this association. Confirming the presence of neurofibromatosis is of relevance as the prognosis tends to be worse in children with this association. If NF1 is suspected, the spine must be evaluated.

4.4.3 Congenital Posteromedial Bowing of the Tibia

The severity of bowing and the degree of shortening have a bearing on the management (Napiontek and Shadi 2014). While milder degrees of bowing are likely to resolve spontaneously, complete resolution may not occur in children with severe bowing and this may necessitate surgery later in childhood (Pappas 1984; Badgley et al. 1952; Shah et al. 2009). If the shortening is mild, simple measures of limb length equalization such as contralateral epiphysiodesis may suffice; the more severe shortening may warrant formal limb lengthening procedures.

4.4.4 Rarer Causes of Bowing of the Tibia in the Newborn

Osteogenesis Imperfecta

A child with a severe form of osteogenesis imperfecta may have a bowed tibia on account of an intrauterine fracture. A family history of brittle bones, presence of blue sclera, and deformities of other long bones help to establish a diagnosis.

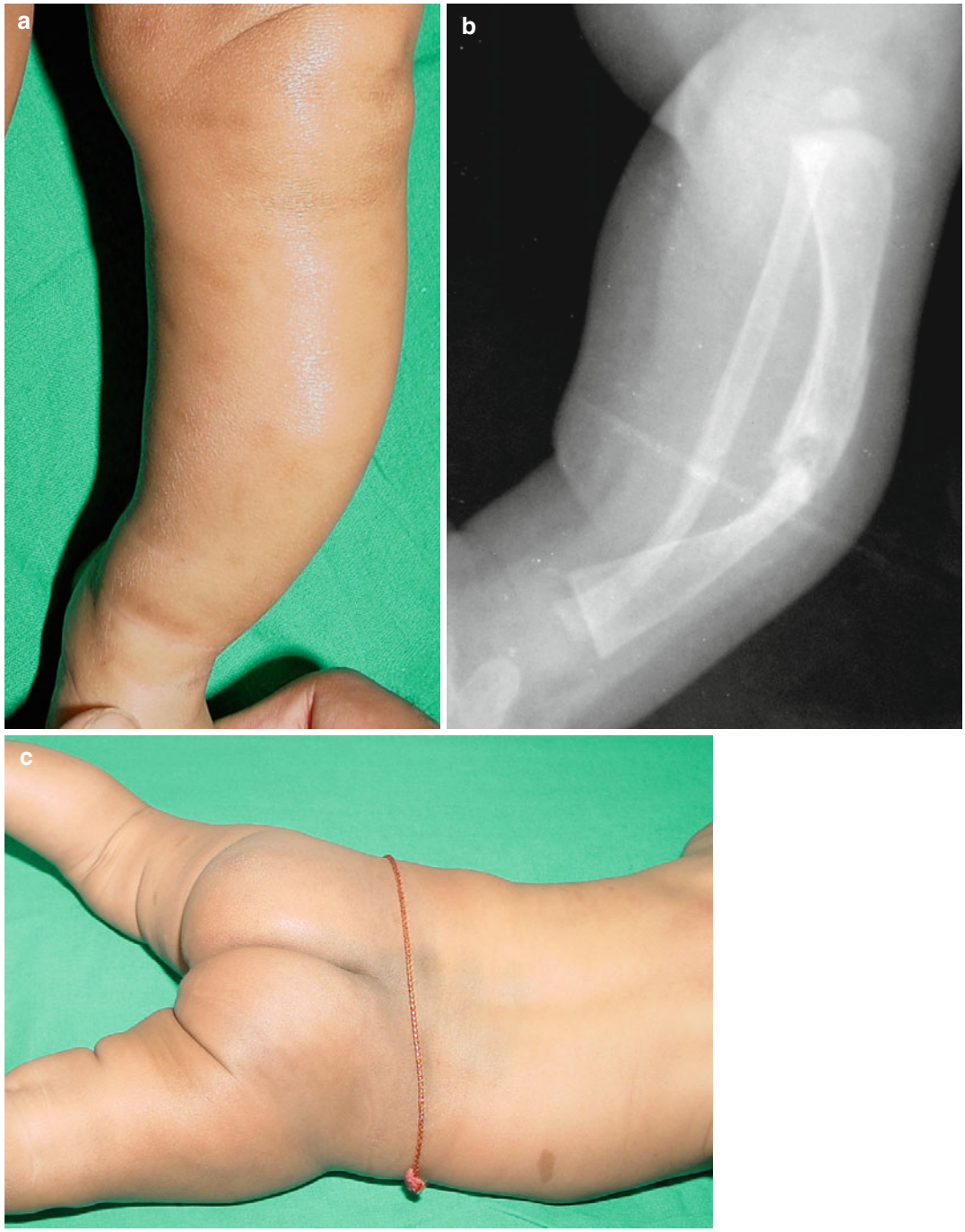

Fig. 4.6 Anterolateral bowing of the leg in a child with neurofibromatosis Type I (**a**). The pseudarthrosis of the tibia is seen (**b**); a café au lait spot is visible on the right side of the torso (**c**)

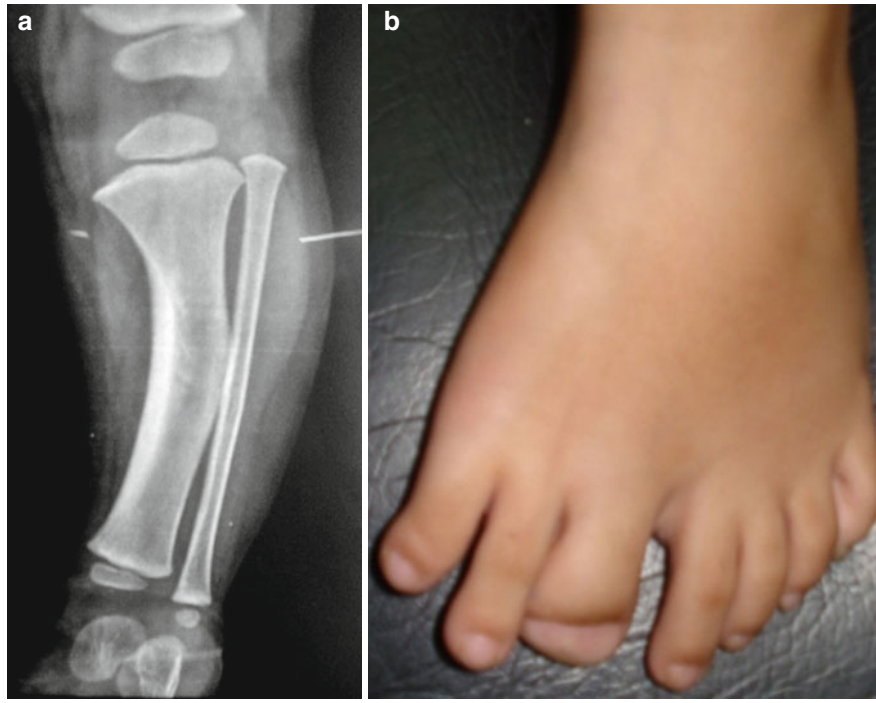

Fig. 4.7 Congenital anterolateral bowing of the tibia (**a**) with duplication of the hallux (**b**); the child also has preaxial mirror polydactyly (Courtesy of Dr. Ranjit Deshmukh, Pune, India)

Congenital Anterolateral Bowing of the Tibia with Associated Duplication of the Hallux

This is a rare deformity that resembles anterolateral bowing associated with congenital pseudarthrosis of the tibia (Tuncay et al. 1994; Bressers and Castelein 2001) (Fig. 4.7a, b). Partial or complete spontaneous resolution of the deformity is the norm. Surgery will be needed to deal with the anomalous great toe. Surgery may also be needed at a later date if the tibial deformity does not resolve completely.

There is often a suggestion of incomplete tibial duplication in the mid-shaft which is the cause of the bowing of the tibia. Occasionally two distinct cortices and separate medullary canals may be seen; this regresses over time and a single medullary cavity eventually develops.

Campomelic Dwarfism

The features of this syndrome include dwarfism, craniofacial abnormalities, shortening of upper and lower limbs with bowing of the femur and tibia, and scoliosis (Kozlowski et al. 1978). There may be associated DDH, equinovarus, or calcaneovalgus deformities of the feet.

Characteristically cutaneous dimples are seen over the summit of the femoral and tibial bow.

The Delta Tibia

All the cases described in the literature have been unilateral with a predilection for the left side of male infants. The bow is characteristically anterolateral and is associated with shortening of the affected tibia (Fig. 4.8). The fibula is totally straight and longer than the tibia resulting in disruption of the proximal tibiofibular joint and proximal migration of the fibular head. Duplication of toes is often seen in this condition; preaxial duplication is more common than postaxial duplication. Polysyndactyly of the hands may also occur (Currarino et al. 2003).

The changes noted in the lateral radiograph is diagnostic; the tibia appears to be divided into a proximal and distal segment, both of which taper towards the apex of the bow that is in the mid-shaft or at the junction of the middle and lower thirds. A triangular piece of bone which lacks a medullary cavity is seen in the concavity of the bow often separated from the tibial segments by a

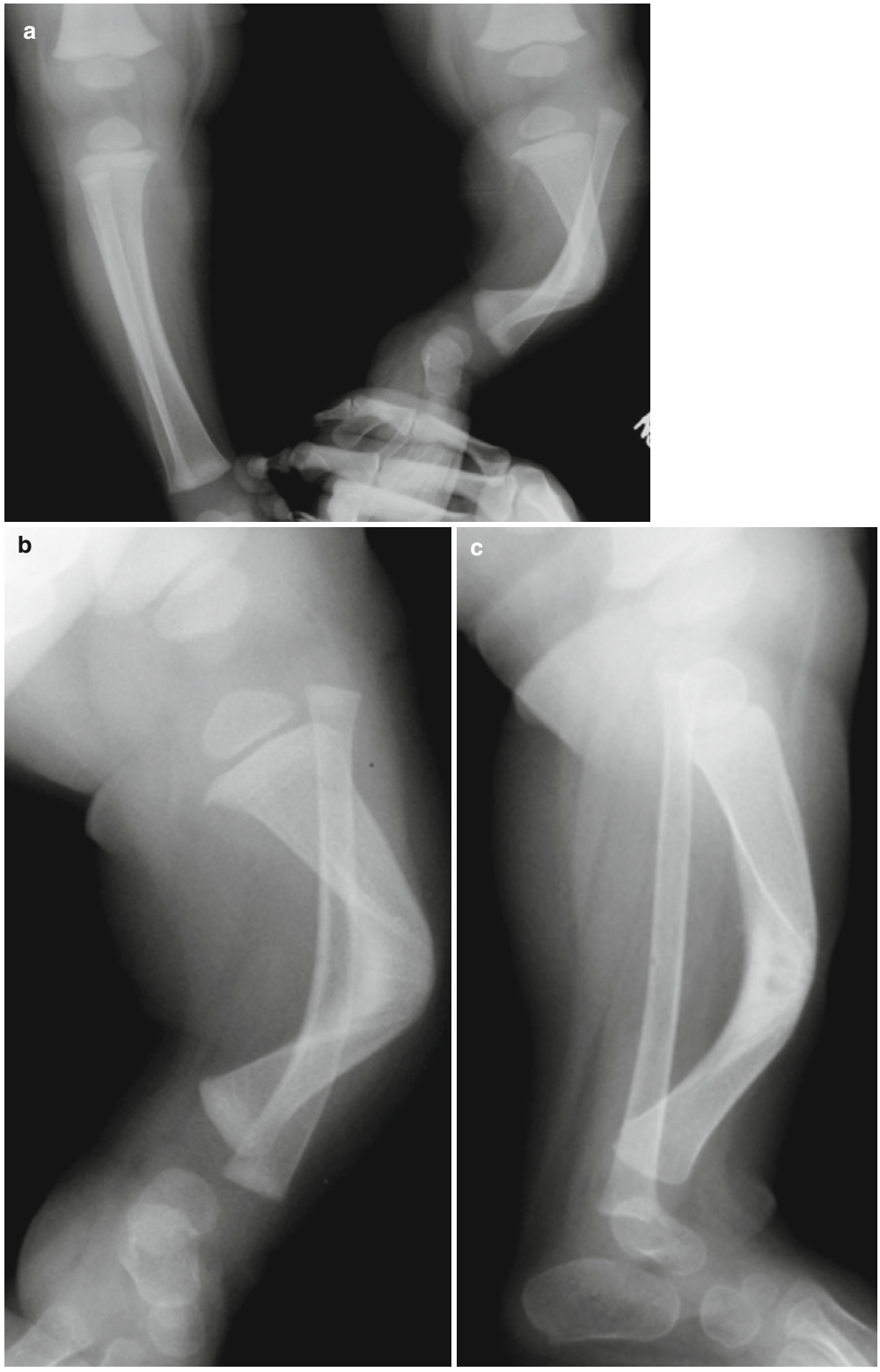

Fig. 4.8 An example of delta tibia in an infant (**a**); the extent of spontaneous resolution of the deformity over 1 year is evident (**b**, **c**). The characteristic pattern of tapering segments of the tibia and the triangular fragment filling the concavity of the bow are seen in the lateral radiograph of an older child (**d**). The deformity is virtually resolved by the age of 9 years though the shortening persists (**e**) (Courtesy: Dr. Charles Johnston, Dallas, USA)

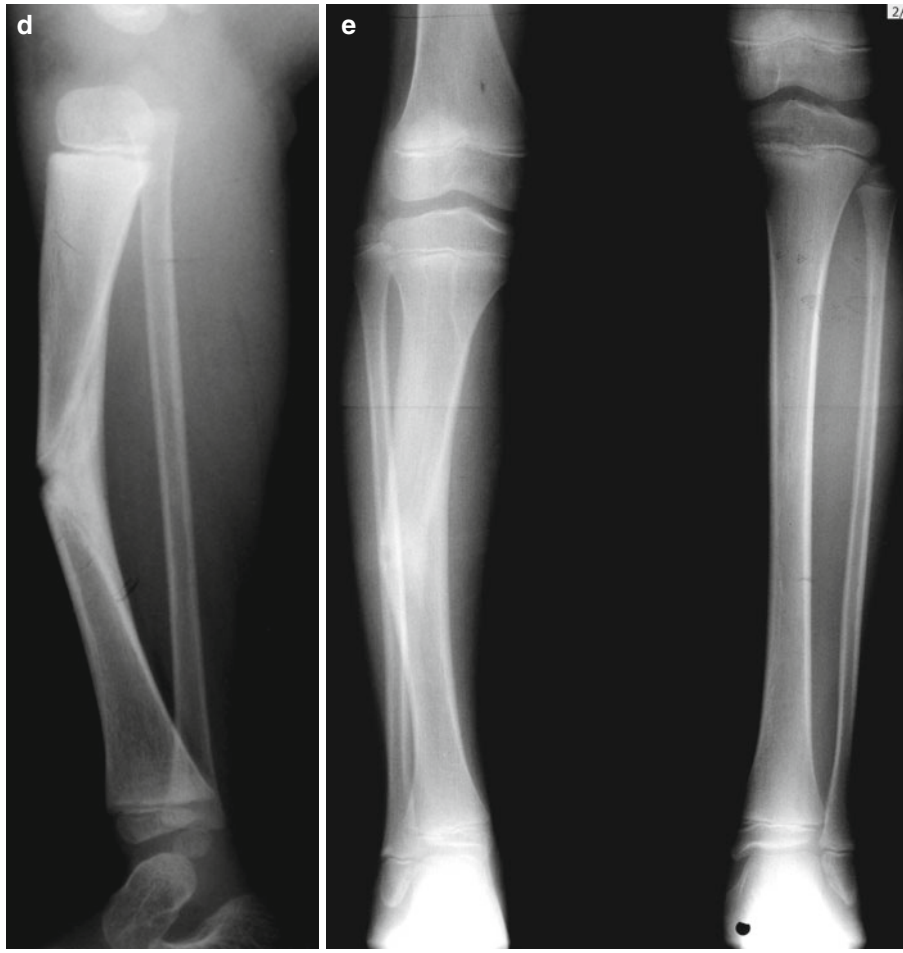

Fig. 4.8 (continued)

translucent line. Spontaneous resolution of the deformity invariably occurs.

Infantile Cortical Hyperostosis (Caffey's Disease)

Though typically this occurs in infants under the age of 6 months, in a small proportion of instances the child may be born with the disease (Kitchin 1951) (Fig. 4.9). There may be malaise and low-grade fever, local warmth, and mild tenderness. The disease is self-limiting with resolution of the swelling and bowing occurring over time.

Weismann-Netter-Stuhl Syndrome

The syndrome is a familial autosomal dominant dysplasia which was originally described as congenital non-progressive anterior bowing of the tibia and fibula. It is now recognized that bowing of the femur is also an important feature of the condition. In addition, there may be short stature, delayed walking, kyphoscoliosis, facial dysmorphism, and some shortening of the upper limbs (Tieder et al. 1995).

4.5 Establishing the Diagnosis

An outline of the process of establishing a diagnosis of the cause of a bowed tibia at birth is shown in Table 4.1.

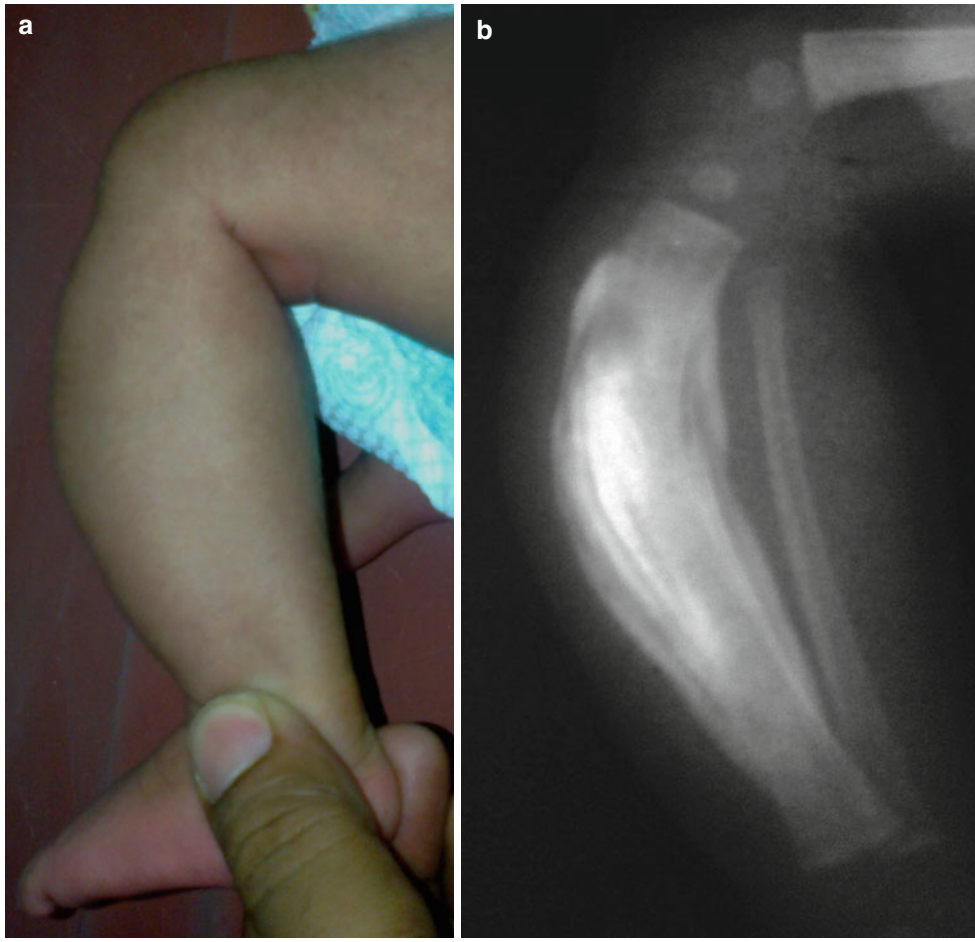

Fig. 4.9 Anterior bowing of the tibia seen in a newborn (**a**); the leg was warm to touch and slightly tender. The radiograph shows florid periosteal reaction with an onion-peel pattern that is characteristic of Caffey's disease (**b**) (Courtesy: Dr. Pabitra Sahoo, Cuttack, India)

Table 4.1 Outline of the diagnosis of the cause of a bowed tibia at birth

History		
Family history of similar deformity may rarely be present	Family history of neurofibromatosis may be present	No positive family history
Physical examination		
Anteromedial bow	Anterolateral bow	Posteromedial bow
Equinovalgus, equinus, or equinovarus deformity may be present	No foot deformity	Calcaneovalgus deformity
Variable degree of shortening of the tibia	No shortening of the tibia	Shortening of the tibia proportionate to the severity of bowing
Mild degree of shortening of the thigh often present. Less frequently severe shortening of the thigh may be present	No shortening of the thigh	No shortening of the thigh
Associated anomalies of the opposite lower limb or the upper limbs may be present	No other anomalies	No other anomalies
No café au lait spots	Café au lait spots may be present	No café au lait spots
Working diagnosis: Fibular hemimelia	Working diagnosis: Congenital pseudarthrosis of the tibia	Working diagnosis: Congenital posteromedial bowing of the tibia
Investigations		
Plain radiograph: Fibula totally or partially absent Tibia bowed anteromedially; medullary cavity and cortex normal Massive talocalcaneal coalition may be present but may only show up once the child is older	Plain radiograph: Tibia bowed anterolaterally; the medullary cavity at the site of the bow may be partially or totally obliterated Very rarely, in the newborn there may be a fracture at the site of the bow The fibula may be normal or bowed and show changes similar to the tibia The bones of the foot are normal	Plain radiograph: Tibia bowed posteromedially at the junction of the middle and lower third Medullary cavity not obliterated but may be narrow The tibia is shorter than the opposite tibia, and the degree of shortening is related to the severity of bowing Fibula may be bowed The bones of the foot are normal
Diagnosis		
Fibular hemimelia	Congenital pseudarthrosis of the tibia	Congenital posteromedial bowing of the tibia

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The Inverted Foot

5

Benjamin Joseph

5.1 Introduction

The most common congenital deformity of the foot requiring early treatment is clubfoot – the inverted and plantarflexed foot. Though the diagnosis of clubfoot per se is straightforward and obvious, the underlying causes of the deformity are many (Fig. 5.1), and the prognosis and treatment of clubfeet of different etiologies may vary a great deal. It is for this reason that an attempt should be made to determine the underlying cause. On one end of the spectrum is the foot that is mildly inverted on account of intrauterine molding – the postural clubfoot that is completely amenable to nonoperative treatment. On the other end is the extremely rigid clubfoot associated with multiple congenital contractures or the syndromic clubfoot that may on occasion stubbornly defy the most radical treatment.

In the vast majority of instances, clubfoot occurs as an isolated anomaly with no apparent underlying cause; this is the idiopathic clubfoot. The second group of clubfeet is associated with some neurological abnormality – the neurogenic clubfoot. Clubfeet may be seen in association with lower limb deficiencies, and the rarer forms of clubfeet include clubfoot associated with syndromes and skeletal dysplasias.

5.2 Questions to Establish a Diagnosis

- **Is there a history suggestive of intrauterine crowding?**
- **Is there a family history of clubfoot?**
- **Is the spine clinically normal?**
- **Are there symmetric deformities of the upper and/or lower limbs?**
- **Are there other deformities of the proximal joints of the limb?**
- **Is the range of flexion of the knee normal?**
- **Is the number of toes of the foot normal?**
- **Are there constriction bands in the upper or lower limbs?**
- **Are there dysmorphic features or dwarfism?**
- **How supple or rigid is the deformity of the affected foot?**

Is there a history suggestive of intrauterine crowding?

When there is decreased space in the uterine cavity on account of twin pregnancy, uterine fibroids or due to congenital anomalies of the uterus postural clubfoot may occur.

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_5

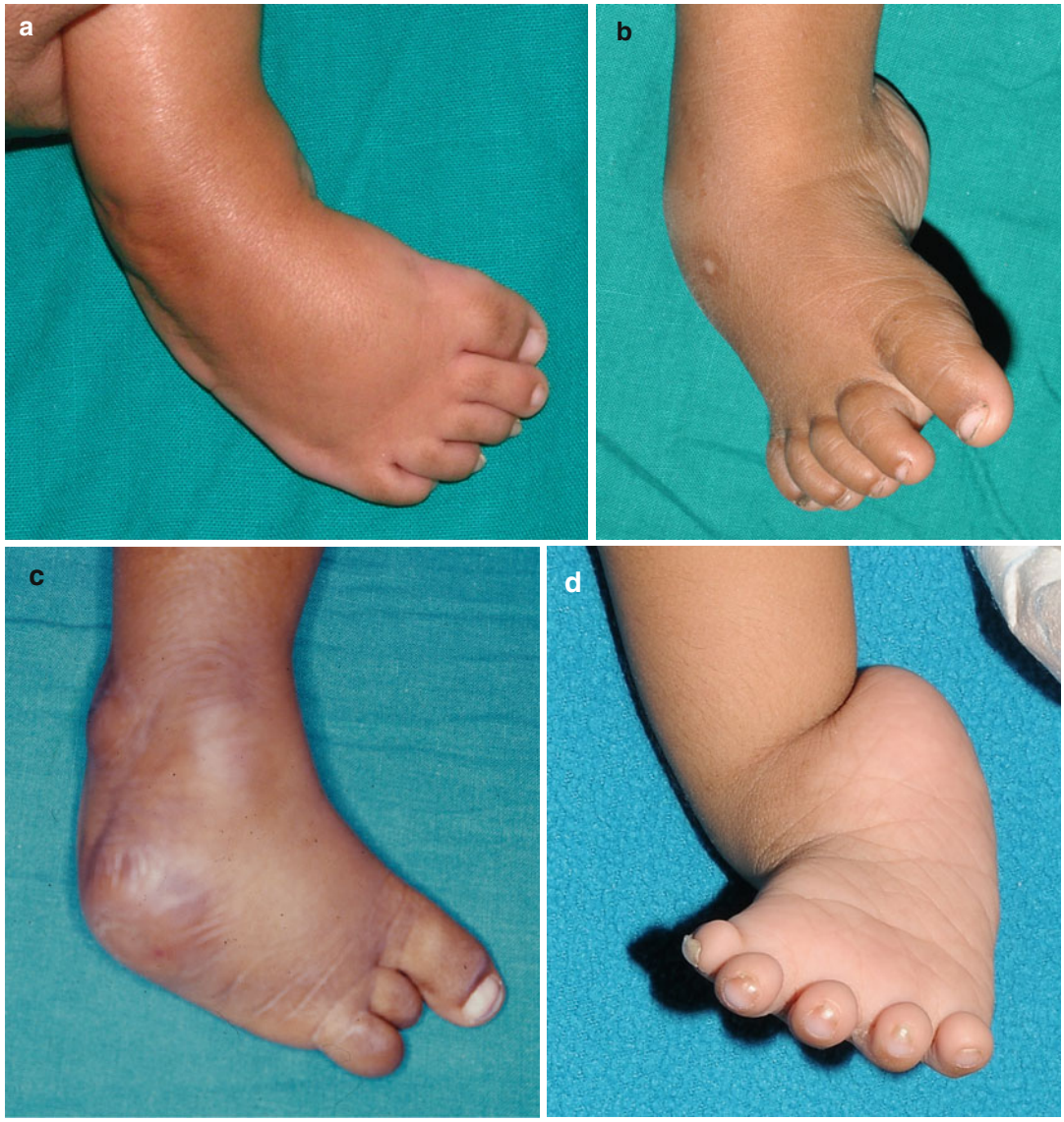

Fig. 5.1 Examples of a postural clubfoot (**a**), a rigid idiopathic clubfoot (**b**), clubfoot associated with fibular aplasia and loss of postaxial rays of the foot (**c**), and clubfoot associated with tibial aplasia with a very hypoplastic great toe (**d**)

Is there a family history of clubfoot?

A family history of clubfoot may be forthcoming in idiopathic clubfoot and clubfoot associated with syndromes or skeletal dysplasia. A positive family history is usually lacking in babies with postural clubfoot.

Is the spine clinically normal?

Clubfoot may be associated with spina bifida and with sacral agenesis. While spina bifida cystica and complete sacral agenesis can be diagnosed on clinical examination, minor degrees of spinal dysraphism or partial sacral agenesis are difficult to diagnose without MRI scans.

Are there symmetric deformities of the upper and/or lower limbs?

Symmetrical deformities of the upper and lower limbs are characteristically seen in multiple congenital contractures or arthrogryposis.

Are there other deformities of the proximal joints of the limb?

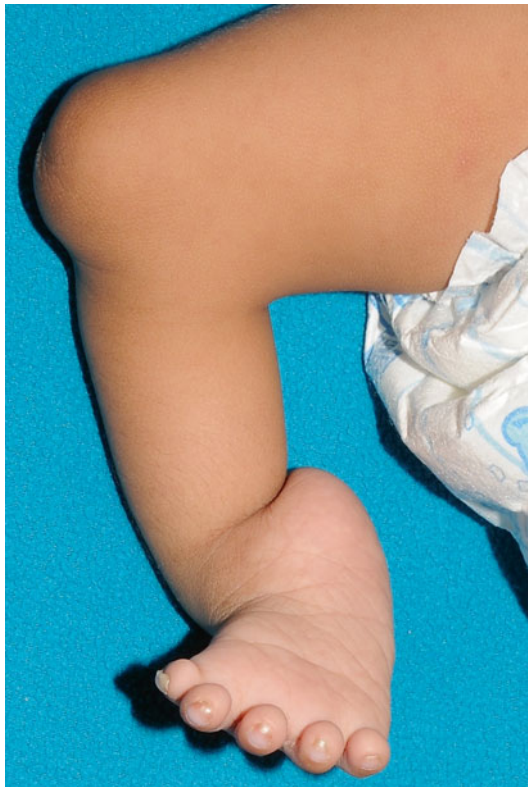

Fig. 5.2 Flexion deformity of the knee and equinovarus deformity of the foot in a child with complete tibial aplasia

Flexion and abduction deformities of the hips may be seen in complete sacral agenesis, and flexion deformities of the knee may be present in sacral agenesis, spina bifida, multiple congenital contractures, and complete tibial aplasia (Fig. 5.2).

Is the range of flexion of the knee normal?

Congenital quadriceps contracture and congenital knee dislocation may be associated with clubfoot. Milder degrees of quadriceps contracture can only be detected by testing passive flexion of the knee (Fig. 5.3). If the knee does not flex up to 90°, casting for clubfoot will be difficult. Knee flexion should be restored early to facilitate expeditious treatment of clubfoot.

Is the number of toes of the foot normal?

Equinovarus deformity in children with agenesis of the tibia may often be associated with either loss of medial (preaxial) rays of the foot or less commonly with preaxial polydactyly. Clubfoot associated with fibular agenesis is infrequent; these children often have absence of lateral (postaxial) rays (Fig. 5.1c).

Are there constriction bands in the upper or lower limbs?

Amniotic band syndrome or congenital constriction band syndrome may be associated with

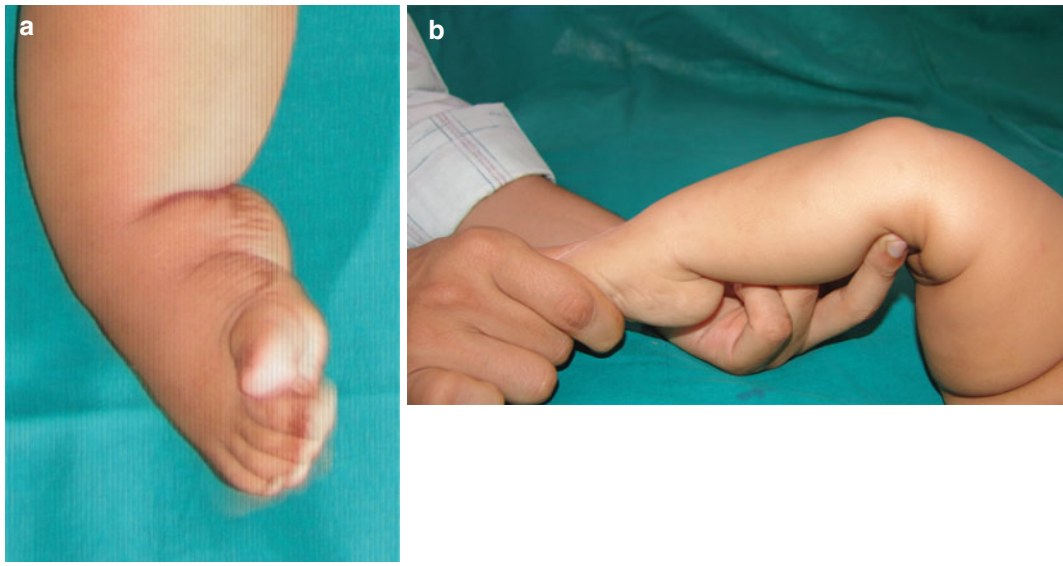

Fig. 5.3 Congenital clubfoot (**a**) in an infant with limited knee flexion due to a congenital quadriceps contracture (**b**)

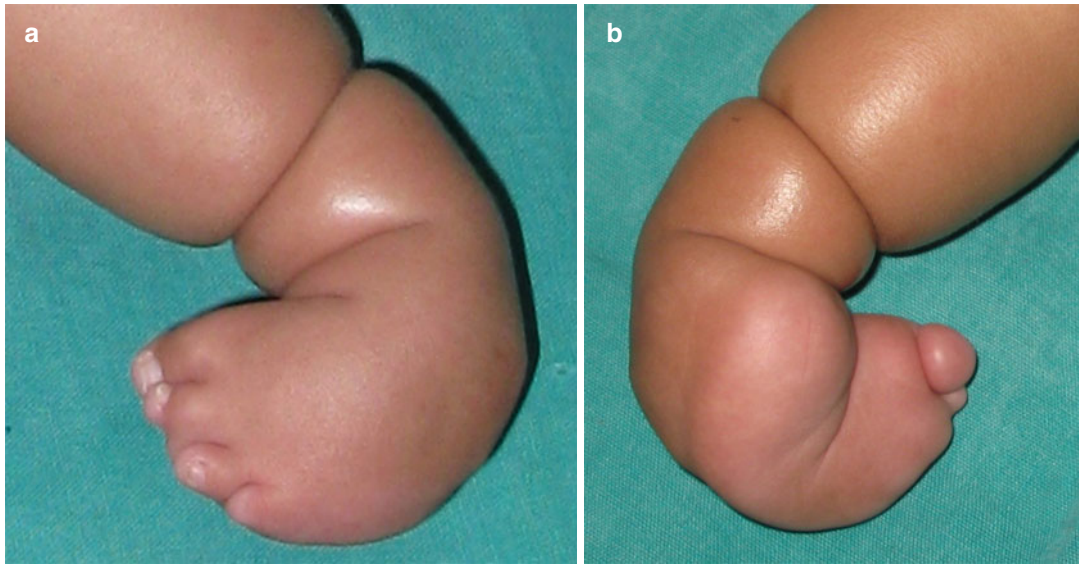

Fig. 5.4 Front (**a**) and back (**b**) of the foot and ankle of an infant with clubfoot associated with amniotic band syndrome. The proximal constriction band is complete and encircles the limb

clubfoot (Fig. 5.4). In a proportion of these children, there is a neurogenic component to the clubfoot deformity if the peroneal nerve has been damaged by the constriction band (Allington et al. 1995; Gomez 1996; Hennigan and Kuo 2000).

Are there dysmorphic features or dwarfism?

A condition associated with a particularly rigid form of clubfoot is diastrophic dwarfism. This form of skeletal dysplasia is characterized by severe dwarfism with anomalies of the outer ear (cauliflower ears), an abducted thumb (hitchhiker's thumb), and deformities of the feet.

How supple or rigid is the deformity of the affected foot?

An attempt at gentle passive correction of the deformity of the foot will give valuable insight into the probable underlying pathology. A foot that can be almost completely corrected passively on the very first examination is in all likelihood a postural clubfoot. On the other hand, if none of the deformities can even be partially corrected, it may indicate that there are bony anomalies like tarsal coalition.

5.3 Physical Examination

5.3.1 Look

Look for dysmorphic features and dwarfism. Examine the spine for anomalies, especially the lumbosacral spine. Look for deformities of the hip and knee on the same side and for deformities or deficiencies of the opposite lower limb and the upper limbs.

Inspect the foot for presence of deep creases on the medial aspect of the foot and the back of the heel or a short first ray, all of which are features of a rigid foot. Note the number of rays of the foot and note if they are all well developed and of normal relative lengths.

5.3.2 Feel

Palpate the lumbosacral spine to exclude spina bifida and sacral agenesis. Note the distance between the posterior superior iliac spines; this distance is markedly reduced in complete sacral agenesis. Palpate the head of the fibula and the lateral malleolus and tibia throughout its length to

verify if they are developed. Feel the heel to see if the calcaneus is palpable in the heel. If it is palpable, it implies that there is negligible equinus as in a postural clubfoot. In the more severe degrees of clubfoot, the heel feels empty. Palpate the lateral part of the head of the talus anterior to the lateral malleolus.

5.3.3 Move

Attempt to correct the deformity by passively everting and dorsiflexing the foot. This should be done gently without causing any discomfort to the baby. The degree of correction of each of the individual deformities of clubfoot (hindfoot equinus, hindfoot varus, medial rotation of the foot around the talus, and forefoot adduction) should be assessed and documented.

Special Tests
Simple tests to assess the integrity of the peripheral nervous system should be performed to ensure that there is no neurogenic cause for the clubfoot.

The Plantar Grasp
The test: *Stroking the sole causes the toes to flex or curl*.

The reflex will be absent if the toes are paralyzed.

The Babinski Sign
The Babinski sign is usually positive in the normal newborn. Dorsiflexion of the great toe and fanning of the lesser toes will not be noted if the lower limb is paralyzed.

5.4 Assessment of Severity of the Deformity

Most surgeons would attempt to assess the severity of the deformity either to plan treatment or to document the response to treatment. Several classification systems have been devised to assess the extent of the deformity (Catterall 1991; Dimeglio et al. 1995; Harrold and Walker 1983; Hennigan and Kuo 2000), and studies have demonstrated that the efficacy and reproducibility of some of these classification systems are not entirely satisfactory (Shaheen et al. 2012; Dyer and Davis 2006; Flynn et al. 1998; Fopma et al. 2003). The classifications of Dimeglio et al. (1995) and Pirani (Shaheen et al. 2012) are the ones most widely used (Figs. 5.5 and 5.6).

The Dimeglio Classification
Dimeglio et al. evaluate equinus, hindfoot varus, forefoot adduction, and rotation of the foot around the talus. Passive correction of each deformity is attempted, and the angle of deformity is then measured and scored as shown in Fig. 5.5. The points for the four deformities and additional points for presence of medial or posterior creases and calf wasting are summated to arrive at a final score. Based on the score, the foot may be graded as one of four possible grades of severity (Table 5.1).

The Pirani Score
In the Pirani grading system, three aspects of the deformity of the hindfoot and three features of the midfoot are scored as 0 (if normal), 0.5 (if moderate), or 1 (if severe). These individual scores are added to give the final score (0–6). As in the Dimeglio system, the lower the score, the milder the deformity. The Pirani score is easier to apply than the Dimeglio score.

5.5 Investigations to Confirm the Diagnosis

Radiography
Though radiographs are not generally required to make a diagnosis of clubfoot, they can provide useful information about the alignment of the tarsals of the hindfoot, provided the radiographs are taken in a standardized manner (Ritchie and Keim 1964; Herbsthofer et al. 1998; Munshi et al. 2006; Fridman and de Almeida Fialho 2007). The talocalcaneal angles which measure the posture of the subtalar joint are reduced in both the anteroposterior and the lateral views in clubfeet, and the angles improve

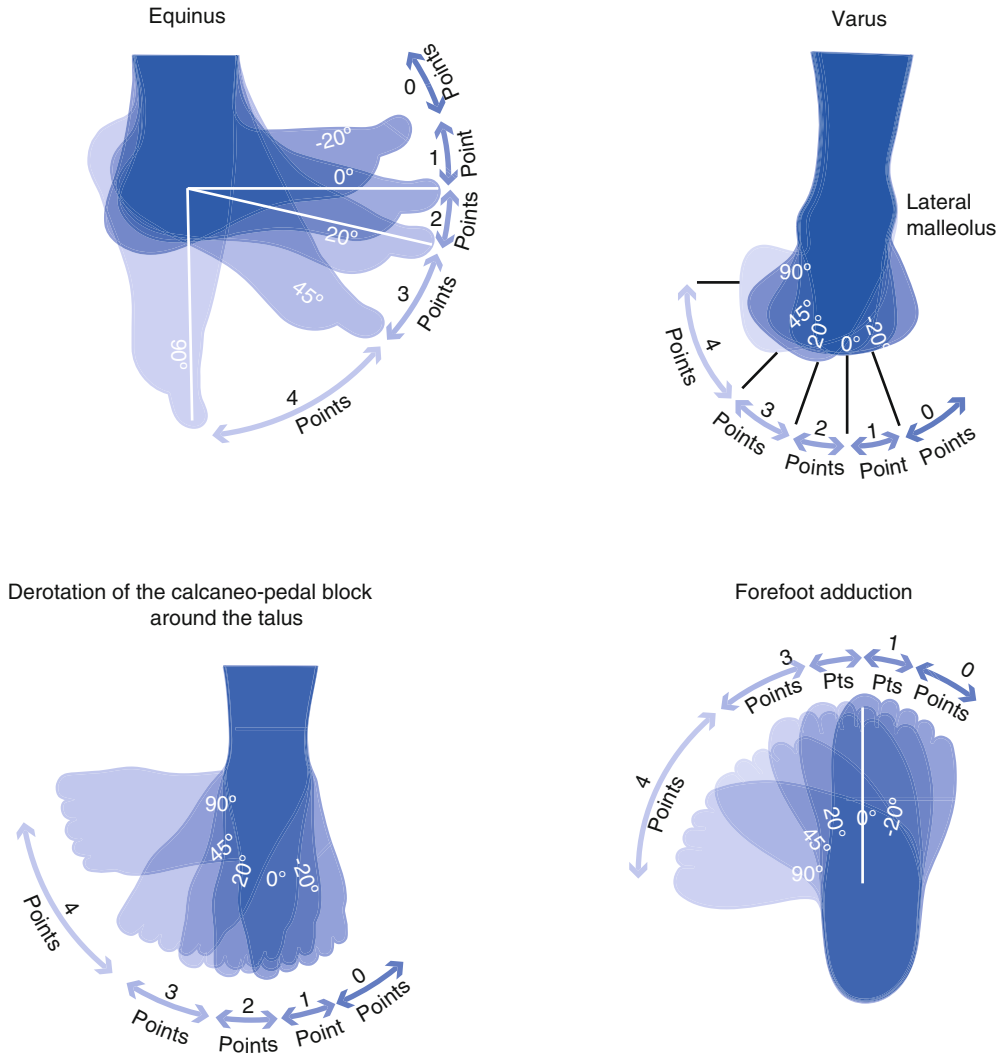

Fig. 5.5 Diagrammatic representation of the Dimeglio classification of clubfoot modified from Dimeglio et al. (1995). An attempt is made to passively correct each of the four deformities defined by the authors, and they are scored as shown. Four additional points are scored for the presence of a medial or posterior crease and wasting of the calf. The sum of all the scores is the final score that categorizes the foot into one of four grades of severity – the lower the grade, the more supple the foot

as the hindfoot varus is corrected. The angle of calcaneal dorsiflexion in a stress dorsiflexion lateral view is a good indicator of the severity of equinus (Fridman and de Almeida Fialho 2007; Munshi et al. 2006). If the foot is very rigid and does not appear to correct at all on gentle manipulation, radiographs of the feet are mandatory to help exclude massive talocalcaneal coalition which may sometimes be associated with what appears to be idiopathic clubfoot (Spero et al. 1994; Rao and Joseph 1994; Callahan 1980). Not all coalitions will be evident on radiographs in the infant. Tarsal coalition associated with clubfoot may be part of a multiple synostosis syndrome (Rebello and Joseph 2003). Radiographs are also essential if the lateral rays of the foot are missing with features of fibular aplasia as massive talocalcaneal coalition is frequently associated with this form of limb deficiency.

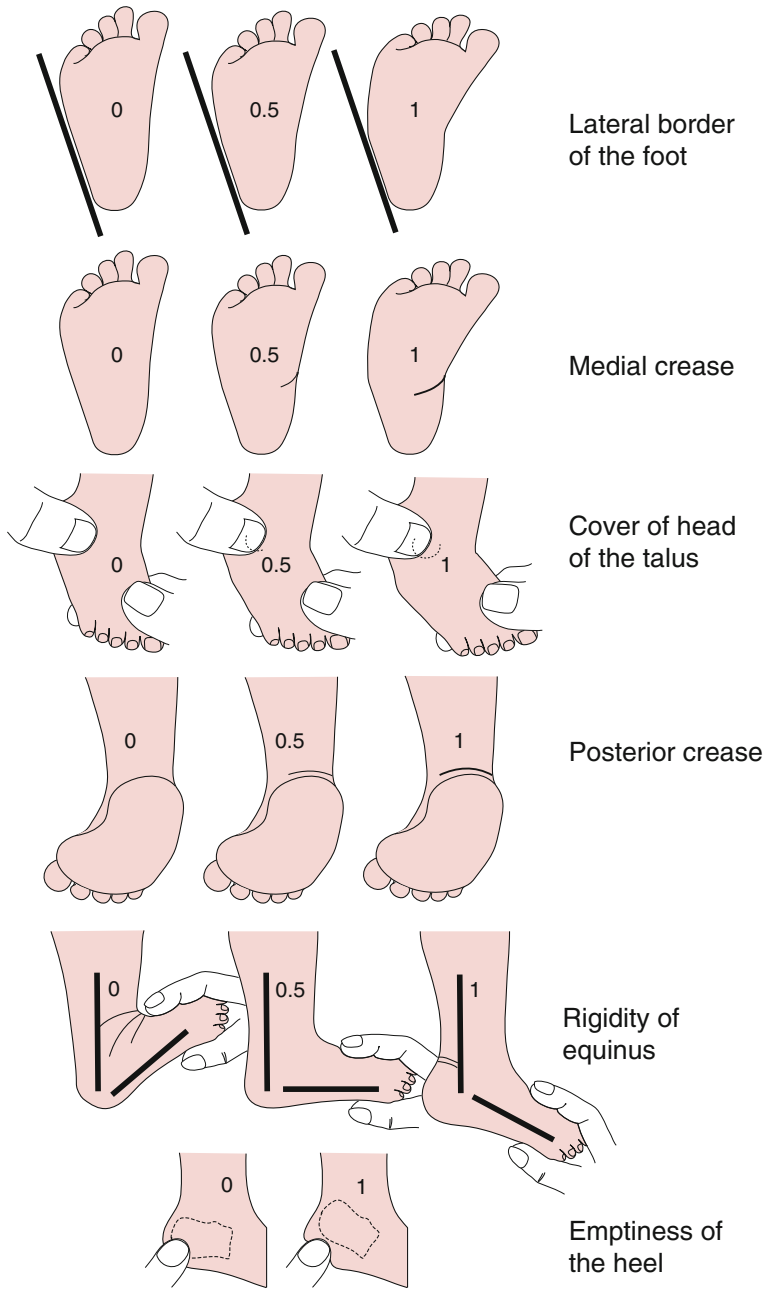

Fig. 5.6 Diagrammatic representation of the Pirani scoring system. The heel crease, the severity of equinus, and the fullness or emptiness of the heel are assessed in the hindfoot, while the curve of the lateral border of the foot, the crease on the medial aspect of the foot, and the extent of uncovering of the head of the talus onto the dorsolateral aspect of the foot are assessed in the midfoot (Fig. 5.6)

Ultrasound

Some surgeons use ultrasound to quantify the severity of the deformity and to monitor response to nonoperative treatment (Coley et al. 2007; Shiels et al. 2007). One measurement that is particularly useful is the distance between the medial malleolus and the tuberosity of the navicular bone; the less this distance, the more severe the deformity.

Table 5.1 Dimeglio classification of congenital clubfoot

Score	Grade	Severity	Suppleness
<5	I	Mild	Over 90 % reducible Soft-soft foot
5–9	II	Moderate	50–90 % reducible Soft-stiff foot
10–14	III	Severe	10–50 % reducible Stiff-soft foot
>15	IV	Very severe	Less than 10 % reducible Stiff-stiff foot

5.6 Differential Diagnosis

Postural Clubfoot

Postural clubfoot develops on account of intrauterine positioning, and this is often due to crowding in utero. The deformity is supple and responds to simple stretching in many instances and invariably to a few casts after serial manipulation. The long-term outcome is excellent.

Idiopathic Clubfoot

The vast majority of clubfeet (~90 %) fall into this category, and they are not associated with any demonstrable neurological abnormality, syndromes, or limb deficiencies. The majority respond to standard methods of serial manipulation and casting. A proportion will need subsequent surgery for relapse, often a tendon transfer.

Idiopathic Complex Clubfoot

It is important to recognize a small proportion of idiopathic clubfeet that are very rigid and difficult to treat. Ponseti et al. refer to them as "complex idiopathic clubfeet" (Ponseti et al. 2006). They are characterized by severe equinus and varus, a short gastroc-soleus muscle belly, deep creases on the sole of the foot and the heel, markedly plantarflexed metatarsals, and extreme rigidity. These feet need to be identified at the outset as the standard method of treatment often fails (Ponseti et al. 2006).

Clubfoot in Multiple Congenital Contractures (Arthrogryposis)

The underlying problem in multiple congenital contractures (comprising over 140 entities) is often the depletion of anterior horn cells, and consequently, there is a neurological basis for deformities in this group of conditions. However, this form of clubfoot is considered separately as it differs from other forms of neurogenic clubfoot. The clubfeet tend to be very rigid, and flexion or extension deformities of the knees, elbows, and wrists will be evident in children with multiple congenital contractures (Fig. 5.7). In one form of multiple congenital contractures, only the hands and feet are affected; this is referred to as distal arthrogryposis.

Neurogenic Clubfoot

Irrespective of the nature of the precise neurological condition, the basic cause in all neurogenic clubfeet is an imbalance between the muscles acting across the axes of the ankle and subtalar joints (Song et al. 2008). The plantarflexors and invertors are stronger than the dorsiflexors and evertors on account of paresis or complete paralysis of the latter groups (Fig. 5.8). There is an associated sensory loss in the foot in spina bifida and peripheral nerve damage, and neuropathic ulcers can develop if adequate care is not taken during casting.

Clubfoot of Limb Deficiency

Clubfoot is more frequently encountered if there is aplasia of the tibia (Fig. 5.9) than if the fibula has failed to develop. Characteristically the leg is shorter than the unaffected side if the deficiency is unilateral. Tarsal coalition is frequently seen in children with fibular aplasia, and in some instances, this may preclude nonoperative treatment of the deformity.

Syndromic Clubfoot and Clubfoot Associated with Skeletal Dysplasias

Equinovarus deformity has been reported in several syndromes such as Freeman-Sheldon syndrome, Larsen syndrome, nail-patella syndrome, Gordon syndrome (Gordon et al. 1969),

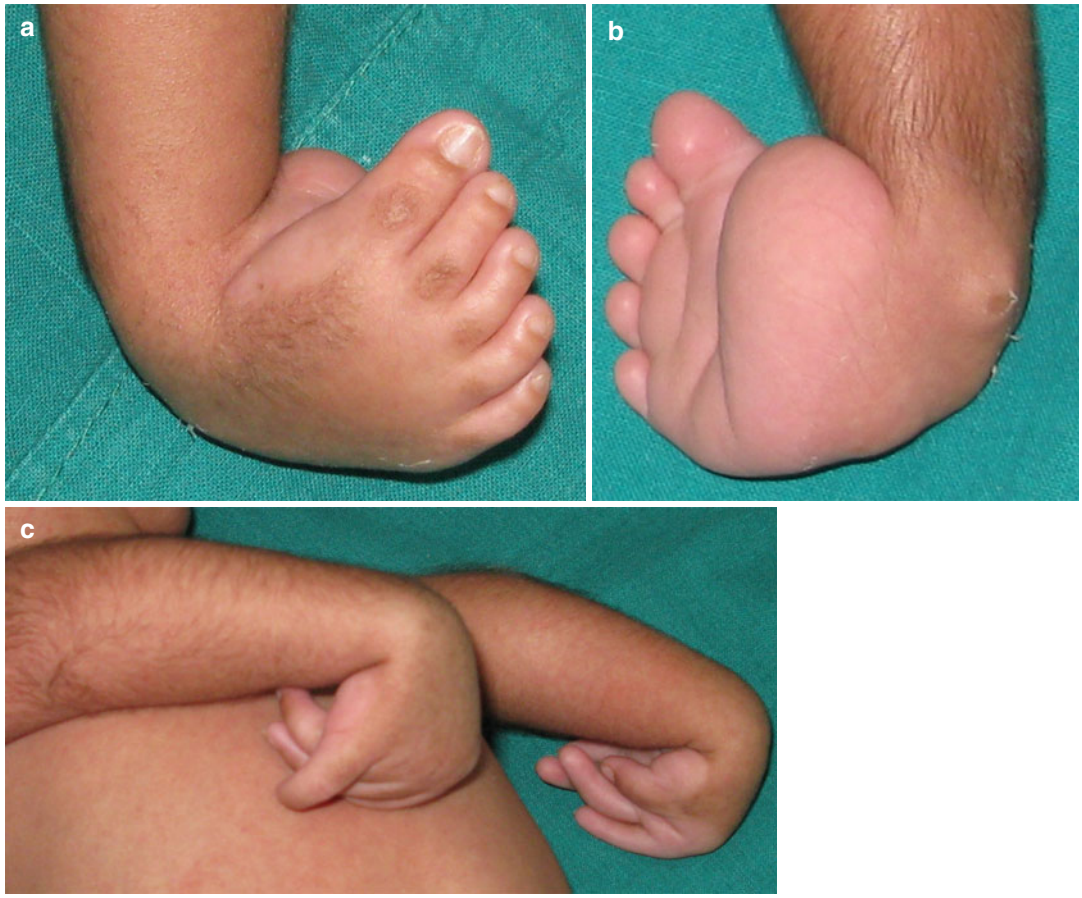

Fig. 5.7 Clubfoot (**a**, **b**) associated with flexion deformities of the wrists (**c**) in a newborn with multiple congenital contractures; the clubfoot deformity is severe

prune-belly syndrome, and Möbius syndrome. A varus deformity of the hindfoot has been observed in the Nievergelt-Pearlman type of multiple synostosis syndrome (Rebello and Joseph 2003). The feet behave generally like idiopathic clubfeet in some of these conditions. In Larsen syndrome, the hyperlaxity of ligaments may increase the propensity for overcorrection particularly if surgery is undertaken. In multiple synostosis syndromes, nonoperative measures are ineffective on account of the underlying bony abnormality (Rebello and Joseph 2003).

Clubfoot is seen as part of the spectrum of foot deformities in diastrophic dysplasia; it is not the most frequently encountered deformity (Ryoppy et al. 1992; Weiner et al. 2008). The foot deformities in diastrophic dwarfism are often very rigid and difficult to treat.

The importance of diagnosing a syndrome associated with clubfoot is to enable identification of other anomalies that may require more urgent intervention. Furthermore, the treatment of the clubfoot itself may need to be modified on account of these anomalies.

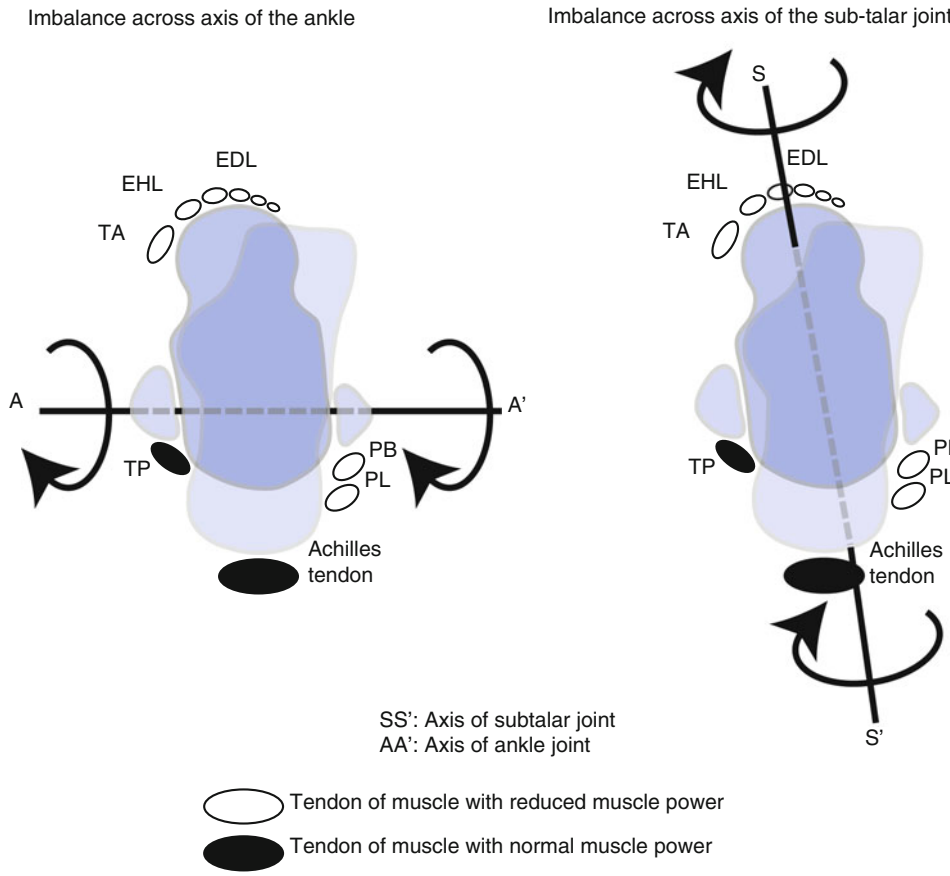

Fig. 5.8 Diagrammatic representation of how muscle imbalance across the axis of the ankle and subtalar joint produce the equinovarus deformity in neurogenic clubfoot

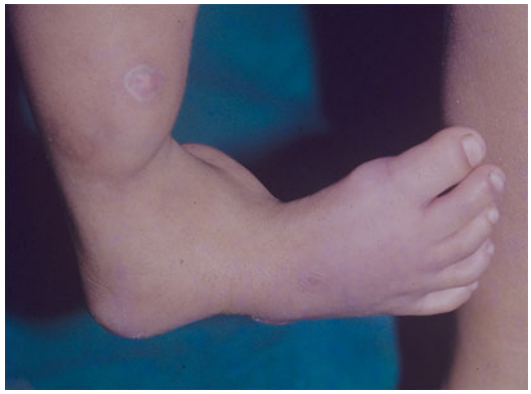

Fig. 5.9 Severe equinovarus deformity in a child with tibial aplasia

5.7 Establishing the Diagnosis

The importance of establishing the etiology of the deformity is because it has a definite bearing on the response to nonoperative measures and the outcome of treatment (Gurnett et al. 2008). An outline of the approach to establishing a diagnosis is shown in Tables 5.2 and 5.3.

Table 5.2 Establishing the diagnosis of the cause of the inverted foot in the newborn

History			
A family history of clubfoot is usually not present	A positive family history may be present	A positive family history may be present	A positive family history may be present
History suggestive of intrauterine crowding may often be present	History of intrauterine crowding seldom present	History of intrauterine crowding seldom present	History of intrauterine crowding seldom present
Physical examination			
Facial dysmorphism absent	Facial dysmorphism absent	Facial dysmorphism absent	May be present
Spine and sacrum normal	Spine and sacrum normal	Spine and sacrum normal	Spina bifida, sacral agenesis, or scoliosis may be present
Symmetric deformities of the upper limbs, knees, or hips not present	Symmetric deformities of the upper limbs, knees, or hips not present	Symmetric deformities of the upper limbs, knees, or hips not present	Symmetric deformities of the upper limbs, knees, or hips may be present
No deformities of the knee or hip of the same side	No deformities of the knee or hip of the same side	No deformities of the knee or hip of the same side	Deformities of the knee or hip of the same side may be present
Normal number of toes and the development of the toes are normal	Normal number of toes and the development of the toes are normal	Normal number of toes. The great toe may be short	There may be a reduction of the number of toes, polydactyly, or varying degrees of hypoplasia of the toes
Shortening of the affected leg is not present	Shortening of the affected leg is not appreciable	Appreciable (very mild) shortening of the leg may be present	Significant shortening of the leg may be present
No muscle weakness or sensory loss	No muscle weakness or sensory loss	No muscle weakness or sensory loss	Muscle weakness or sensory loss may be present
Supple foot	Rigid foot	Very rigid foot	Variable
Dimeglio score: <5	Dimeglio score: 5–15	Dimeglio score: >15	Dimeglio score: 5–20
Working diagnosis: Postural clubfoot	Working diagnosis: Idiopathic rigid clubfoot	Working diagnosis: Complex idiopathic clubfoot	Working diagnosis: Non-idiopathic clubfoot
Investigations			
Not indicated	Not usually indicated	Plain radiograph of the foot to rule out tarsal coalition (may be of limited use in the new born as it may only manifest later)	Plain radiographs of the feet, spine, and pelvis as indicated based on the clinical findings
Diagnosis			
Postural clubfoot	Idiopathic rigid clubfoot	Complex idiopathic clubfoot (if tarsal coalition is absent) Clubfoot associated with tarsal coalition (if coalition seen on radiograph)	Non-idiopathic clubfoot (see Table 5.3)

Table 5.3 Establishing the diagnosis of the cause of non-idiopathic clubfoot in the newborn

History				
		Family history may be present		Family history may be present
Physical examination				
Face and head: Facial dysmorphism may or may not be present	Face and head: No facial dysmorphism Hydrocephalus may be present	Face and head: Facial dysmorphism usually not present	Face and head: Facial dysmorphism not present	Face and head: Facial dysmorphism often present
Stature and body proportions: Normal	Stature and body proportions: Normal	Stature and body proportions: Normal	Stature and body proportions: Normal	Stature and body proportions: Dwarfism frequent Body proportions often altered
Spine and sacrum: Normal	Spine and sacrum: Spina bifida or sacral agenesis	Spine and sacrum Normal	Spine and sacrum normal	Spine and sacrum: Scoliosis may be present
Upper limbs: Symmetric deformities of the shoulders, elbows and wrists present	Upper limbs: Normal	Upper limbs: May have longitudinal or transverse deficiencies	Upper limbs: Syndactyly and varying degrees of hypoplasia of the fingers with constriction bands may be present	Upper limbs: May be short The thumb may be abducted
Knee: Deformities of the same knee often present (flexion or hyperextension)	Knee: Flexion deformity may be present	Knee: Flexion deformity may be present	Knee: Normal	Knee: Often normal Flexion deformity may be present
Leg: Not short	Leg: Not short	Leg: Short	Leg: Normal in length but may have partial/complete constriction band	Leg: May be short
Toes: Normal in number and development	Toes: Normal in number and development	Toes: Preaxial or postaxial rays may be deficient	Toes: Syndactyly and varying degrees of hypoplasia Constriction bands	Toes: May be normal or hypoplastic
Muscle power: Muscle weakness around foot and ankle may be demonstrable	Muscle power: Muscle weakness around foot and ankle present	Muscle power: Muscle weakness may be present (e.g., quadriceps weakness with tibial aplasia)	Muscle power: Often normal Occasionally compromised on foot	Muscle power: Normal
Sensation: No sensory loss on foot	Sensation: Sensory loss on foot may be present	Sensation: Normal	Sensation: Often normal Occasionally compromised on foot	Sensation normal
Suppleness of the clubfoot: Rigid foot	Suppleness of the clubfoot: Rigid foot frequently, sometimes supple	Suppleness of the clubfoot: Rigid foot	Suppleness of the clubfoot: Rigid foot	Suppleness of the clubfoot: Rigid foot frequently

(continued)

Table 5.3 (continued)

Working diagnosis: Arthrogrypotic clubfoot	Working diagnosis: Neurogenic clubfoot	Working diagnosis: Clubfoot associated with limb deficiency	Working diagnosis: Clubfoot in amniotic band syndrome	Working diagnosis: Syndromic clubfoot or clubfoot associated with skeletal dysplasia
Investigations				
Radiographs not indicated	Radiographs of spine and pelvis: May confirm spina bifida or sacral agenesis	Radiographs of the limbs, pelvis, and spine: Will show the nature and extent of congenital malformation	Radiographs seldom required (except when a deep constriction band encircles the calf to see if the tibia is affected)	Skeletal survey required to establish the nature of the dysplasia or syndrome
Other investigations not indicated	MRI of spine/brain may be required at a later date	Ultrasound of the abdomen to exclude visceral anomalies	Other investigations not indicated	Genetic studies may be indicated later
Diagnosis				
Arthrogrypotic clubfoot	Neurogenic clubfoot due to spina bifida/sacral agenesis/peripheral nerve lesion	Clubfoot associated with tibial or fibular deficiency	Clubfoot in amniotic band syndrome	Syndromic clubfoot or clubfoot associated with skeletal dysplasia

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The Everted Foot

6

Benjamin Joseph

6.1 Introduction

The foot in the newborn may appear everted if the hindfoot is in valgus either at the ankle or the subtalar joint or if the forefoot is pronated.

The most frequent reason for the foot being everted at birth is a congenital calcaneovalgus deformity which is a benign condition that resolves spontaneously. In contrast, there are a few conditions where the deformity persists. Eversion of the foot and a valgus deformity of the hindfoot could be due to lack of adequate bony support to the lateral aspect of the ankle, contracture of the evertor muscles or tendons, or imbalance of muscle power between the invertors and evertors of the foot (Fig. 6.1) (Edwards and Menelaus 1987; Yang and Lee 2002; Cho et al. 2006; Lampasi et al. 2008).

6.2 Questions to Establish a Diagnosis

- **Is there a history suggestive of intrauterine crowding?**
- **Is the spine clinically normal?**
- **Are there café au lait spots anywhere on the body?**
- **Are there symmetrical deformities of the upper and/or lower limbs?**
- **Is there a bowing deformity of the leg and shortening?**
- **Can the lateral malleolus and the lower end of the fibula be palpated?**
- **Is the number of toes of the foot normal?**
- **Are the hips stable?**
- **How supple or rigid is the deformity of the affected foot?**

Is there a history suggestive of intrauterine crowding?

As in the case of postural clubfoot, it has been assumed that congenital calcaneovalgus may be associated with situations where there is a packaging effect on account of crowding in utero.

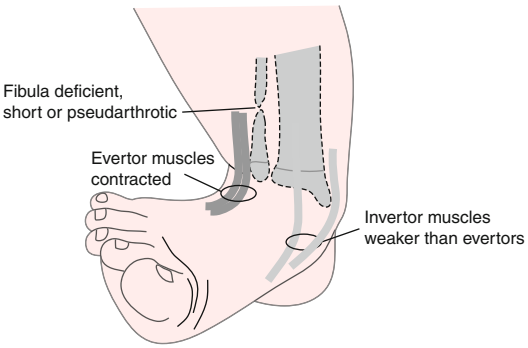

Fig. 6.1 Factors that may be the underlying cause for an everted foot at birth

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_6

However, Wynne-Davies et al. (1982) did not observe an association between hydramnios and oligohydramnios and calcaneovalgus, and similarly an association with twin pregnancies could not be demonstrated in another study (Nunes and Dutra 1986).

Is the spine clinically normal?

Valgus deformity of the foot may be associated with spina bifida and sacral agenesis, and hence it is essential that the lumbosacral spine is examined.

Are there café au lait spots anywhere on the body?

Café au lait spots suggestive of neurofibromatosis type I (NF-1) may be seen in children with fibular pseudarthrosis.

Are there symmetrical deformities of the upper and/or lower limbs?

Valgus, calcaneovalgus, or congenital vertical talus (congenital convex pes valgus) deformities may all be seen as part of symmetrical deformities of the upper and lower limbs in multiple congenital contractures.

Is there a bowing deformity of the leg and shortening?

Congenital posteromedial bowing of the tibia and anteromedial bowing of the tibia seen in fibular aplasia are often associated with a calcaneovalgus or equinovalgus deformity, respectively (Pappas 1984; Shah et al. 2009). In both these conditions, the leg is shorter than the normal side when the anomaly is unilateral.

Can the lateral malleolus and the lower end of the fibula be palpated?

Total absence of the fibula or partial absence involving the distal part of the fibula is often associated with an everted posture of the foot (Fig. 6.2). Similarly fibular pseudarthrosis may be associated with a valgus deformity of the ankle (Yang and Lee 2002; Cho et al. 2006; Lampasi et al. 2008).

Is the number of toes of the foot normal?

Equinovalgus deformity is typically seen in association with fibular aplasia where the lateral rays of the foot may be absent (Fig. 6.2).

Are the hips stable?

An association between developmental dysplasias and congenital calcaneovalgus deformity has been noted in some studies (Paton and Choudry 2009).

How supple or rigid is the deformity of the affected foot?

Idiopathic congenital calcaneovalgus deformity and calcaneovalgus associated with posteromedial bowing of the tibia are supple deformities that can be corrected to a very large extent by passively plantarflexing and inverting the foot. Congenital vertical talus and deformities associated with fibular aplasia or fibular pseudarthrosis are more rigid.

6.3 Physical Examination

6.3.1 Look

Disrobe the baby and carefully look for café au lait spots on the body. Examine the lumbosacral spine and exclude spina bifida and sacral agenesis. Look for symmetrical deformities of the extremities. Look for bowing of the leg and note the plane of bowing if present (see Chap. 4). Look for shortening of the leg and note the number of rays of the foot.

Look at the foot from the front to identify if the forefoot is pronated and from the back to identify the position of the hindfoot, and look at the sole to assess the alignment of the lateral border of the foot.

6.3.2 Feel

Palpate the lumbosacral spine to exclude spina bifida and sacral agenesis. Note the distance between the posterior superior iliac spines; this distance is markedly reduced in complete sacral agenesis. Palpate the fibula throughout its length to verify if it is completely developed and to see if there is a palpable discontinuity in the lower third of the bone. Note the position of

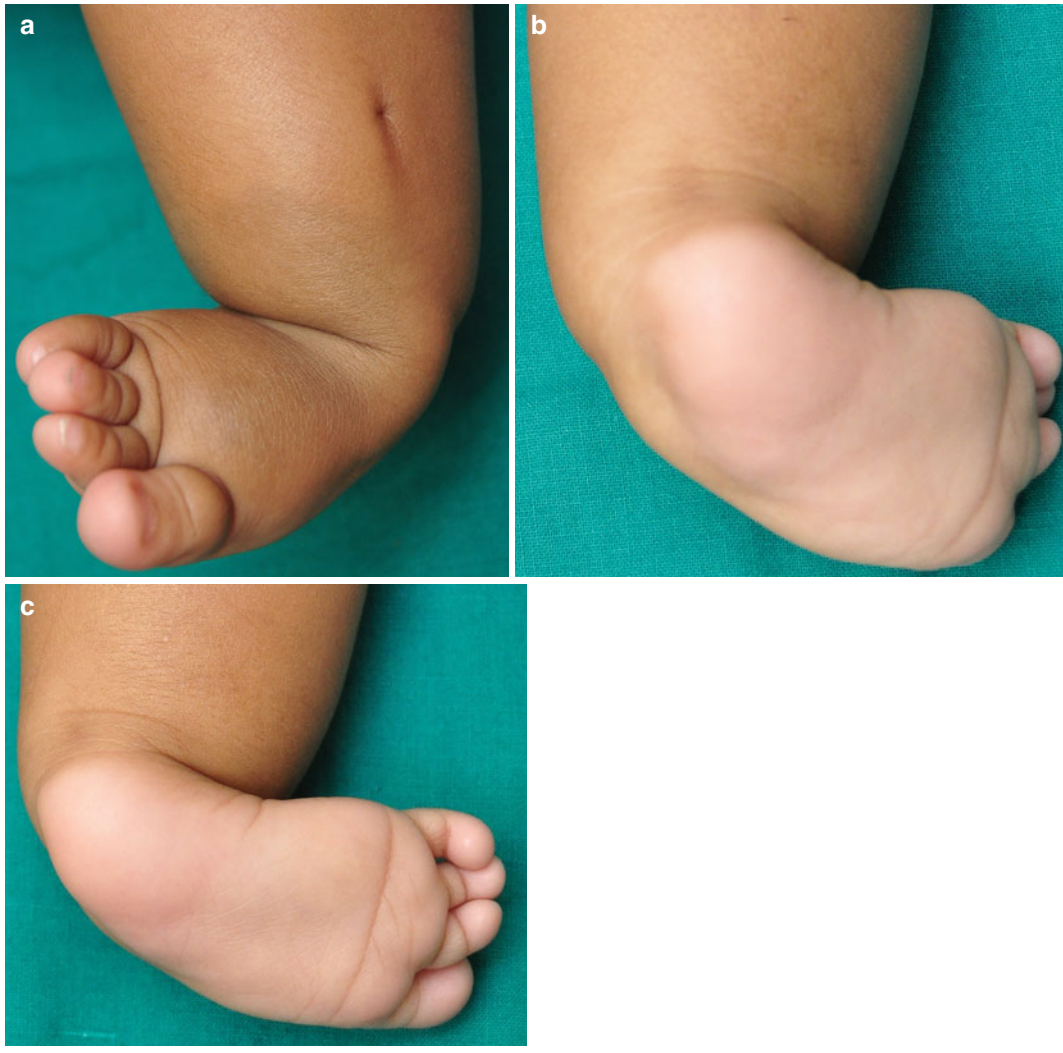

Fig. 6.2 The foot of a newborn infant with an everted foot secondary to fibular aplasia. The pronated forefoot is appreciated in the frontal view (**a**), while the hindfoot valgus is evident when the foot is viewed from the back (**b**). Forefoot abduction is best seen by looking at the lateral border of the sole (**c**). A dimple is often present over the tibia (**a**)

the peroneal tendons in relation to the lateral malleolus.

6.3.3 Move

Perform the Barlow and Ortolani maneuvers to exclude neonatal hip instability (see Chap. 1). Attempt to correct the foot deformity by passively inverting the foot, and assess if the deformity is supple or rigid.

6.3.4 Special Tests

The integrity of the peripheral nervous system should be tested to ensure that there is no neurogenic cause for the deformity of the foot.

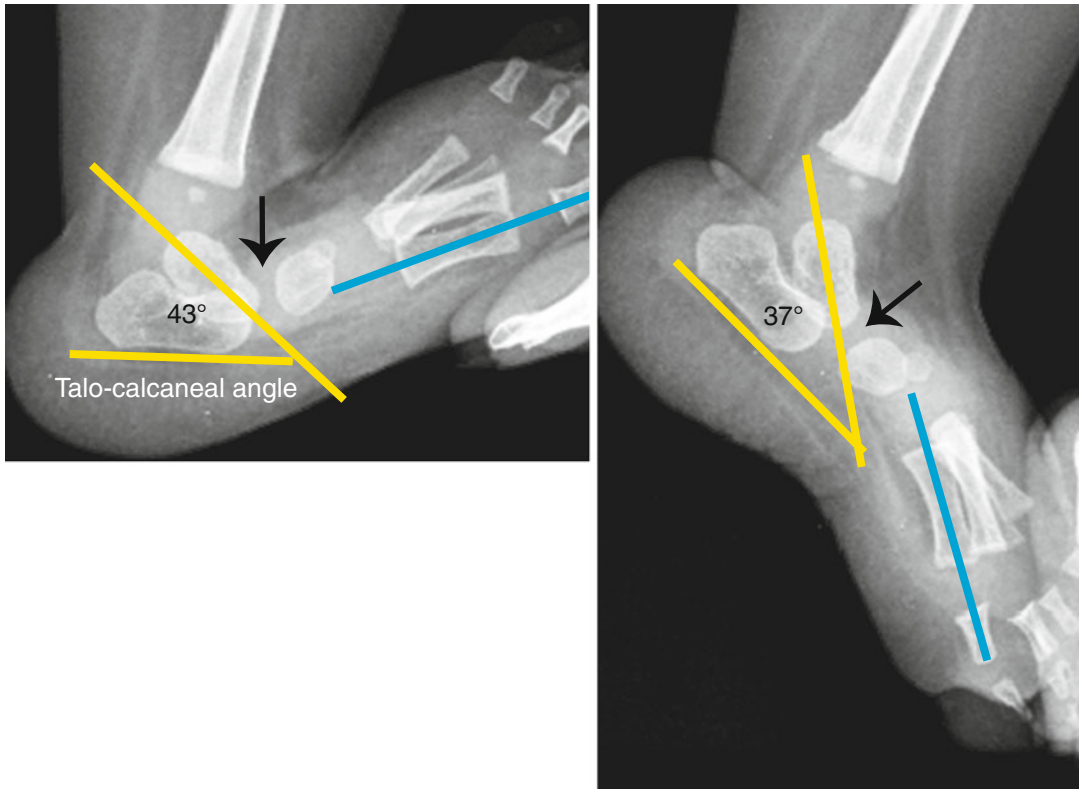

Fig. 6.3 Lateral radiographs of the foot of an infant with an oblique talus. In the dorsiflexion film, the talus appears severely plantarflexed, the midtarsal joint is subluxed (*arrow*), and the axis of the first metatarsal (*blue line*) is not aligned with the long axis of the talus. When the foot is plantarflexed, the talocalcaneal angle between the long axis of the talus and the long axis of the calcaneum (*yellow lines*) reduces, the midtarsal subluxation improves, and the axis of the first metatarsal becomes almost parallel to the talar axis. This pattern of improvement on plantarflexion will not be seen in vertical talus

The Plantar Grasp

The test: *Stroking the sole causes the toes to flex or curl.*

The reflex will be absent if the toes are paralyzed.

The Babinski Sign

The Babinski sign is usually positive in the normal newborn. Dorsiflexion of the great toe and fanning of the lesser toes will not be noted if the lower limb is paralyzed.

6.4 Investigations to Confirm the Diagnosis

Radiography

Plain radiographs are indicated when abnormalities of the fibula are suspected. A lateral radiograph of the foot in plantarflexion is also useful to distinguish between oblique talus and vertical talus. The normal relationship of the tarsal bones is restored on plantarflexion of the foot when there is an oblique talus, but it is not restored in a foot with a vertical talus (Fig. 6.3).

6.5 Differential Diagnosis

Congenital Idiopathic Calcaneovalgus

Idiopathic calcaneovalgus deformity is characterized by dorsiflexion and eversion of the foot. The deformity may appear very severe with the dorsum of the foot lying almost in contact with the shin. On attempting to passively correct the deformity, the foot can almost always be brought to neutral at least. In the vast majority of instances, the deformity resolves spontaneously or with

passive stretching. A small proportion may require a few serial casts with progressive plantarflexion and inversion of the foot. Surgery is only required in the very rare instance of a recalcitrant deformity (Edwards and Menelaus 1987; Yu and Hladik 1994).

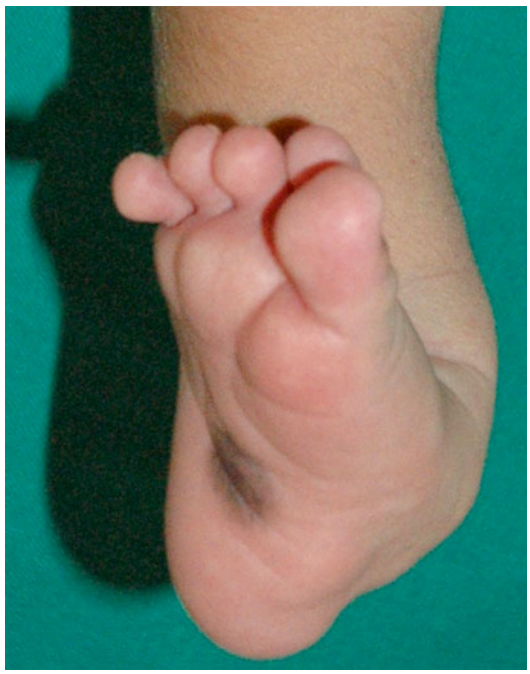

Fig. 6.4 Calcaneovalgus deformity associated with posteromedial bowing of the tibia

Calcaneovalgus Associated with Congenital Posteromedial Bowing of the Tibia

Calcaneovalgus deformity is frequently associated with congenital posteromedial bowing of the tibia (Pappas 1984; Shah et al. 2009). The foot deformity and its response to treatment are identical to isolated congenital idiopathic calcaneovalgus (Fig. 6.4).

Oblique and Vertical Talus

It is important to differentiate congenital calcaneovalgus from congenital convex pes valgus or vertical talus and the less severe oblique talus (Fig. 6.5) (Greenberg 1981; Kumar et al. 1982). Congenital vertical talus is a complex rigid deformity with equinus and valgus of the hindfoot and dorsiflexion, abduction, and pronation of the forefoot in relation to the hindfoot. The gastrocsoleus, ankle, and toe dorsiflexors and the evertors are all contracted in vertical talus; the gastrocsoleus is never contracted in congenital calcaneovalgus. The pronation of the forefoot may not always be appreciable in vertical talus because the tibialis anterior and the peronei are contracted (i.e., tendons on both sides of the axis of the midtarsal joint are contracted).

Multiple Congenital Contractures (Arthrogryposis)

Valgus, calcaneovalgus, or vertical talus may be seen in the newborn with multiple congenital

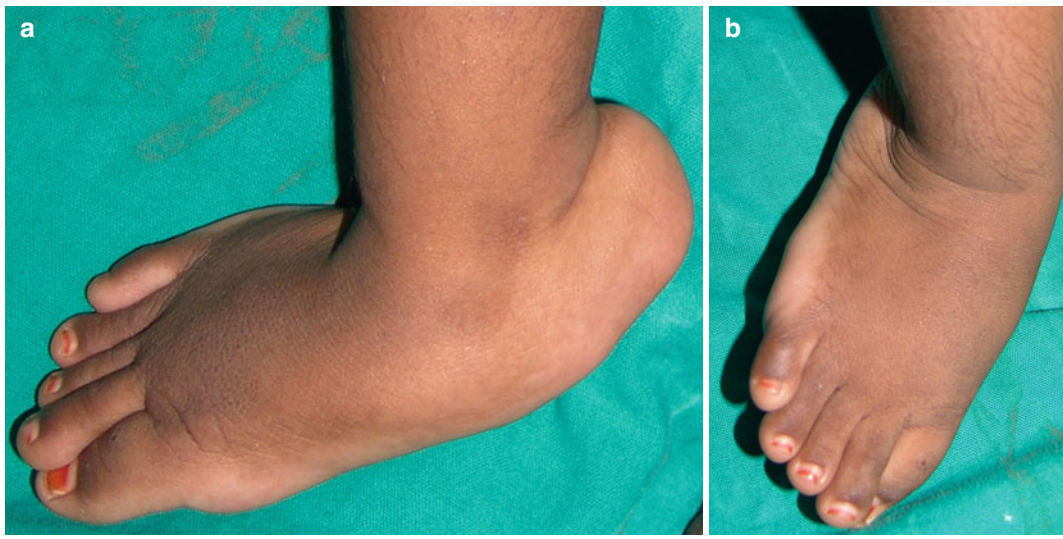

Fig. 6.5 The foot of a child with isolated vertical talus; the convex medial arch (**a**) and the eversion of the foot (**b**) are evident

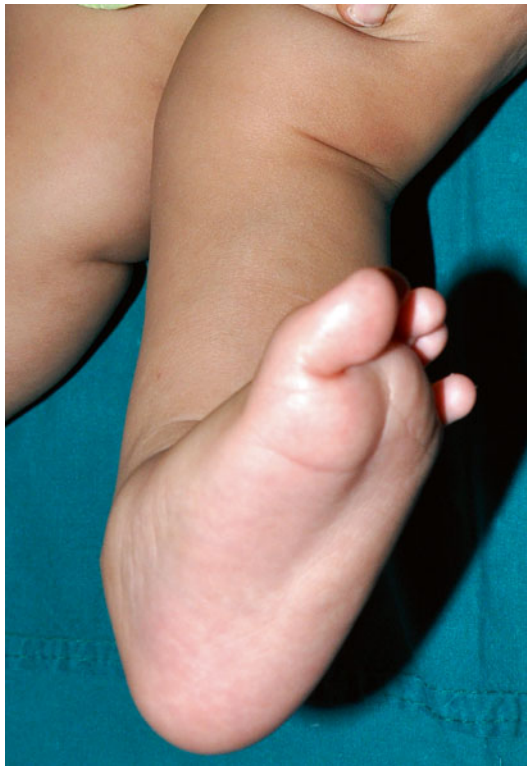

Fig. 6.6 Calcaneovalgus deformity of the foot and hyperextension of the knee in a newborn infant with multiple congenital contractures

contractures (Fig. 6.6). These deformities which are less frequent than equinovarus (see Chap. 5) are rigid and may not always respond to non-operative measures.

Neurogenic Foot

Valgus, calcaneovalgus, and vertical talus have all been seen in association with spina bifida (Rodrigues and Dias 1992; Frawley et al. 1998; Swaroop and Dias 2011). These deformities often are on account of muscle imbalance, but in some instances, the pattern of paralysis cannot account for the foot deformity (Frawley et al. 1998). Contractures of muscles and tendons are frequently seen, and tenotomies of the contracted tendons result in correction of the deformities in a large proportion of instances (Rodrigues and Dias 1992).

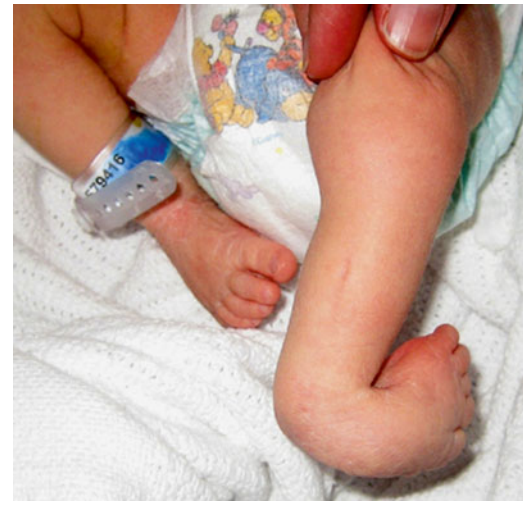

Fig. 6.7 Extreme eversion of the foot in a newborn with left-sided fibular aplasia

The Everted Foot in Limb Deficiency

Complete or partial aplasia of the fibula is usually associated with some degree of ankle valgus; occasionally the deformity can be very severe (Fig. 6.7). There is frequently an associated equinus deformity.

Congenital Pseudarthrosis of the Fibula

Isolated fibular pseudarthrosis is rare, but fibular pseudarthrosis is frequently seen in association with tibial pseudarthrosis. A significant proportion of children have features of neurofibromatosis type I, and there may be a clear family history of neurofibromatosis. A valgus deformity of the ankle is often seen if the distal fibula is dysplastic.

Rare Conditions

Congenital Dislocation of the Peroneal Tendons

Purnell et al. reported four cases of calcaneovalgus deformities in infants secondary to congenital dislocation of the peroneal tendons (Purnell et al. 1983). The deformity is more difficult to correct than the idiopathic form of calca-

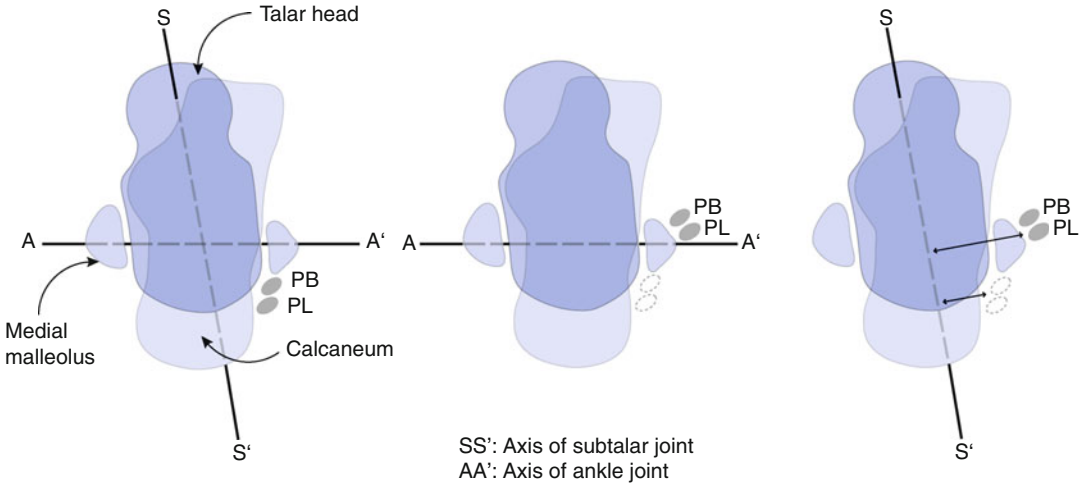

Fig. 6.8 Diagrammatic representation of the position of the peroneal tendons in relation to the axes of the ankle and subtalar joints in the normal foot (*left*) and when the peroneal tendons are dislocated (*middle* and *right*). When dislocated, the tendons lie anterior to the axis of the ankle joint and the evertor moment arm of the peronei increase as the perpendicular distance of the tendons from the axis increases quite substantially

neovalgus. The dislocated peroneal tendons act as perverted dorsiflexors of the ankle as they move anterior to the axis of the ankle joint when they dislocate. The evertor moment arm also increases when these tendons dislocate (Fig. 6.8).

6.6 Establishing the Diagnosis

An approach to the diagnosis of the cause of an everted foot in the newborn is outlined in Table 6.1.

Table 6.1 Establishing the diagnosis of the cause of the everted foot in the newborn

History						
History suggestive of crowding in utero may be present			History of reduced fetal movements (akinesia) may be present			Family history of neurofibromatosis may be present
Physical examination						
Skin normal	Skin normal	Skin normal	Skin normal	Skin normal	Skin normal	Café au lait spots on the body
Spine normal	Spine normal	Spine normal	Spine usually normal	Spina bifida or sacral agenesis	Spine normal	Spine usually normal at birth
May be associated with neonatal hip instability	No deformities of other limbs	No deformities of other limbs	Symmetrical deformities of the upper and lower limbs	Deformities of the hips and knees may be present	Other limb deficiencies may occasionally be present	No deformities of other limbs
Tibia normal	Tibia bowed posteromedially	Tibia normal	Tibia normal	Tibia normal	Tibia may be bowed anteromedially	Tibia may be normal/bowed anterolaterally or a pseudarthrosis of the tibia may be present
Tibia of normal length	Tibia short	Tibia of normal length	Tibia of normal length	Tibia of normal length	Tibia short	Tibia may be short
Fibula normal	Fibula bowed posteromedially	Fibula normal	Fibula normal	Fibula normal	Fibula partially or totally absent	Fibular pseudarthrosis in the lower third of the bone
No neurological deficit	No neurological deficit	No neurological deficit	Motor deficit may occasionally be demonstrable No sensory loss	Motor or sensory deficit may be demonstrable	No neurological deficit	No neurological deficit
The foot: Supple deformity (foot can usually be brought to the neutral posture passively) Gastrocsoleus *not* contracted	The foot: Supple deformity (foot can usually be brought to the neutral posture passively) Gastrocsoleus *not* contracted	The foot: Rigid deformity Gastrocsoleus always contracted	The foot: Rigid deformity Gastrocsoleus may be contracted (i.e., if the deformity is vertical talus)	The foot: Rigid deformity often Gastrocsoleus may be contracted (i.e., if the deformity is vertical talus)	The foot: Rigid deformity Gastrocsoleus may be contracted (if equinovalgus deformity is present)	The foot: Rigid deformity Gastrocsoleus *not* contracted

Working diagnosis Idiopathic congenital calcaneovalgus	Working diagnosis Calcaneovalgus associated with congenital posteromedial bowing of the tibia	Working diagnosis Isolated oblique or vertical talus	Working diagnosis Valgus/calcaneovalgus/ vertical talus associated with multiple congenital contractures	Working diagnosis Valgus/calcaneovalgus/ vertical talus associated with spina bifida or sacral agenesis	Working diagnosis Valgus deformity associated with fibular aplasia	Working diagnosis Valgus ankle associated with congenital pseudarthrosis of the fibula
Investigations						
Radiographs not indicated	Radiographs can confirm the bowing of the tibia	Lateral radiograph in plantarflexion to distinguish between oblique and vertical talus	Radiographs not routinely indicated	Radiographs of the spine to confirm the nature of spinal anomaly	Radiographs of the limb to confirm the extent of aplasia	Radiographs of the limb to assess the nature and extent of pseudarthrosis
Diagnosis						
Idiopathic congenital calcaneovalgus	Calcaneovalgus associated with congenital posteromedial bowing of the tibia	Isolated oblique talus (if the plantarflexion film shows restoration of normal tarsal relationships) Or Isolated vertical talus (if the plantarflexion film does not show restoration of normal tarsal relationships)	The everted foot in multiple congenital contractures	Neurogenic valgus/ calcaneovalgus/vertical talus	Valgus deformity of the foot associated with fibular aplasia	Ankle valgus secondary to congenital fibular pseudarthrosis

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The Deformed Shoulder Girdle

7

Benjamin Joseph

7.1 Introduction

Congenital anomalies of the scapula, clavicle, and shoulder are relatively rare (McClure and Raney 1975; Williams 2003) but when present can affect the range of motion of the shoulder and alter its contour. The more common anomalies of the scapula include abnormalities in the position, mobility, size, and shape of the scapula and its processes; agenesis and duplication are exceedingly rare. Anomalies of the clavicle include a short clavicle, partial failure of formation, and pseudarthrosis (Beals 2000; Currarino and Herring 2009; Fairbank 1949); a pseudarthrosis can be mistaken for a birth fracture of the clavicle. Abnormalities of muscles include contractures and aplasia; contractures of muscles of the shoulder that result in deformities and limitation of motion are relatively common (Axt et al. 1997).

7.2 Questions to Establish a Diagnosis

- **Is there a history of akinesia (decreased fetal movements) in pregnancy?**
- **Is the scapula small, at the normal level, and are the acromion and coracoid processes well formed?**
- **Is the clavicle palpable throughout its length?**
- **Is the passive range of motion of the shoulder normal? If not, what are the movements that are restricted?**
- **Are the skull, spine, and other extremities deformed or normal?**

Is there a history of akinesia (decreased fetal movements) in pregnancy?

A history of akinesia is often present in mothers of children with arthrogryposis.

Is the scapula small, at the normal level, and are the acromion and coracoid processes well formed?

Alterations in the shape and size and a proximal location of the scapula are features of the Sprengel anomaly. Rare anomalies of the scapula include failure of normal formation of the coracoid or the acromion.

Is the clavicle palpable throughout its length?

Total or partial agenesis of the clavicle is seen in cranioclеidodysostosis. A swelling in the middle of the clavicle and abnormal mobility will be present in congenital pseudarthrosis of the clavicle.

Is the passive range of motion of the shoulder normal? If not, what are the movements that are restricted?

Restriction of shoulder abduction is a feature of arthrogryposis and Sprengel anomaly. However,

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in arthrogryposis, external rotation of the shoulder is usually limited. Limitation of adduction of the shoulder will be seen in children with congenital deltoid contracture.

Are the skull, spine, and other extremities deformed or normal?

Delayed closure of the fontanelles is seen in cranioclеidodysostosis. A short neck with or without webbing and congenital scoliosis may be seen in association with the Sprengel anomaly. Symmetric deformities of other joints with limitation of movement will be seen in children with arthrogryposis.

7.3 Physical Examination

Clinical examination is often sufficient to establish the diagnosis.

7.3.1 Look

Note the contour of the shoulder and the position of the scapula and the clavicle. Look for asymmetry of the chest wall, and note if the hand and fingers are well developed. Observe if the spine is straight and if the neck is of normal length and if the hairline at the back is at the normal level. Note if there are symmetric deformities of the shoulders and other joints of the upper and lower limb.

7.3.2 Feel

Systematically palpate the scapula including the body, the medial and lateral borders, the superior and inferior angles, and the acromion and the coracoid processes. Palpate the clavicle throughout its length. Note if there is a discontinuity of the bone or a palpable swelling in the middle third.

7.3.3 Move

Test the passive ranges of abduction, adduction, flexion, extension, and rotations of the scapulohumeral joint. Note if the scapula moves on the rib cage. Move the ends of the clavicle to detect abnormal mobility. See if the shoulders can be approximated in the midline in the front of the chest.

7.3.4 Special Tests

Scapula elevation sign: If there is limitation of abduction of the glenohumeral joint, passively adduct the shoulder and observe if the angle of the scapula becomes prominent (see Sect. 16.2).

Auscultate the chest to determine if there is dextrocardia.

7.4 Investigations to Confirm the Diagnosis

Plain Radiographs

Obtain plain radiographs of the shoulders, chest, and clavicles and radiographs of the cervical and thoracic spines if there is a spinal deformity.

Magnetic Resonance Imaging

Obtain a MRI of the spinal cord if there is a deformity of the cervical or thoracic spine.

7.5 Differential Diagnosis

7.5.1 Anomalies of the Scapula

Congenital Elevation of the Scapula (Sprengel Anomaly)

This is the commonest anomaly of the scapula. The term “congenital elevation of the scapula” is a misnomer as the anomaly is due to failure of normal descent of the scapula during fetal development (Grogan et al. 1983). The scapula is smaller than normal, situated more proximally than normal and rotated such that the glenoid faces slightly downwards (Fig. 7.1a). The Sprengel anomaly can occur in isolation, but associated vertebral anomalies such as scoliosis, hemivertebrae, segmentation defects, spina bifida, diastematomyelia, anomalies of the rib cage, and the Klippel-Feil syndrome are frequently seen. Limitation of abduction of the shoulder is characteristically present in children

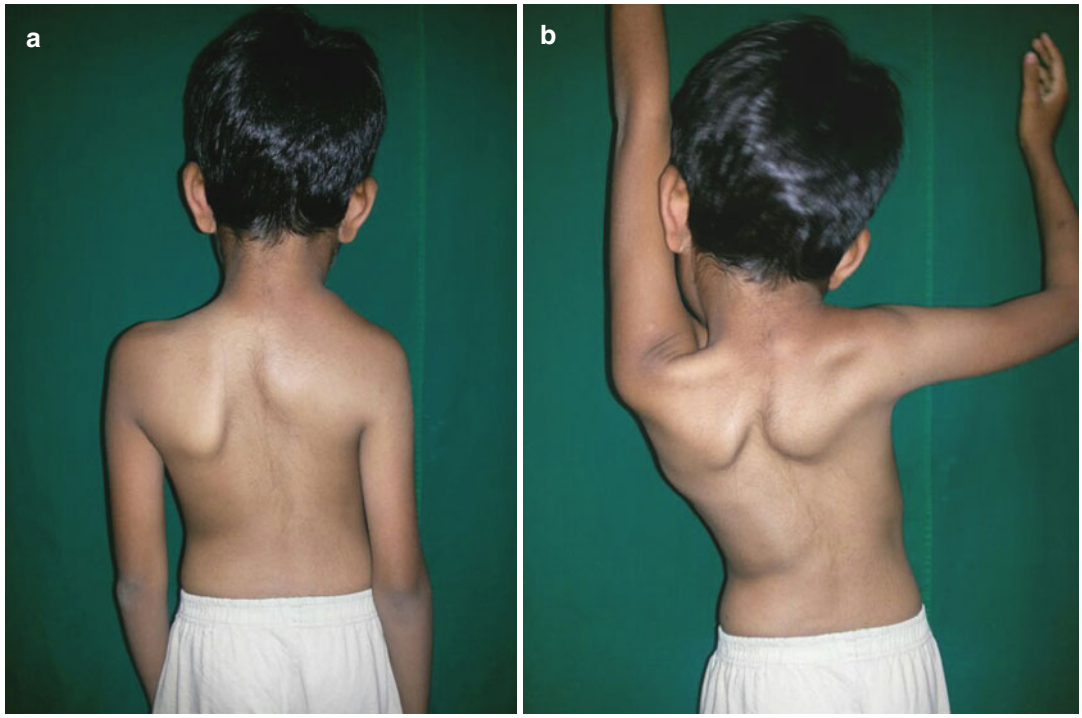

Fig. 7.1 Appearance of the back of a child with Sprengel anomaly on the right side (**a**); the scapula is proximally located and rotated. There is significant limitation of abduction of the shoulder in the side of the anomaly (**b**)

with this anomaly (Fig. 7.1b). This is partly due to the downward tilt of the glenoid. A bony, cartilaginous, or fibrous connection between the scapula and the posterior elements of the vertebrae (omovertebral connection) is present in up to half of children with the Sprengel anomaly; it restricts scapulothoracic movement and consequently abduction. Less frequently, a bony connection between the scapula and the clavicle may be present (Mikawa et al. 1991). The severity of Sprengel anomaly has been graded on how high the scapula is located; minor grades of Sprengel anomaly may go undetected, while the more severe grades are clearly visible and unsightly (Cavendish 1972).

Hypoplasia of the Scapula

The scapula is always hypoplastic in children with the Sprengel anomaly; hypoplasia of the scapula is also seen in campomelic dysplasia (Natasha et al. 2006). Hypoplasia of the scapula itself does not cause functional or cosmetic impairment, but its recognition may help in diagnosing an underlying dysplasia.

7.5.2 Anomalies of the Clavicle

Congenital Pseudarthrosis of the Clavicle

Congenital pseudarthrosis of the clavicle is almost invariably right sided (Lloyd-Roberts et al. 1975), and this has been attributed to pressure on the developing clavicle from the subclavian artery that is higher on the right than on the left side. Left-sided involvement has been noted with dextrocardia (Lloyd-Roberts et al. 1975). Bilateral involvement is very rare (Mooney and Koman 1991). The pseudarthrosis is usually in the middle third of the clavicle; the presenting feature is a swelling in the region (Fig. 7.2). It is distinguished from a birth fracture by the lack of pain and tenderness. Thoracic outlet syndrome in adolescents with congenital pseudarthrosis of the clavicle has been reported (Sales de Gauzy et al. 1999; Watson et al. 2013).

Cleidocranial Dysostosis

The clavicles may be totally or partially absent, and as a result, the shoulders can often be approximated in the midline (Fig. 7.3). The contour of

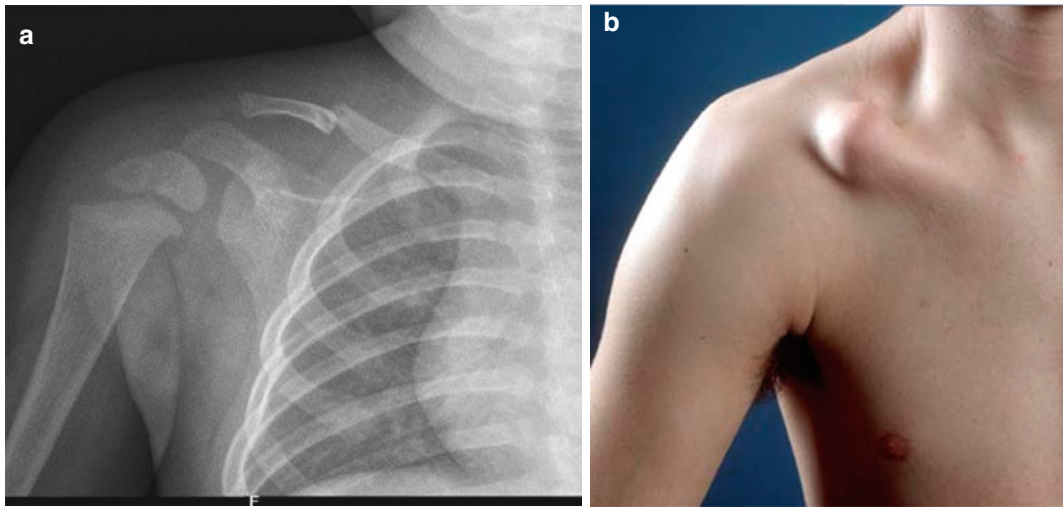

Fig. 7.2 Radiograph of a child with a congenital pseudarthrosis of the right clavicle (**a**) and the clinical appearance in an older child who did not have any treatment (**b**)

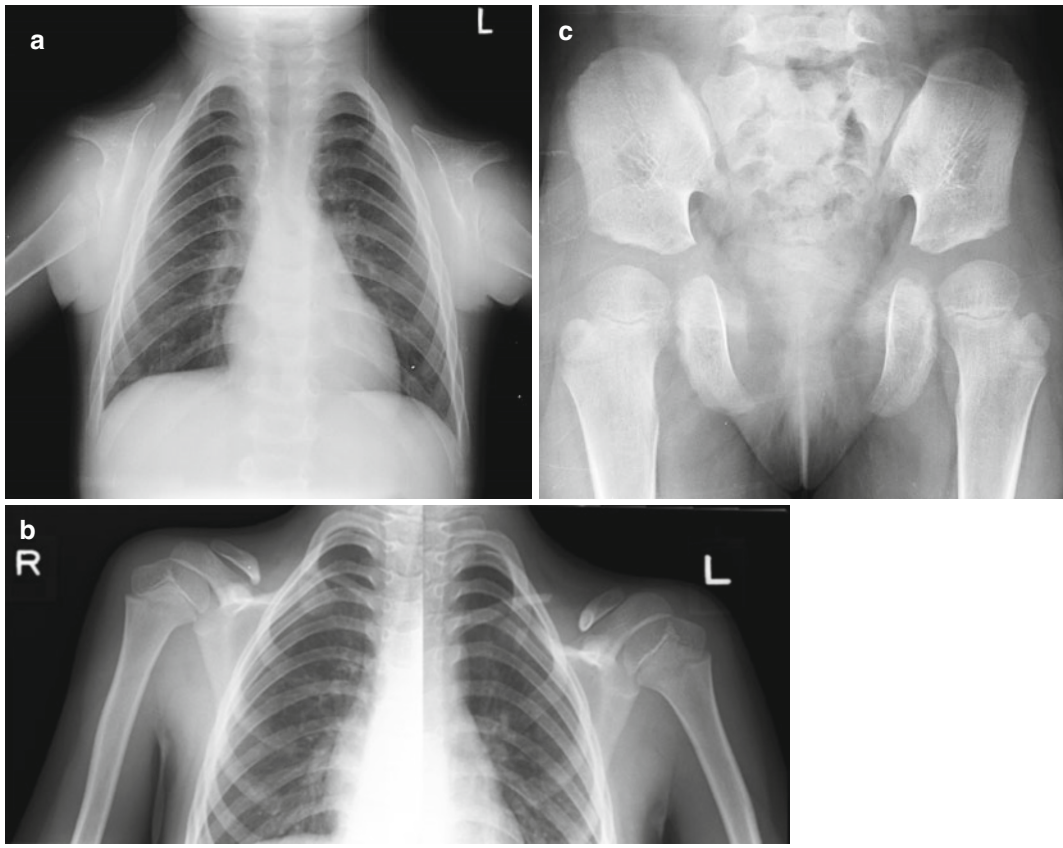

Fig. 7.3 Radiographs of the chest of two children with craniocleidodysostosis; in one child, the clavicles are totally absent (**a**), while in the other child, the lateral ends of the clavicles are present (**b**). The pubic bones are also not formed (**c**)

the shoulder is altered. Associated anomalies include delayed closure of the fontanelles, abnormal dentition, failure of ossification of the pubic bones, scoliosis, and genu valgum.

7.5.3 Abnormalities of Muscles

Contractures

Internal rotation and adduction contractures of the shoulder are commonly seen in children with arthrogryposis (Axt et al. 1997; Bennett et al. 1985; Williams 1985; Zlotolow and Kozin 2012). Congenital deltoid contracture may rarely occur, warranting surgical release in early childhood (Bhagat et al. 2008; Chari et al. 1979).

Aplasia

Aplasia of muscles around the shoulder girdle has been recognized; the Poland syndrome includes one such anomaly. In Poland syndrome, the sternocostal head of the pectoralis major is absent, and the hand on the affected side is hypoplastic with short fingers and syndactyly. Shortening or aplasia of the middle phalanges of the fingers has been recognized as the cause for the short fingers (Al-Qattan and Al Thunayan 2005).

7.6 Establishing the Diagnosis

An outline of the process of establishing a diagnosis of the cause of diminished movements of a limb in the neonate is shown in Table 7.1.

Table 7.1 Diagnosis of the cause of a deformed shoulder

Physical examination				
Shoulder Contour altered Chest normal Back: prominence of upper angle of the scapula (in the back of the neck) Neck may be short and webbed	Shoulder Contour drooping May be possible to approximate the shoulders in the midline Chest narrow Back may be normal or scoliosis may be present Neck normal	Shoulder Contour normal or drooping Chest : swelling over the middle of the clavicle Back normal Neck normal	Shoulder Contour normal Chest normal Back: prominence of angle of the scapula Neck normal	Shoulder Contour normal Chest: absent pectoralis major (sternal head) Back normal Neck normal
Side involved: Left/right/both	Side involved: Both	Side involved: Almost always right (when left is involved, dextrocardia may be present)	Side involved: Left/right/both	Side involved: Left/right
Scapula: More proximally situated Small Rotated so that the glenoid faces more caudally	Scapula: Normal	Scapula: Normal	Scapula: Rotated so that the glenoid faces more caudally Angle of scapula prominent when the arm is at the side	Scapula: Normal
Clavicle: Normal	Clavicle: Totally or partially absent	Clavicle: Abnormal mobility Bony swelling at the site of abnormal mobility	Clavicle: Normal	Clavicle: Normal
Hand normal	Hand normal	Hand normal	Hand normal	Brachydactyly Syndactyly

(continued)

Table 7.1 (continued)

Working diagnosis: Sprengel anomaly	Working diagnosis: Craniocleidodysostosis	Working diagnosis: Congenital pseudarthrosis of the clavicle	Working diagnosis: Congenital deltoid contracture	Working diagnosis: Poland syndrome
Investigations				
Plain radiographs: Confirms proximal location and hypoplasia of the scapula May show omovertebral bone	Plain radiographs: Confirms absence of part or all of the clavicle, failure of formation of pubic rami and body of the pubis and open fontanelles	Plain radiograph: Confirms pseudarthrosis of the clavicle	Plain radiograph: Confirms rotated scapula of normal size and shape	Plain radiograph: Normal
Diagnosis				
Sprengel anomaly	Craniocleidodysostosis	Congenital pseudarthrosis of the clavicle	Congenital deltoid contracture	Poland syndrome

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The Deformed Elbow

8

Benjamin Joseph

8.1 Introduction

The elbow in the newborn may be flexed or extended, and the underlying pathology may involve the soft tissue or the bones. Minor degrees of flexion deformity may be seen in a host of conditions and seldom merit any intervention. The causes of the more severe degrees of deformity and limitation of motion are considered in this chapter.

8.2 Questions to Establish a Diagnosis

- **Was there a history of reduced fetal movements (akinesia) during pregnancy?**
- **Is there symmetrical bilateral involvement?**
- **Is any passive motion possible from the deformed position?**
- **Are there associated anomalies of the involved limb?**
- **Is there webbing of the front of the elbow?**
- **Are there anomalies of the ears, face, or fingers?**

Was there a history of reduced fetal movements (akinesia) during pregnancy and is there symmetrical bilateral involvement?

A history of akinesia and symmetrical bilateral deformities are characteristically seen in the amyoplasia form of multiple congenital contractures (classical arthrogryposis).

Is any passive motion possible from the deformed position?

No passive motion will be possible beyond the deformed position if there is a synostosis between the humerus and the radius or ulna. If there is a contracture of the flexor or extensor muscle that is so severe that it holds the elbow completely extended (when there is a contracture of the triceps) or completely flexed (when there is a contracture of the biceps and brachialis), no passive motion will be possible.

Are there associated anomalies of the involved limb?

Symmetrical deformities of other joints are characteristic of multiple congenital contractures, while associated reduction defects such as aplasia of the digits indicate a congenital anomaly of the elbow joint.

Is there webbing of the front of the elbow?

Webbing of the elbow is diagnostic of antecubital pterygium which may occur in association with webbing of the neck and popliteal pterygia (Escobar syndrome) or in association with humeroradial synostosis.

Are there anomalies of the ears, face, or fingers?

Crumpled ears are characteristic of Beals syndrome, as is arachnodactyly. Facial dysmor-

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phism may be noted in syndromes associated with synostosis at the elbow.

8.3 Physical Examination

Examination of the elbow can often distinguish between soft tissue and bony causes of congenital elbow deformities.

8.3.1 Look

Look for anomalies of the skull, face, ears, neck, and spine. Note if there are symmetrical deformities of the limbs. If the deformity involves only one elbow, look if the affected limb is hypoplastic and if there is absence of rays of the hand. If the elbow is in flexion, note if the front of the elbow is webbed.

Examine the hand and look for flexion deformity of the fingers, arachnodactyly, and symphalangism.

8.3.2 Feel

Palpate the humerus, ulna, and radius throughout their entire lengths, and note if they are completely formed and of normal length.

8.3.3 Move

Attempt to passively move the elbow and record if any movement if possible.

8.4 Investigations to Confirm the Diagnosis

Plain Radiographs
Plain radiographs of the entire extremity are mandatory if the limb is maldeveloped or if no passive motion is possible at the elbow.

8.5 Differential Diagnosis

8.5.1 Multiple Congenital Contractures

The elbow in classical arthrogryposis or amyoplasia may be in extension (Fig. 8.1) due to a contracture of the triceps with weakness of flexor muscles or in flexion due to contracture of the biceps and brachialis muscles with weakness of extension (Williams 1985; Axt et al. 1997).

8.5.2 Beals Syndrome

Beals syndrome or congenital contractural arachnodactyly is characterized by flexion contractures resembling arthrogryposis, arachnodactyly resembling Marfan syndrome, crumpled ears, and muscle hypoplasia. The contractures that are most pronounced at the elbows, knees, and fingers tend to improve spontaneously to a variable degree (Green and Lesser 2006; Tuncbilek and Alanay 2006).

8.5.3 Antecubital Pterygium

Webbing of the flexor aspect of the elbow with a flexion deformity is referred to as the antecubital pterygium (Fig. 8.2). It may occur in isolation or as part of the multiple pterygium syndrome (Gillin and Pryse-Davis 1976; Escobar et al. 1978; Chen et al. 1980). The anomaly has been observed in families, and both autosomal and recessive patterns of inheritance have been noted. Absence of the long head of the triceps was noted in one family (Wallis et al. 1988).

8.5.4 Humeroradial Synostosis

Synostosis of the humerus and radius may occur very rarely as an isolated anomaly, in association with ulnar aplasia, as part of multiple synostosis syndromes, or as a component of other generalized syndromes (Lambert 1947;

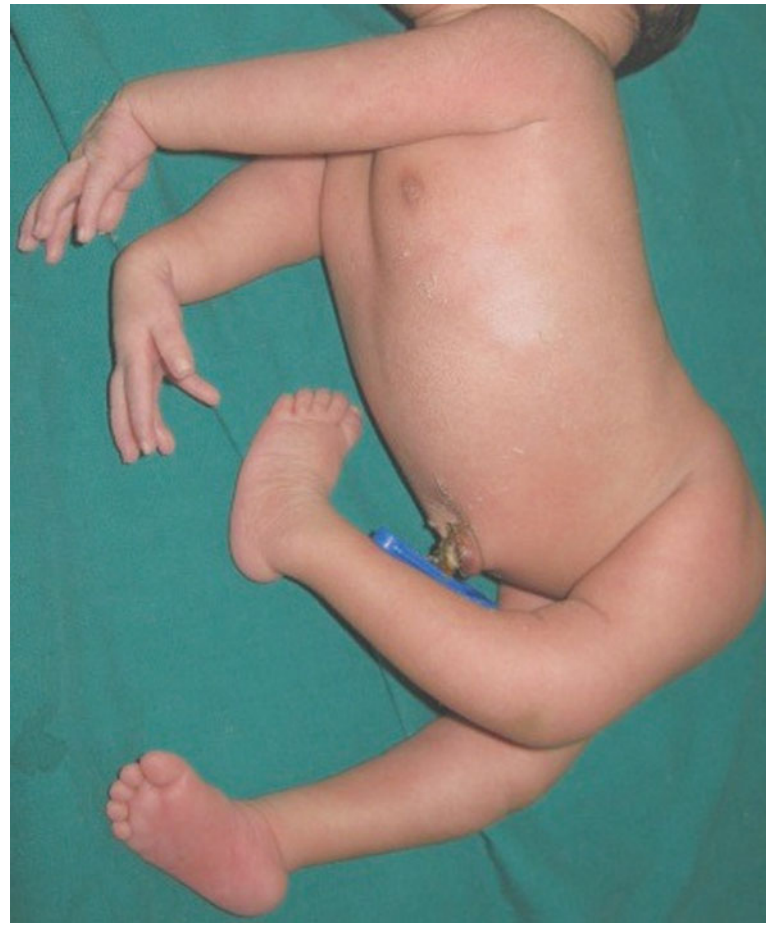

Fig. 8.1 A newborn infant with arthrogryposis and extended elbows. Symmetrical deformities of the lower limbs are also evident

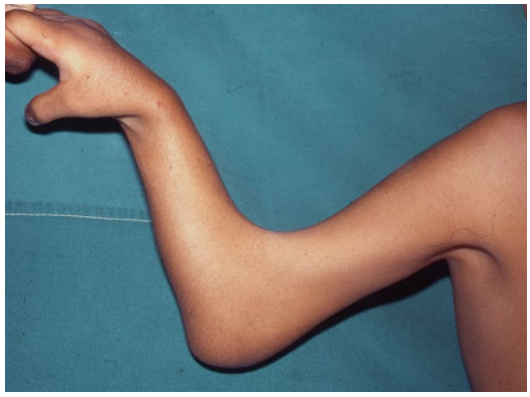

Fig. 8.2 Antecubital pterygium

Jacobsen and Crawford 1983; Drawbert et al. 1985; McIntyre et al. 2003). The fused elbow may be in extension or flexion (McIntyre and Benson 2002). They classified humeroradial synostosis into two main groups, those with primary joint maldevelopment without hypoplasia of the bones and those with hypoplasia or aplasia of the bones, and then subdivided these groups according to whether the elbow is ankylosed in extension or flexion (Fig. 8.3). The commonest association with this anomaly is ulnar aplasia (Fig. 8.4), which in turn may be part of a syndrome. Among the various

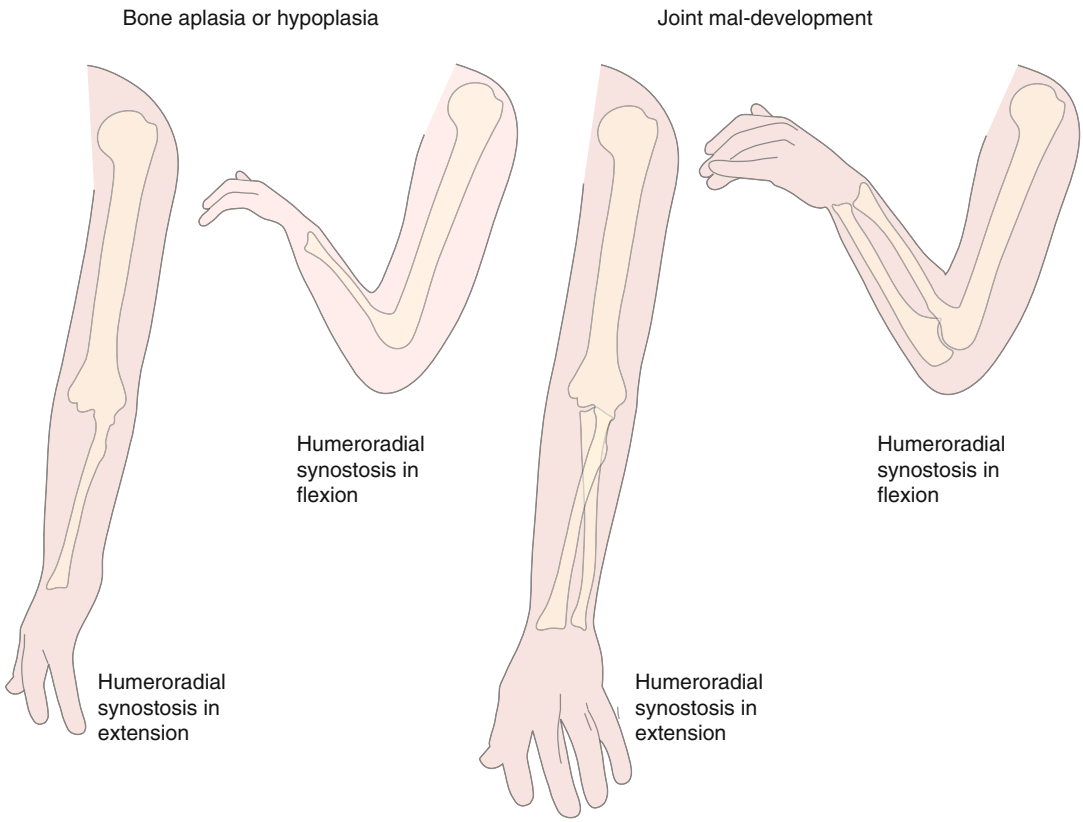

Fig. 8.3 Classification of humeroradial synostosis suggested by McIntyre and Benson

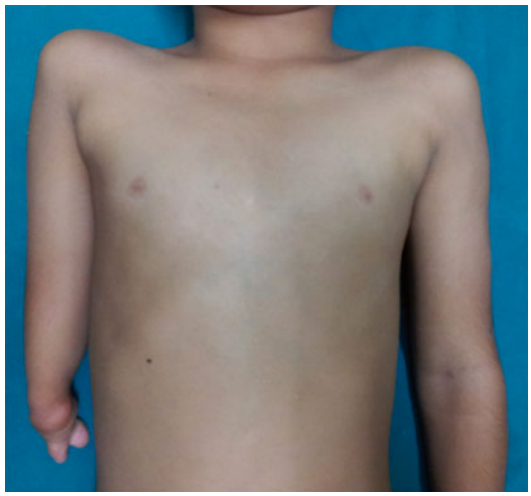

Fig. 8.4 The upper limb of a child with ulnar aplasia and humeroradial fusion in extension. The hypoplasia of the limb and the absence of postaxial rays are seen

syndromes identified with this anomaly, some demonstrate an autosomal dominant inheritance pattern, while others show a recessive trait (Table 8.1) (Drawbert et al. 1985; Bianchi et al. 1991; Kantaputra and Mongkolchaisup 1999). Sporadic cases have also been described, and hence, a positive family history of similar defects may not always be present (McIntyre and Benson 2002). Symphalangism is often present in multiple synostosis syndromes (Drawbert et al. 1985).

8.5.5 Rare Conditions

Humeroulnar and Humeroradioulnar Synostosis

Humeroulnar synostosis is three times less frequent than humeroradial synostosis; isolated synostosis and synostosis as part of a more complex syndrome have been described (Marles et al. 2003; Musa 2004). Humeroradioulnar synostosis is exceedingly rare (Kakarala et al. 2006); it is estimated that this form of elbow synostosis is

Table 8.1 Classification of the types of humeroradial synostosis

Humeroradial synostosis with bony aplasia or hypoplasia (usually ulnar deficiency)		Humeroradial synostosis with joint maldevelopment with no bone hypoplasia	
Autosomal dominant (AD)	Autosomal recessive (AR)	Autosomal dominant (AD)	Autosomal recessive (AR)
Cornelia de Lange syndrome (AD inheritance if mutation in NIPBL or SMC3 gene but X-linked if mutation in SMC1A gene) Limb hypoplasia–renal disease syndrome	SC phocomelia syndrome Robert's syndrome Femoral–fibula–ulna complex	Multiple synostosis syndromes Pfeiffer syndrome Apert syndrome	Antley–Bixler syndrome

Modified from McIntyre and Benson (2002)

ten times less frequent than humeroradial synostosis (McIntyre and Benson 2002).

Omodysplasia and Distal Humeral Dysplasia

Omodysplasia and distal humeral dysplasia are rare anomalies of the distal humerus, associated with dwarfism and characteristic facial dysmorphism (Joseph and Varghese 2003; Sauvegrain and Maroteaux 1992). In omodysplasia, the distal end of the humerus is dysplastic, the radial head is dislocated, and there may be a flexion deformity of the elbow (Sauvegrain and Maroteaux 1992).

Congenital Absence of Muscles

Congenital absence of the biceps results in an extended elbow with no active flexion (Ethans and Leahey 2001). Similarly, congenital absence of the triceps results in a flexion deformity (Wallis et al. 1988).

8.6 Establishing the Diagnosis

An outline for establishing the causes of deformities of the elbow at birth is shown in Table 8.2.

Table 8.2 An outline of the process of establishing the diagnosis of deformities and diminished movement of the elbow in the newborn

History					
Family history seldom present		A positive family history may be present		A positive family history may be present	
History of akinesia during pregnancy may be present		History of akinesia may be present		History of akinesia not present	
Physical examination					
Symmetrical contractures of multiple joints		Webbing on the flexor aspect of the elbow		Ankylosis of the elbow	
Elbow extended or flexed	Elbow flexed	Elbow flexed		Elbow extended or flexed	
External ears normal	External ears crumpled	Webbing of the neck absent	Webbing of the neck may be present	Affected limb hypoplastic	Affected limb not hypoplastic
Arachnodactyly absent	Arachnodactyly present	Webbing in the popliteal region absent	Webbing of the popliteal region may be present	Absence of rays of the hand often	No absence of the rays of the hand
				Major reduction defects may be present	Major reduction defects not present
				Symphalangism not present	Symphalangism often present
Investigations					
Radiograph not indicated (unless the elbow is totally stiff – to rule out bony ankylosis)	Radiograph not indicated	Radiograph not indicated	Radiograph not indicated	Radiograph of the entire limb Will show: The nature and extent of synostosis The nature and extent of bony hypoplasia or aplasia	Radiograph of the entire limb Will show: The nature and extent of the elbow synostosis Carpal synostosis or symphalangism if present
Diagnosis					
Arthrogryposis	Beals syndrome (congenital contractural arachnodactyly)	Isolated antecubital pterygium	Multiple pterygium syndrome	Humeroradial synostosis associated with ulnar hypoplasia or aplasia	Humeroradial synostosis (isolated or part of multiple synostosis syndrome)

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The Deformed Wrist

9

Benjamin Joseph

9.1 Introduction

Deformities of the wrist seen in the newborn include flexion and radial or ulnar deviation. Flexion deformity is most frequently seen in children with arthrogryposis (Smith and Drennan 2002), while radial or ulnar deviation is seen in association with radial or ulnar deficiencies (Broudy and Smith 1979; Roth et al. 1996).

9.2 Questions to Establish a Diagnosis

- **Are both wrists symmetrically deformed?**
- **Are there deformities of other joints associated with limitation of motion?**
- **Is the forearm short and is there aplasia of the thumb or fingers?**
- **Are the stature and body proportions normal?**

Are both wrists symmetrically deformed?

Symmetrical deformities are the norm in multiple congenital contractures (arthrogryposis) and occasional in children with radial or ulnar deficiencies. Symmetrical deformities may also be seen in mesomelic skeletal dysplasia.

Are there deformities of other joints associated with limitation of motion?

Deformities of the proximal joints of the upper limb and joints of the lower limb are seen in multiple congenital contractures. Deformities of the elbow may be seen in children with limb deficiencies.

Is the forearm short and is there aplasia of the thumb or fingers?

Aplasia of the fingers or thumb and a short forearm are characteristically seen in deficiencies of the radius or ulna. Short forearms will be present in mesomelic dysplasia.

Are the stature and body proportions normal?

Dwarfism and altered body proportions are seen in skeletal dysplasias associated with wrist deformities.

9.3 Physical Examination

9.3.1 Look

Note if there are deformities of the proximal joints of the limb and of joints of other limbs.

9.3.2 Feel

Palpate the forearm and note if the radius and the ulna can be palpated and if they are of normal length.

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9.3.3 Move

Move the wrist and note if the range of motion of the wrist is reduced. Check the range of motion of the elbow.

9.4 Investigations to Confirm the Diagnosis

Plain Radiographs
Plain radiographs of the affected limb are needed if hypoplasia or aplasia of the radius or ulna is suspected.

9.5 Differential Diagnosis

9.5.1 Multiple Congenital Contractures

Flexion deformity of the wrist is a common deformity in classical arthrogryposis. The deformity can be quite severe (Fig. 9.1).

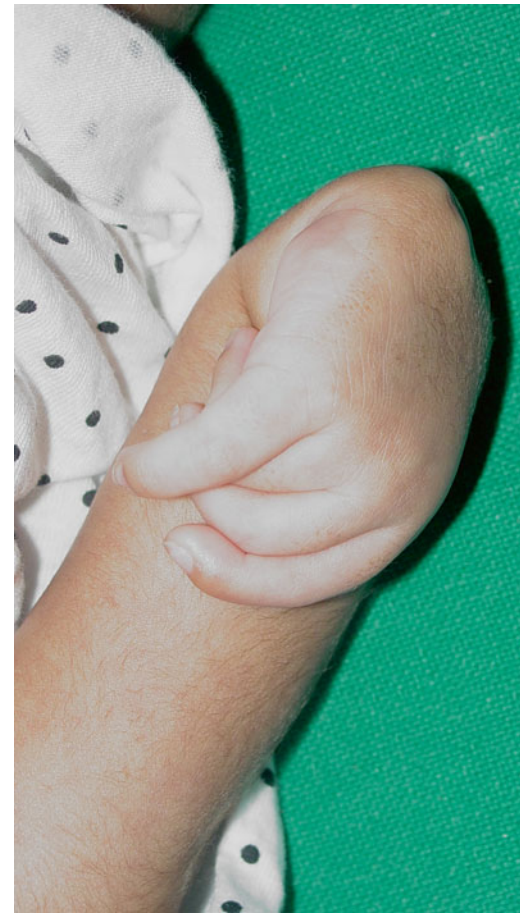

Fig. 9.1 Severe flexion deformity of the wrist in a newborn with multiple congenital contractures

9.5.2 Radial and Ulnar Deficiency

Radial deviation of the wrist is characteristically seen in radial deficiency, and the extent of radial deviation reflects the severity of the deficiency (Fig. 9.2). However, ulnar deviation is not invariably associated with ulnar aplasia (Fig. 9.3); in one study, only three out of 26 children with ulnar aplasia had ulnar deviation of the wrist greater than 20° (Broudy and Smith 1979). Another fact that needs to be borne in mind is that while radial aplasia is characteristically associated with absence of the radial rays, digital aplasia associated with ulnar aplasia does not always follow a comparable pattern. Both ulnar digit aplasia and aplasia of the thumb can occur with ulnar aplasia (Broudy and Smith 1979).

9.5.3 Langer Mesomelic Dysplasia, Leri-Weill Dyschondrosteosis, and Turner Syndrome

Langer mesomelic dysplasia is characterized by disproportionate dwarfism, very short ulna and fibula with bowing of the radius and tibia. This form of dysplasia can be diagnosed at birth or even prenatally (Roth et al. 1996). It is now clear that mutations of the short stature homeobox-containing gene (SHOX gene) cause the characteristic forearm and wrist deformities in Langer mesomelic dysplasia, Leri-Weill dyschondrosteosis, Turner syndrome, and Madelung deformity (Grigelioniene et al. 2001; Leka et al. 2006).

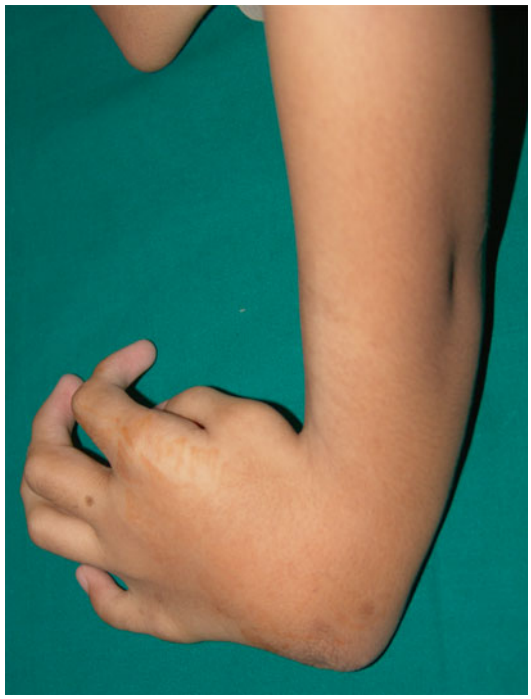

Fig. 9.2 Severe radial deviation of the wrist in a child with radial aplasia

Ulnar deviation of the wrist is seen in Leri-Weill syndrome and Turner syndrome (Tauber et al. 2004). However, since the wrist changes are more manifest in the older child, these conditions are discussed in Chapter 19.

9.5.4 Rare Conditions

A rare novel syndrome with ulnar deviation of the wrist associated with carpal aplasia and joint contractures has been described (Phadke and Dalal 2007).

9.6 Establishing the Diagnosis

An outline of the process of establishing the diagnosis of the cause of a deformed wrist in the newborn is shown in Table 9.1.

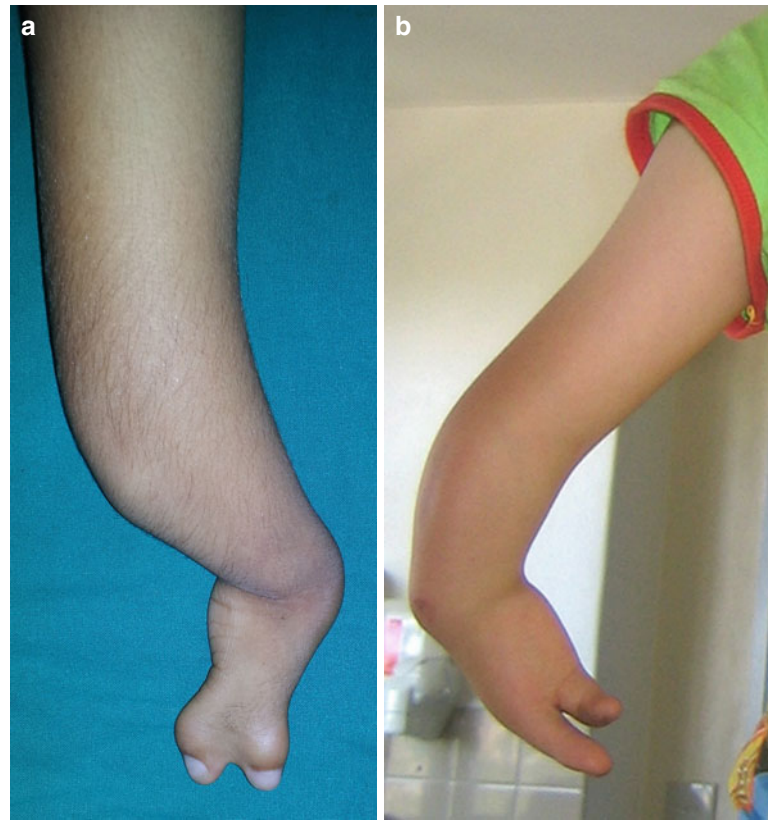

Fig. 9.3 Ulnar deviation of the wrist and absence of rays in a child with ulnar aplasia (**a**). Bowing of the forearm is also evident in another child (**b**)

Table 9.1 An outline of the process of establishing the diagnosis of the cause of a wrist deformity at birth

History			
Family history absent	Family history may be present	Family history may be present	Family history may be present
History of akinesia (reduced fetal movement) during pregnancy may be present	No history of akinesia	No history of akinesia	No history of akinesia
Physical examination			
Flexion deformity of the wrist	Radial deviation of the wrist (valgus) frequently seen	Ulnar deviation of the wrist may be present or the wrist may not be deviated	Ulnar deviation of the wrist always present
No deficiencies of fingers	Thumb may be hypoplastic or absent	Fingers missing	No deficiencies of fingers
Thumb may be adducted	Thumb may have poor function	Thumb anomalies often seen	No thumb anomalies
Forearm not short	Forearm short	Forearm short	Forearm short
Forearm not bowed	Forearm may be bowed	Forearm may be bowed	Forearm and leg bowed
Symmetric involvement	May be symmetric – often asymmetric	Seldom symmetric	Symmetric involvement
Limitation of movements of joints of the lower limb present	No limitation of movements of joints of the lower limb present	No limitation of movements of joints of the lower limb present	No limitation of movements of joints of the lower limb present
Lower limbs of normal length	Lower limbs of normal length	Lower limbs of normal length	Leg disproportionately short
Stature normal	Stature normal	Stature normal	Short stature
Working diagnosis: Wrist flexion deformity in arthrogryposis	Working diagnosis: Radial deviation of the wrist in radial longitudinal deficiency	Working diagnosis: Wrist deformity associated with ulnar longitudinal deficiency	Working diagnosis: Langer mesomelic dysplasia
Radiographs of the upper limb not indicated	Radiographs of the upper limb will show extent of radial longitudinal deficiency	Radiographs of the upper limb will show extent of ulnar longitudinal deficiency	Radiographs will show disproportionately short ulna and fibula with bowing of the radius and tibia bilaterally
Diagnosis			
Wrist deformity in arthrogryposis	Wrist deformity associated with radial longitudinal deficiency	Wrist deformity associated with ulnar longitudinal deficiency	Langer mesomelic dysplasia

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The Deformed Spine in the Newborn

10

Ian Torode

10.1 Introduction

Spinal deformity as a visual entity in the newborn is relatively uncommon although the deformity itself may take many forms and be due to a myriad of causes. Paradoxically, the more obvious deformities of the spine at birth are at the more benign end of the spectrum, whereas the babies with more severe underlying congenital anomalies often do not show the deformity at birth but the deformity develops with growth.

The spectrum includes normal children born with benign resolving scoliosis through congenital abnormalities, named syndromes, and even potentially lethal conditions such as Jarcho-Levin syndrome.

Diagnosis of both the condition and the spinal deformity will follow examination of antenatal ultrasounds and physical observation of the newborn on arrival.

10.2 Questions to Establish a Diagnosis

- **What were the prenatal ultrasound findings?**
- **Is there a family history?**
- **Is there a history of maternal alcohol abuse?**
- **Is the child apparently normal apart from a deformity of the neck or trunk?**
- **Are there features of proportionate or disproportionate dwarfism?**
- **Are there anomalies of the limbs or other anatomical features?**
- **Is the baby in pain or respiratory distress?**
- **Are there abnormalities of the skin over the spinal column (swelling, dimples, hairy areas, etc.)?**
- **Do the limbs move spontaneously or with stimulation?**
- **What is the site of the deformity?**
- **What is the plane of the deformity?**

What are the prenatal ultrasound findings?

An antenatal ultrasound should be able to delineate the formation of the vertebral column and detect abnormalities in vertebral formation such as the presence of single or multiple hemivertebrae. The overall alignment of the vertebral column should be visible and any acute deviations or discontinuity noted. Furthermore, the routine antenatal ultrasound will examine the transnuchal line thickness which is associated with genetic abnormalities. Diastematomyelia, neurenteric cysts, and sacral agenesis have all been reported in ultrasound examinations (Gadodia et al. 2010; Jackson and Rose 1998; Dane et al. 2007; Karasahin et al. 2009).

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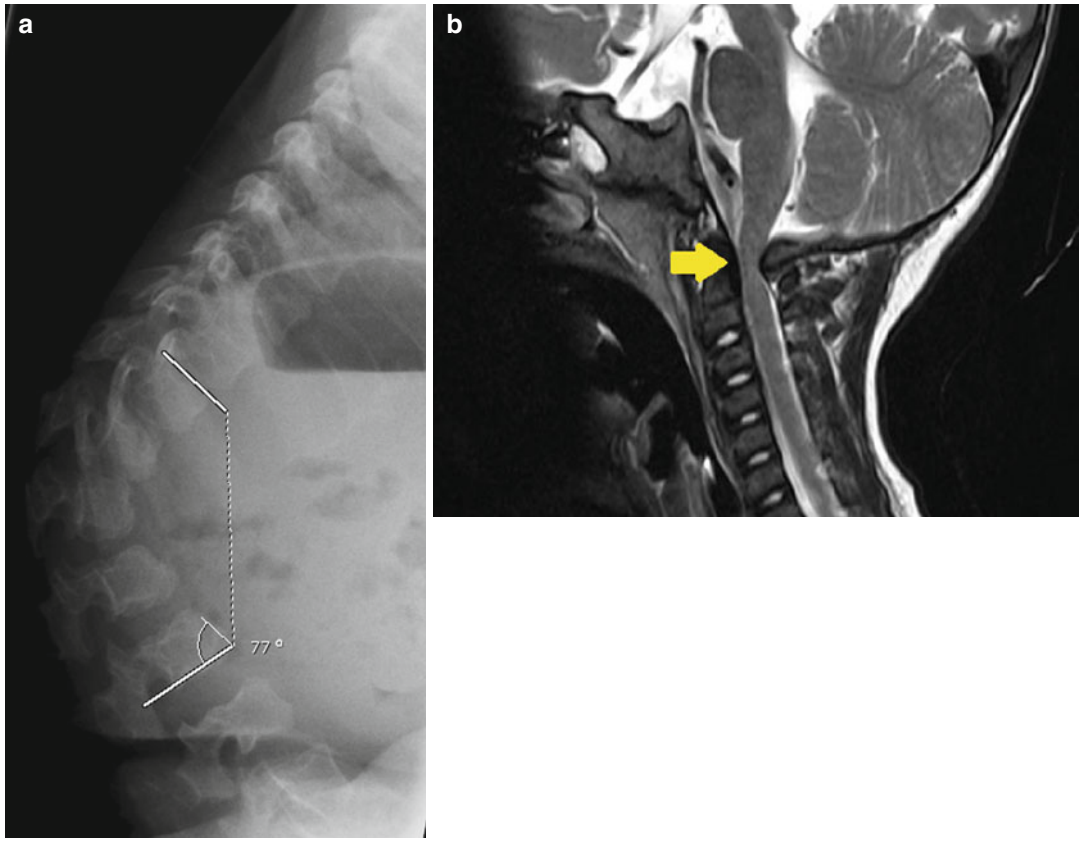

Fig. 10.1 Thoracolumbar kyphosis in an infant with achondroplasia (**a**) and stenosis of the foramen magnum in achondroplasia (**b** – *yellow arrow*)

Is there a family history?

While an isolated hemivertebra is not hereditary, conditions such as spondylocostal dysplasia may be familial. A family history is most important for syndromes such as neurofibromatosis, osteogenesis imperfecta, and achondroplasia.

Is there a history of maternal alcohol abuse?

Eliciting a history of alcohol abuse by the mother is important as fetal alcohol syndrome is associated with congenital spine fusions (Tredwell et al. 1982).

Is the child apparently normal apart from a deformity of the neck or trunk?

When presented with an apparently normal child with a deformity of the head and neck, one should consider the likelihood of congenital anomalies between the occiput and the thoracic spine. The other more common possibility is a muscular torticollis with or without a sternocleidomastoid "tumor."

In the trunk, similar possibilities exist. A thoracic hemivertebra may produce a scoliotic deformity; however, more commonly, a thoracic scoliosis will be an early onset curve which may resolve spontaneously or persist and demand intervention.

Are there features of proportionate or disproportionate dwarfism?

Disproportionate dwarfism is a feature of skeletal dysplasia and achondroplasia is a prime example in this group. There may be a family history. The baby will have the typical features of short limbs and a trunk of relatively normal length. There are two spinal issues in achondroplasia. There is often a thoracolumbar kyphosis (Fig. 10.1a) which in severe cases warrants intervention. Spinal stenosis however may be silent but should be suspected as stenosis at the

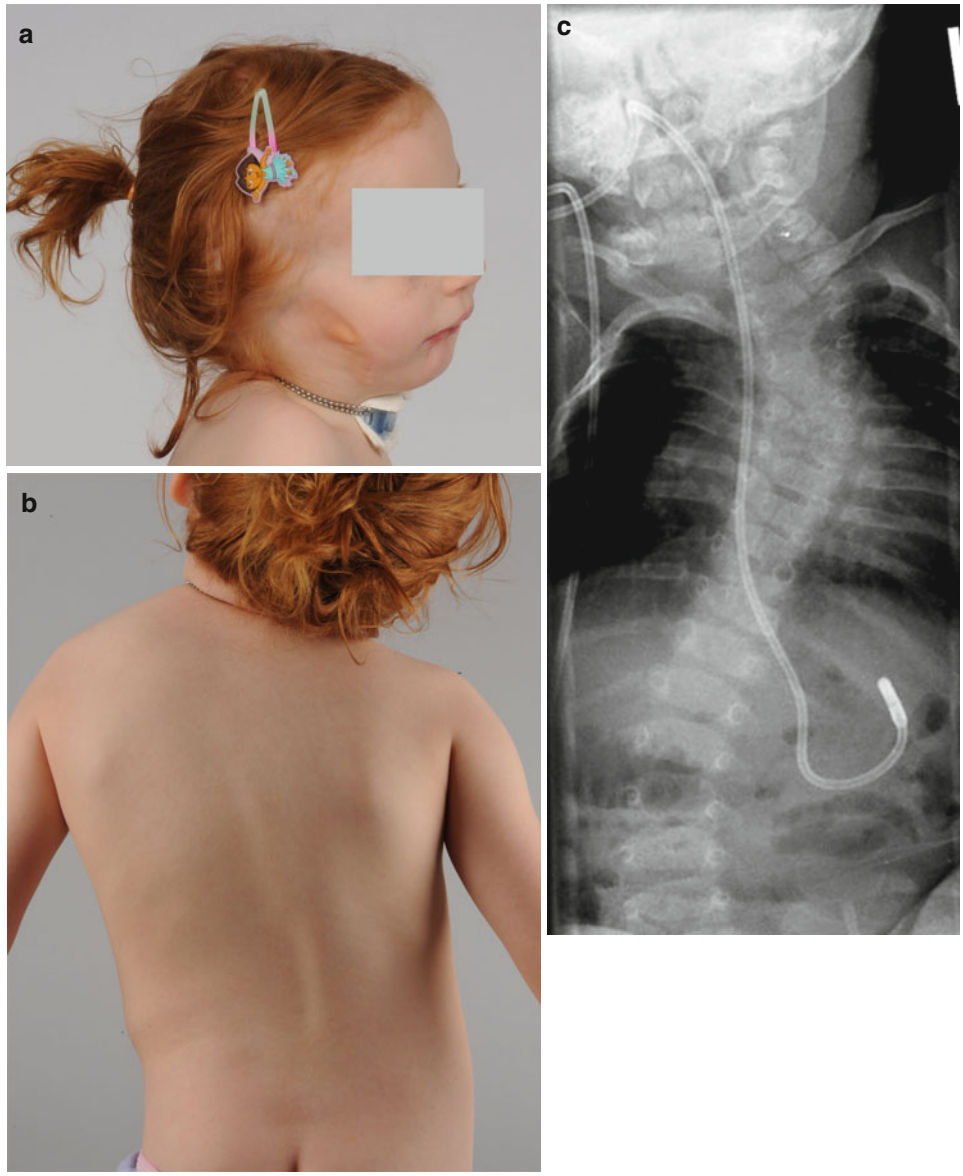

Fig. 10.2 Agenesis of the ear (**a**) and scoliosis (**b**, **c**) in a child with Goldenhar syndrome

foramen magnum can have profound effects on the baby (Fig. 10.1b). Diastrophic dysplasia in addition to short stature typically has hitchhiker thumbs, feet deformities and joint contractures. Scoliosis is common and the curves are usually severe and progressive. Babies severely affected by osteogenesis imperfecta may appear disproportionate due to intrauterine fractures and limb deformity. The skull may be deformed and basilar invagination can be an issue. Scoliosis is common.

Are there anomalies of the limbs or other anatomical features?

Certain physical features are readily apparent in some children. For example, the absence of an ear and facial deformity should alert one to Goldenhar syndrome (Fig. 10.2a) and the underlying vertebral anomalies (Fig. 10.2b, c) (Avon and Shively 1988). A radial club hand raises the possibility of VACTERL syndrome with vertebral anomalies and possible deformity. Ligamentous laxity and joint deformities and

Table 10.1 The site and nature of spinal deformity noted in different conditions

Site of deformity	Nature of deformity	Condition
Cervical spine	Kyphosis	Larsen syndrome Diastrophic dysplasia
Thoracic spine	Scoliosis	Prader-Willi syndrome Infantile idiopathic scoliosis
	Kyphoscoliosis	Campomelic dysplasia
Thoracolumbar spine	Kyphosis	Achondroplasia Mucopolysaccharidoses Pseudoachondroplasia Ehlers-Danlos syndrome Marfan syndrome
	Scoliosis/kyphoscoliosis	Meningomyelocele
Lumbar and lumbosacral spine	Kyphosis	Meningomyelocele Ehlers-Danlos syndrome Marfan syndrome
	Lordosis	Meningomyelocele
	Scoliosis	Meningomyelocele
	Kyphoscoliosis	Meningomyelocele

dislocations should alert one to Larsen syndrome and cervical kyphosis.

In the legs, the finding of a tibial hemimelia should raise suspicion of congenital scoliosis. The finding of a club foot, particularly with a motor or sensory deficit, should drive a search for spinal dysraphism.

Is the baby in pain or respiratory distress?

Pain is a common feature of osteogenesis imperfecta. In addition, the presence of multiple rib fractures in the severely involved child can cause respiratory distress. Respiratory distress is the hallmark of Jarcho-Levin syndrome or thoracic insufficiency syndrome. Scoliosis and rib fusions are the findings in this syndrome. In children with tracheoesophageal fistula, rib fusions and congenital scoliosis are common. Not all rib fusions are due to the thoracotomy performed to repair the fistula.

Are there abnormalities of the skin over the spinal column?

Swelling, dimples, and hairy areas over the lumbosacral region are the typical findings in spinal dysraphism. Examination of the spine and spinal cord is imperative. Underlying congenital anomalies, including diastematomyelia, and anomalies or tethering of the spinal cord are common.

What is the site of the deformity? (See Table 10.1)

While congenital malformations of the spinal column can occur at any level, torticollis obviously can only occur in the neck. However, the typical site of the deformity seen in Larsen is in the neck, and the typical site for deformity seen in achondroplasia and mucopolysaccharidoses is at the thoracolumbar junction. The deformity in Prader-Willi syndrome, Coffin-Siris syndrome, and Williams syndrome is commonly in the thoracic spine.

The combination of a short neck, a low posterior hairline, and limitation of motion points to fusion anomalies of the cervical spine otherwise known as Klippel-Feil syndrome (Fig. 10.3) (Klippel and Feil 1975).

In the distal aspect of the trunk and pelvis, varying deformities are seen due to lumbosacral vertebral anomalies and varying degrees of sacral agenesis, the latter being associated with neurological deficit and the former noted by a pelvic obliquity as a baby or a lumbosacral kyphosis.

What is the plane of the deformity? (See Table 10.1)

Congenital abnormalities can produce deformities in any plane depending on which component of the vertebra has failed to develop and whether or not there are associated failures of segmentation. Failure of development of the vertebral body may produce only a mild deformity as in a butterfly vertebra but may be more severe with the absence of the vertebral body. The most severe deformity in regard to the

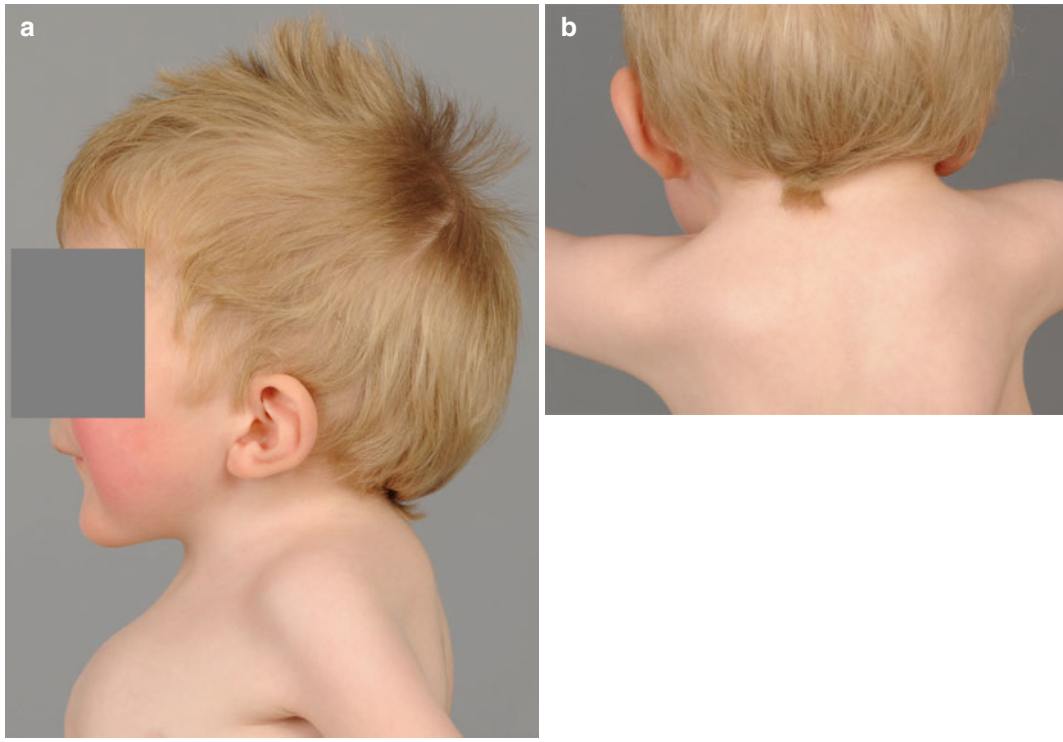

Fig. 10.3 (**a**, **b**) A low hairline and a short neck in a child with Klippel-Feil syndrome

spinal cord is where there is a congenital dislocation of the spine at the junction of two scoliotic curves with failure of formation of one half of the vertebra.

Kyphosis is the hallmark of spinal alignment in Larsen syndrome (Fig. 10.4), achondroplasia, and mucopolysaccharidoses, whereas failure of segmentation of the posterior elements produces a severely lordotic spine. Rotary deformity is most commonly seen in the cervical spine with torticollis and in failure of formation at the upper cervical spine. In campomelic dysplasia, the deformity is typically a kyphoscoliosis of the thoracic spine.

10.3 Physical Examination

10.3.1 Look

Look for abnormal shape and size of the skull. Look for abnormal development of the ears and mandible. Look for abnormal eye motion. Observe the alignment of the head and neck to the trunk. Look for neck length and the hairline. Look for features of disproportionate growth and the completeness of the extremities. Look at the trunk both anteriorly and posteriorly. Observe for any signs of spinal dysraphism and skin pigmentation.

10.3.2 Feel

Examine the extremities and note joint deformity and stability and absence of bones or digits. Feel the vertebral column and note deformity.

10.3.3 Move

Feel the degree of motion and any resistance to motion of the head and neck. Muscular torticollis will feel different to congenital scoliosis, and the motion in torticollis will be biased to one direction. Check the passive mobility of the spine. A normal child will have a very flexible thoracic

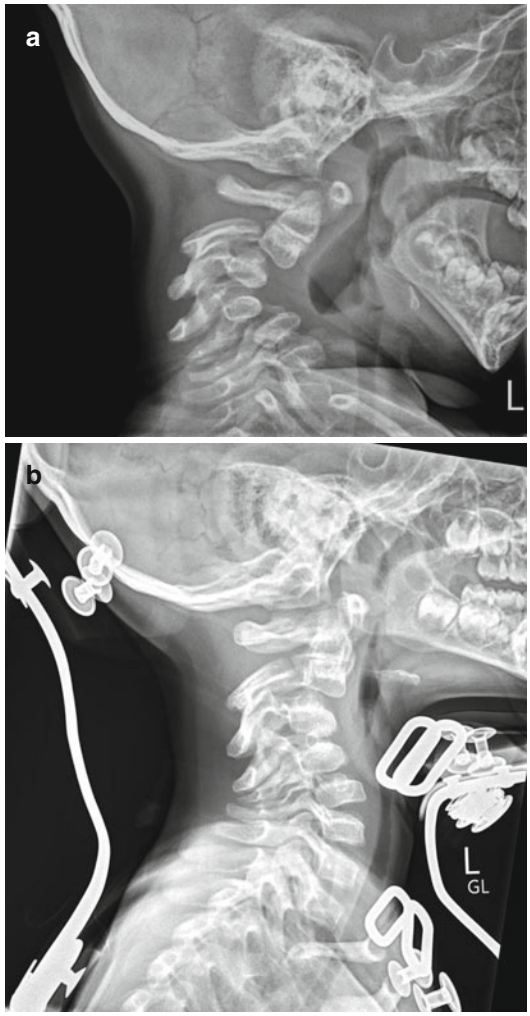

Fig. 10.4 Severe cervical kyphosis in an 18-month-old child with Larsen syndrome (**a**). After bracing for 3 years, the deformity has reduced (**b**)

and lumbar spine, whereas congenital anomalies will restrict motion and congenital fusions will obliterate motion.

10.3.4 Additional Tests

Note power and sensation of the extremities. Though an accurate estimate of muscle power and sensation may not be possible in the infant, the response to stimulation of the sole of the foot will give some idea of the sensation and motor power.

Drape the baby over the examining hand and see if the spinal deformity is corrigible.

10.4 Investigations to Confirm the Diagnosis

Plain Radiographs
Plain radiographs will provide most of the information required. A full-length supine AP radiograph of the spine will be reasonably reproducible for the first 2 years of life. Necessary measurements of idiopathic curves include the Cobb angle, the rib vertebral angle difference (RVAD), and the phase of the rib heads.

CT Scan
CT scans with 3D reconstructions can provide more defined information which is often helpful in planning surgery.

MRI Scan
MRI scans should be performed in congenital scoliosis. Approximately 20 % of cases will have an anomaly of the cord. In patients with a neurological deficit, this scan should be done without delay.

Ultrasound
A renal ultrasound examination should also be performed in congenital scoliosis as approximately 20 % of cases will have an abnormality in the genitourinary system.

10.5 Differential Diagnosis

10.5.1 Torticollis and Congenital Cervical Anomalies

The deformities of these groups are among the most obvious spinal deformities in the newborn. The attitude of the head in torticollis is typical.

The motion of the spine in torticollis is very much one sided. Congenital anomalies have a more global restriction. This part of the examination needs to be carefully performed as the proximal cervical spine provides a significant amount of movement. A tight sternocleidomastoid may be felt, and occasionally, a "tumor" is palpated within the muscle.

10.5.2 Infantile (Early Onset) Idiopathic Scoliosis

These children may have a deformity that is largely a packaging problem or a true idiopathic scoliosis that is progressive. The former will tend to be more corrigible and resolve with observation. The latter will have a RVAD greater than 20° and evidence of rotation in their radiographs, and the deformity will be progressive. Treatment with early casting is appropriate.

10.5.3 Syndromes Involving Congenital Anomalies and Other Stigmata

This group includes Goldenhar syndrome, VACTERL, Klippel-Feil, tracheoesophageal fistulae with spinal anomalies, and others. These groups are identified by their facial and other abnormalities. The behavior of the spinal deformity is not uniform, and all need to be observed and investigated for renal, cardiac, and spinal cord abnormalities.

10.5.4 Spinal Dysraphism/ Meningomyelocele

These newborns are immediately apparent and most diagnosed antenatally. Scoliosis in these children is common but not apparent early on. A kyphotic deformity, however, may benefit from early repair of the displaced erector spinae muscles at the time of closure of the meningocele.

10.5.5 Achondroplasia

The diagnosis is clinically evident. Two issues can arise in these babies; one is stenosis at the foramen magnum which places these babies at risk of spinal cord injury. The second is a kyphotic deformity in the lumbar spine that may require treatment in early childhood.

10.5.6 Osteogenesis Imperfecta

Spinal deformity is common in OI. However, in the newborn, these babies need to be protected from inadvertent injury to the limbs and spine. A carrying shell can be most helpful.

10.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis of the cause of a spinal deformity in the newborn and infant is shown in Table 10.2.

Table 10.2 Establishing the diagnosis of spinal deformity in the newborn

History					
–	–	Prenatal ultrasound may have identified vertebral deformity and anomaly	Prenatal ultrasound may have identified vertebral anomaly	–	Prenatal ultrasound may have identified limb deformities
History of maternal alcohol abuse may be present	–	–	–	–	–
Physical examination					
Deformity of the neck	Deformity of the thoracic spine	Deformity of the thoracic or lumbar spine	Deformity of the lumbar and lumbosacral spine	Deformity of the thoracic or thoracolumbar spine	Deformity of the thoracic and lumbar spine
Limitation of motion of the neck	–	–	–	–	–
Sternomastoid muscle may be contracted	–	–	–	–	–
Body proportions: Neck may be short with low hairline	Body proportions: Normal	Body proportions: Normal	Body proportions: Normal or trunk may be short	Body proportions: Altered	Body proportions: May appear altered
Normal muscle power	Normal muscle power	Normal muscle power	Weakness of the lower limbs	Normal muscle power	Difficult to assess on account of pain
No pain	No pain	No pain	No pain	No pain	Pain often present
–	–	Visceral anomalies may be evident (e.g., tracheoesophageal fistula)	–	–	–
–	–	Other limb anomalies (e.g., radial club hand) may be present	–	–	–
Working diagnosis: Torticollis/cervical vertebral anomaly	Working diagnosis: Infantile idiopathic scoliosis	Working diagnosis: Congenital scoliosis associated with syndromes with or without visceral anomalies	Working diagnosis: Spinal dysraphism/meningomyelocele	Working diagnosis: Skeletal dysplasia	Working diagnosis: Osteogenesis imperfecta

Investigations					
Plain radiograph: Congenital vertebral anomaly of the upper cervical spine may be present (failure of formation or segmentation)	Plain radiograph: Scoliosis without congenital vertebral anomaly	Plain radiograph: Scoliosis with congenital vertebral anomaly (failure of formation or segmentation)	Plain radiograph: Spina bifida and kyphosis/lordosis/scoliosis of lumbar spine	Plain radiograph: Features of specific skeletal dysplasia (e.g., achondroplasia)	Plain radiograph: Fractures of the long bones and scoliosis
Diagnosis					
Congenital torticollis (muscular or bony)	Infantile idiopathic scoliosis	Congenital scoliosis with or without syndromic association	Spinal dysraphism	Scoliosis or kyphosis associated with skeletal dysplasia	Scoliosis associated with osteogenesis imperfecta

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Absent Digits in the Hands or Feet

11

Benjamin Joseph

11.1 Introduction

Reduction in the normal number of digits of the hand or foot may occur as a consequence of failure of formation (agenesis) due to a definite genetic mutation or as a consequence of intrauterine amputation due to constriction by an amniotic band (Dy et al. 2014; Petit et al. 2014). Agenesis of the digits may represent a transverse terminal deficiency where fingers or toes alone are missing or the failure of formation of the digits may be part of a more extensive longitudinal deficiency; longitudinal deficiencies are far more common than transverse deficiencies (Makhoul et al. 2003). The number of missing digits does not necessarily reflect the degree of aplasia of the proximal bones (Petit et al. 2014; Del Campo et al. 1999) (Fig. 11.1). Often, more than one limb is affected, and the pattern of deficiencies may be similar in the affected limbs, or the patterns may be quite dissimilar (Ferda Percin and Yilmaz 2003).

Recognition of the nature and extent of deficiency has a direct bearing on future management.

11.2 Questions to Establish a Diagnosis

- **Which digits are missing?**
- **Is the absent digit (or digits) due to a constriction band or true deficiency due to failure of formation?**
- **If it is true agenesis, is the deficiency a transverse terminal deficiency or is there a longitudinal deficiency?**
- **If there is a longitudinal deficiency, is the ulna, radius, fibula, or tibia partially or totally absent?**
- **Are there deficiencies involving other limbs?**

Which digits are missing?

The deficiency in the hand may be radial (preaxial), ulnar (postaxial), or central. The pattern of missing digits may give a clue to the extent of deficiency in the limb. Absence of the

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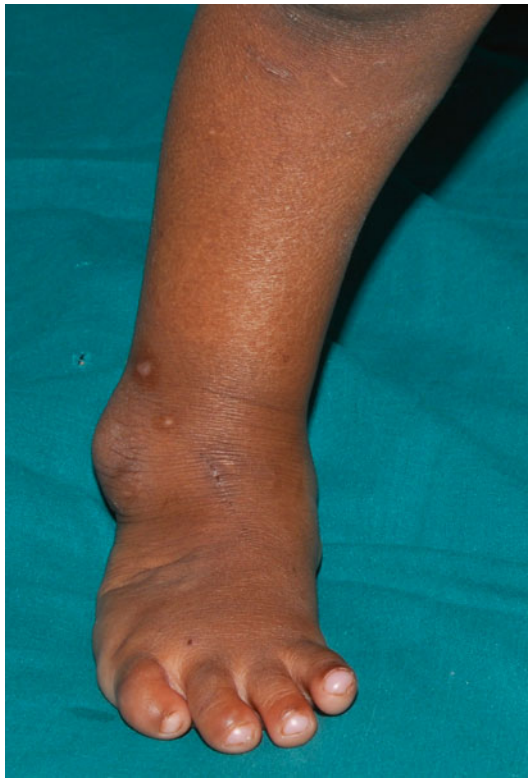

Fig. 11.1 Absence of the great toe in an infant with proximal focal femoral deficiency and partial tibial agenesis

thumb is frequently associated with absence of the radius (Inusha and Prasad 2008). However, it is important to be aware that occasionally the thumb may be absent in children with ulnar agenesis (Malik and Afzal 2013). In the foot, also the deficiency may be preaxial, postaxial, or central.

Is the absent digit (or digits) due to a constriction band or true deficiency due to failure of formation?

Constriction bands in the calf, fingers, and toes with asymmetric transverse deficiencies involving the hands and feet are characteristic of amniotic band syndrome (Al-Qattan 2000; Barros et al. 2014).

If it is true agenesis, is the deficiency a transverse terminal deficiency or is there a longitudinal deficiency?

While major degrees of aplasia of bones of the mesomelic segment (radius, ulna, fibula, and tibia) when associated with an absent digit will be clearly evident, minor degrees of hypoplasia may not be obvious.

If there is a longitudinal deficiency, is the ulna, radius, fibula, or tibia partially or totally absent?

Since treatment may differ profoundly, it is important that the extent of aplasia of the forearm or leg bones is clearly determined.

Are there deficiencies involving other limbs?

Quite frequently, the upper and lower limbs may show varying degrees of deficiency possibly as a result of a common insult to the developing limb buds early in embryonic development (Fuhrmann et al. 1980; Courtens et al. 2005).

11.3 Physical Examination

11.3.1 Look

If a toe or finger is missing, note the pattern of deficiency (preaxial, postaxial, or central). Examine all four limbs to confirm if the deficiency is limited to one hand or foot. Observe if the limb is short and if there are deformities of the proximal joints.

11.3.2 Feel

Palpate the forearm and leg and confirm if the radius, ulna, tibia, and fibula are all present.

11.3.3 Move

Attempt to move the knee and elbow and confirm the range of passive motion.

11.4 Investigations to Confirm the Diagnosis

Radiography

Plain radiographs of the affected limbs must be taken to determine the extent of deficiency.

Table 11.1 Syndromes associated with radial longitudinal deficiencies

Syndrome	Anomalies and hematological disorders	Frequency of association in children with radial deficiency (%)
Thrombocytopenia-absent radius (TAR) syndrome	Thrombocytopenia Radial club hand	15
VACTERAL association	Vertebral defects Anal atresia Cardiac malformations Tracheoesophageal fistula Tracheoesophageal atresia Renal anomalies Radial club hand	13
Holt-Oram syndrome	Atrial and ventricular septal defects Radial club hand	4
Fanconi anemia	Pancytopenia due to bone marrow failure	~1

Ultrasound
Abdominal ultrasound is necessary to exclude associated visceral anomalies.

Hematological Investigations
Platelet, white blood cell, and red blood cell counts are essential in children with a thumb deficiency.

11.5 Differential Diagnosis

11.5.1 Radial Ray Deficiency

Radial ray deficiency is very frequently associated with radial longitudinal deficiency or the radial club hand. Radial club hand occurs most often due to a spontaneous mutation though it may be inherited as a dominant or recessive trait. Radial club hand is frequently associated with serious abnormalities involving the hematological, cardiac, and renal systems, and it is important that the clinician is aware of these associations (Dy et al. 2014). The commonest syndrome of such associations is thrombocytopenia-absent radius (TAR) syndrome (Table 11.1).

In view of the frequency of visceral and systemic abnormalities associated with agenesis of the thumb, it is essential that every child with a hypoplastic or absent thumb is assessed to exclude these serious abnormalities.

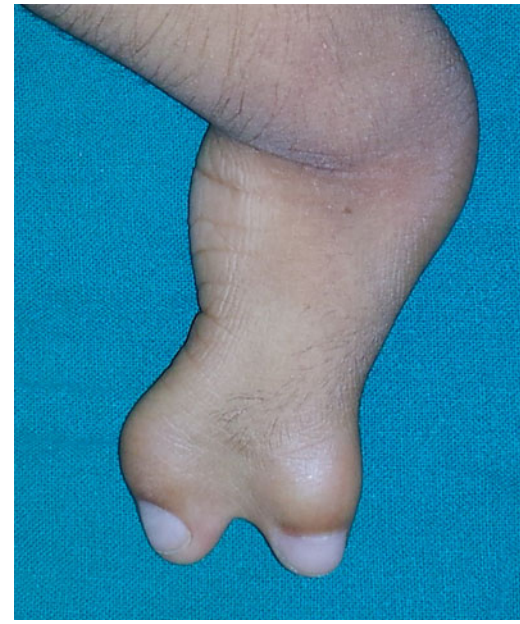

Fig. 11.2 Absent rays in a child with ulnar longitudinal deficiency

11.5.2 Ulnar Ray Deficiency

The medial rays may be absent in children with ulnar longitudinal deficiency (Fig. 11.2). Ulnar longitudinal deficiency is far less common than its radial counterpart, and it is not associated with serious or life-threatening visceral malformations or hematological disorders. Though

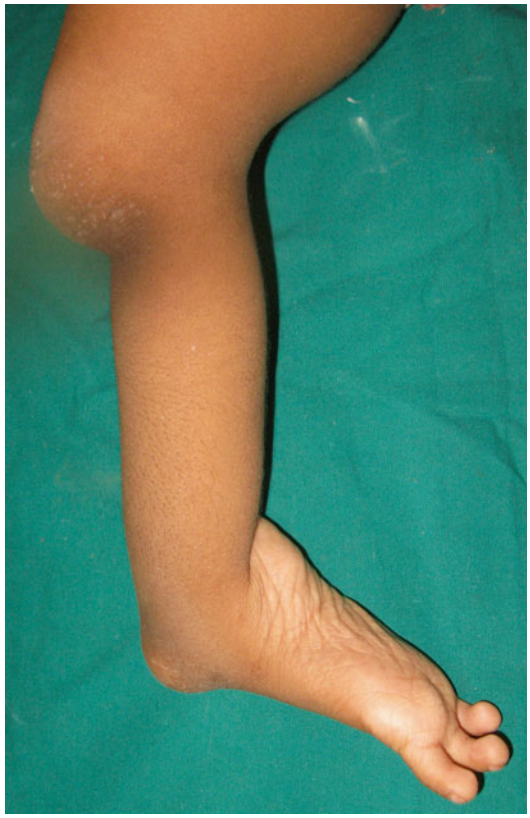

Fig. 11.3 Absent medial rays of the foot in a child with total agenesis of the tibia

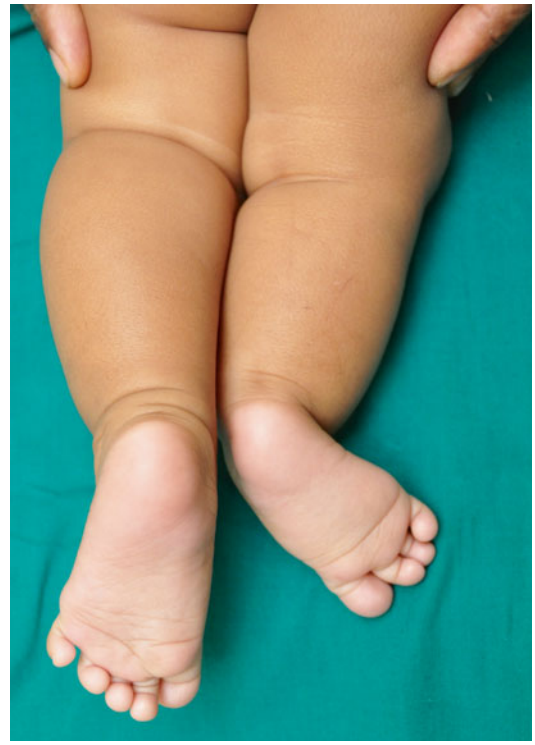

Fig. 11.4 Absent little toe in a child with fibular hemimelia

the ulna is the deficient bone in the limb, anomalies of the thumb are very frequent (almost as high as 90 %), and rarely, the thumb may even be absent (Malik and Afzal 2013; Dy et al. 2014). The entire limb is hypoplastic, and often the elbow is stiff due to synostosis of the radius and humerus.

11.5.3 Ray Deficiency with Tibial Hemimelia

Tibial hemimelia is often associated with hypoplasia or absence of the great toe particularly when the entire tibia is absent (Majewski et al. 1996) (Fig. 11.3).

11.5.4 Ray Deficiency with Fibular Hemimelia

The lateral rays of the foot are usually absent in children with fibular hemimelia (Fig. 11.4); the tibia is often short, and there may be some degree of anteromedial bowing of the tibia. The combination of fibular aplasia and tibial campomelia (bowing) with oligodactyly has been referred to as the FATCO syndrome (Karaman and Kahveci 2010; Courtens et al. 2005).

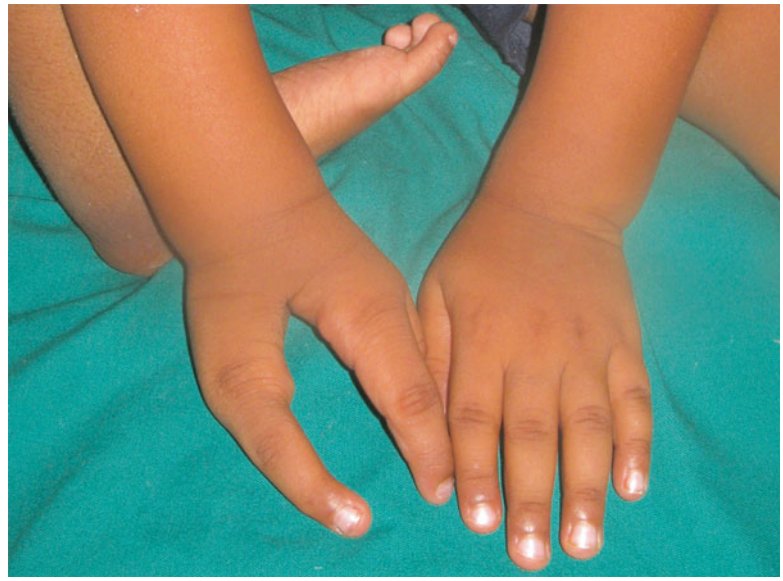

Fig. 11.5 Central ray deficiency involving one hand. The child also had complete tibial hemimelia in one leg

11.5.5 Central Ray Deficiency

Central ray deficiency manifests as the split hand or foot (Fig. 11.5); the anomaly is also referred to as ectrodactyly. The split hand or foot anomaly is often inherited as an autosomal dominant trait. As with other forms of digital aplasia, it may occur as an isolated anomaly or as part of a more extensive limb malformation syndrome (Majewski et al. 1996; Petit et al. 2014; Tos et al. 2011; Wada et al. 2013). Though typically the great toe is deficient in children with aplasia of the tibia, central ray deficiency has also been described (Majewski et al. 1996).

11.5.6 Terminal Transverse Deficiency

Terminal transverse deficiencies of the digits (adactyly and aphalangia) are rare (Makhoul et al. 2003) and more often occur as unilateral, isolated anomalies (Malik and Riaz 2012).

11.5.7 Amniotic Constriction Band

Fetuses exposed to misoprostol (prostaglandin E_1 analogue) in early pregnancy or chorionic villous sampling appear to be susceptible to developing amniotic band syndrome (McGuirk et al. 2001).

Amniotic bands can cause various types of defects in the hands and feet (Fig. 11.6) which include annular constrictions, amputations, acrosyndactyly, and clubfoot (Al-Qattan 2000; Barros et al. 2014; Goldfarb et al. 2009). Deficiencies in amniotic band syndrome are often seen in the terminal parts of the second and third or the third and fourth digits of the hand (McGuirk et al. 2001). More proximally located constriction bands may be present on the limb; these bands

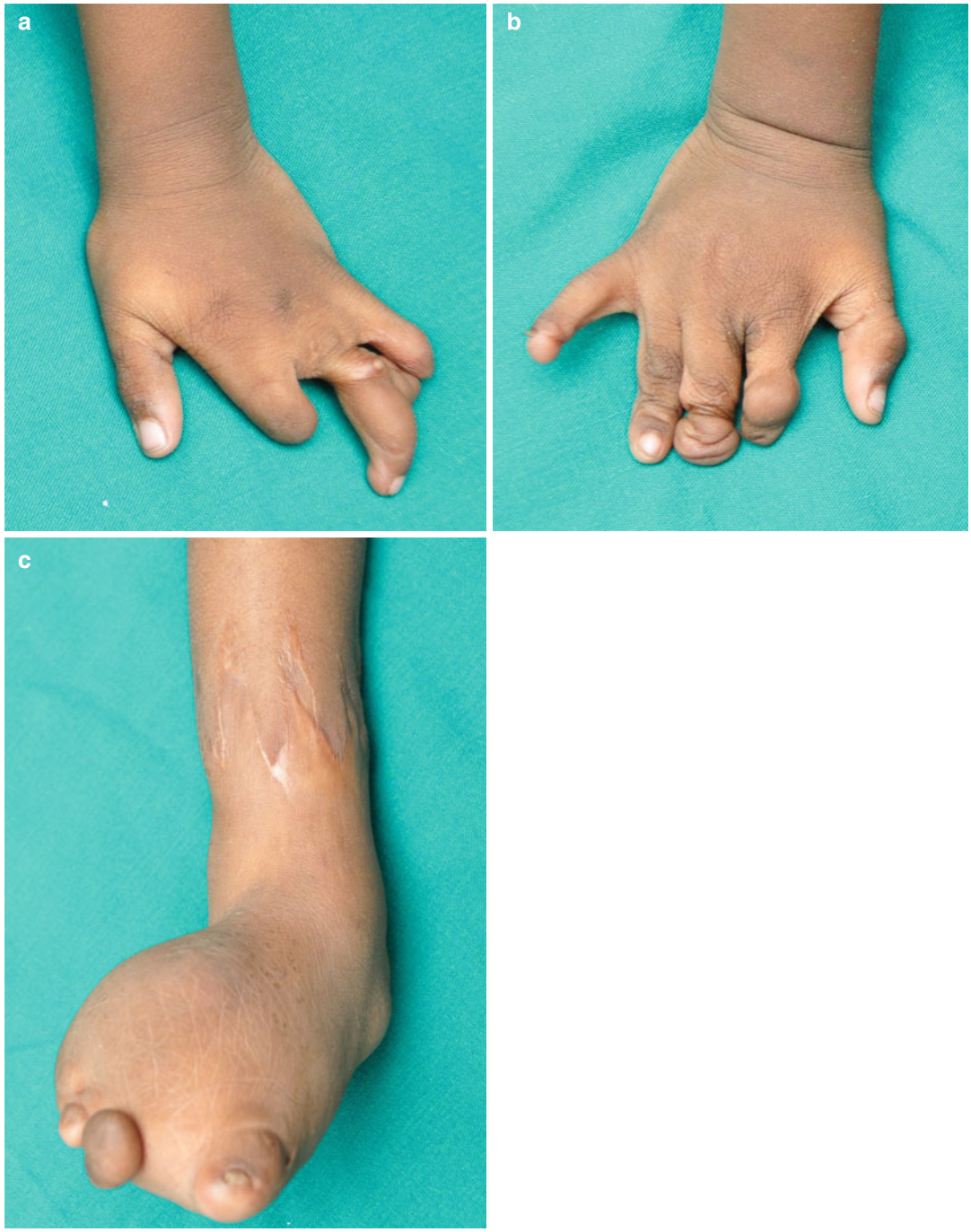

Fig. 11.6 Terminal deficiencies of the digits of the hands (**a**, **b**) and feet (**c**) in children with amniotic band syndrome. The scar of the Z-plasty done to release a constriction in the calf and the residual lymphedema on the dorsum of the foot are evident (**c**)

may be associated with complications such as distal edema (Fig. 11.6c), circulatory compromise, nerve damage, and even a pseudarthrosis of the underlying bone (Magee et al. 2007).

11.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Table 11.2.

Table 11.2 Establishing the diagnosis of the type of anomaly in a child with absent fingers or toes

History			
Family history seldom present History of illness or substance abuse in mother during pregnancy may be present	Family history may be present History of illness or substance abuse in mother during pregnancy may be present	Family history may be present History of illness or substance abuse in mother during pregnancy may be present	Family history not present History of exposure to misoprostol in early pregnancy may be present
–	–	–	History of chorionic villous sampling being performed may be present
No symptoms related to the upper or lower GI tract	No symptoms related to the upper or lower GI tract	Symptoms related to the upper or lower GI tract may be present	No symptoms related to the upper or lower GI tract
Physical examination			
Absent finger(s) or toe(s)	Absent finger(s) or toe(s)	Absent thumb	Absent finger(s) or toe(s)
Pattern of absence of digits: Transverse terminal deficiency involving the phalanx (or entire digit) of one or more digits	Pattern of absence of digits: Longitudinal deficiency	Pattern of absence of digits: Longitudinal deficiency involving the thumb	Pattern of absence of digits: Transverse terminal deficiency often affecting digits 2, 3, or 4
No constriction bands	No constriction bands	No constriction bands	Constriction bands present
Bones of the forearm or leg: Not deficient	Bones of the forearm or leg: Deficiency of the radius or ulna in the forearm or tibia or fibula in the leg	Bones of the forearm or leg: Deficiency of the radius	Bones of the forearm or leg: Not deficient
Working diagnosis: Isolated transverse deficiency	Working diagnosis: Longitudinal deficiency without systemic or visceral involvement	Working diagnosis: Longitudinal deficiency with systemic or visceral abnormalities	Working diagnosis: Deficiency in amniotic constriction band syndrome
Investigations			
Plain radiographs of the limb: No abnormality other than the localized terminal deficiencies	Plain radiographs of the limb: Longitudinal deficiency affecting the proximal segment of the limb present	Plain radiographs of the limb: Partial or total radial aplasia will be present	Plain radiographs of the limb: Often no bony abnormality beyond the terminal deficiency Rarely, constriction band in the calf may be associated with a pseudarthrosis of the tibia

(continued)

Table 11.2 (continued)

Ultrasound of the abdomen: No visceral abnormality	Ultrasound of the abdomen: No visceral abnormality	Ultrasound of the abdomen: Visceral abnormality involving the kidneys often present	Ultrasound of the abdomen: Not indicated
–	–	Echocardiogram: May demonstrate atrial or ventricular septal defects	–
–	–	Hematological investigations: May show features of anemia, thrombocytopenia, or pancytopenia	–
Diagnosis			
Isolated transverse terminal deficiency involving the digits of the hand or foot	Longitudinal deficiency of the limb (of which digital aplasia is a part) without visceral or systemic abnormality	Longitudinal deficiency of the radius (of which thumb aplasia is a part) with visceral or hematological abnormality	Transverse deficiency associated with amniotic constriction band syndrome

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Duplication of Fingers

12

Benjamin Joseph

12.1 Introduction

Duplication of fingers or polydactyly may occur on the preaxial (radial) or postaxial (ulnar) side; central polydactyly is rare. The proximal extent of duplication varies a great deal; attempts have been made to classify the patterns of duplication comprehensively (Blauth and Olason 1988; Wassel 1969). Polydactyly may be associated with other anomalies of the hand, the commonest of which is syndactyly (polysyndactyly). Children with polydactyly of the hands may have comparable patterns of polydactyly of the feet also (Miura et al. 1987). It is also important to recognize that polydactyly may just be part of a more generalized malformation of the limb and in some instances part of a genetically distinct syndrome with multisystem involvement (Schwabe and Mundlos 2004).

12.2 Questions to Establish a Diagnosis

- **Is the supernumerary finger preaxial, central, or postaxial in location?**
- **What is the proximal extent of duplication?**
- **Is the duplication present in only one limb or is it bilateral and symmetric?**
- **Are there associated limb anomalies?**
- **Are there extra-skeletal anomalies or defects?**
- **Is the duplication an isolated anomaly or is it associated with a syndrome?**

Is the supernumerary finger preaxial, central, or postaxial in location?

Racial differences in the frequency of these anomalies have been noted; preaxial polydactyly is more common in white races, while postaxial polydactyly is far more common in colored races (Kozin 2003). Certain syndromes have a predilection for preaxial polydactyly, while others tend to have associated postaxial polydactyly.

What is the proximal extent of duplication?

The proximal extent of duplication will influence the surgical approach, and hence it is important that this is identified.

Is the duplication present in only one limb or is it bilateral and symmetric?

Bilateral symmetric duplication is more likely to be associated with a syndrome.

Are there associated limb anomalies?

Associated limb anomalies again will influence the treatment.

Are there extra-skeletal anomalies or defects?

If extra-skeletal anomalies are present, it is more likely that the polydactyly is part of a syndrome.

Is the duplication an isolated anomaly or is it associated with a syndrome?

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Table 12.1 Syndromes associated with different patterns of polydactyly

Preaxial (radial) polydactyly	Postaxial (ulnar) polydactyly	Central polydactyly
Acrocephalopolysyndactyly syndrome Townes-Brocks syndrome	Ellis-van Creveld syndrome (chondroectodermal dysplasia) Smith-Lemli-Opitz syndrome McKusick-Kaufman syndrome Bardet-Biedl syndrome	Pallister-Hall syndrome
Greig cephalopolysyndactyly syndrome Orofaciodigital syndrome Short rib-polydactyly syndrome		

Several syndromes are associated with polydactyly of the hand, and the pattern of duplication may vary with the underlying syndrome (Schwabe and Mundlos 2004) (Table 12.1).

12.3 Physical Examination

12.3.1 Look

Look for the location of the duplication (ulnar or radial side). Note if other anomalies of the upper and lower limb are present. Observe if there is evidence of facial dysmorphism. Inspect the mouth, and note if the palate is high arched or cleft.

12.3.2 Feel

Palpate the hand, and attempt to identify the proximal extent of the skeletal elements of the duplicated digit.

12.3.3 Move

Ensure that the individual joints of the digits are mobile.

12.4 Investigations to Confirm the Diagnosis

Radiography
Plain radiographs are essential to determine the proximal extent of duplication and if there are associated skeletal anomalies.

12.5 Differential Diagnosis

12.5.1 Preaxial Polydactyly

Preaxial polydactyly is a genetically heterogeneous group. The commonest type is duplication of the biphalangeal thumb. This form of thumb duplication is frequently sporadic, unilateral, and not associated with any syndrome, and treatment is limited to the functional and cosmetic problems associated with the thumb alone. The second type is duplication of a triphalangeal thumb. The third type is polydactyly of the index finger which replaces the normal thumb. Yet, another form of preaxial polydactyly is associated with syndactyly of the third and fourth finger. Preaxial mirror polydactyly is a rare form which may be associated with ulnar dimelia and mirror polydactyly of the feet.

Duplication of the Biphalangeal Thumb
Thumb duplication varies with regard to the proximal extent of duplication, and the anomaly has been classified on the basis of this (Wassel 1969). Seven types have been identified; the commonest is type IV (~50 % of cases) where there is complete duplication of the distal and proximal phalanges (Wassel 1969; Al-Qattan 2010) (Fig. 12.1).

12.5.2 Postaxial Polydactyly

Postaxial polydactyly is frequently inherited as an autosomal dominant trait (Schwabe and Mundlos 2004). Two distinct types of postaxial polydactyly have been identified; type A is a well-developed digit articulating with the fifth metacarpal (Fig. 12.2) or a supernumerary metacarpal, while type B is a rudimentary digit. Postaxial polydac-

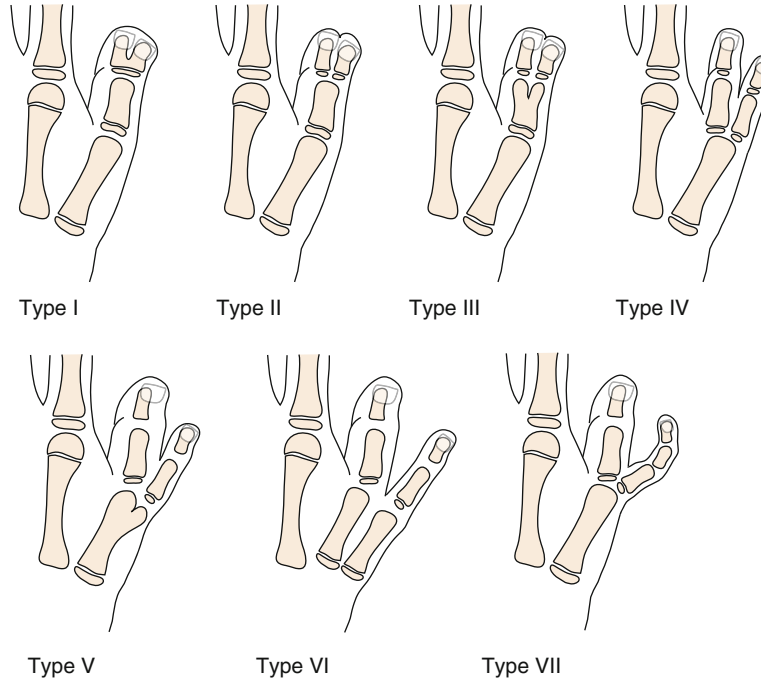

Fig. 12.1 Diagram showing the patterns of thumb duplication

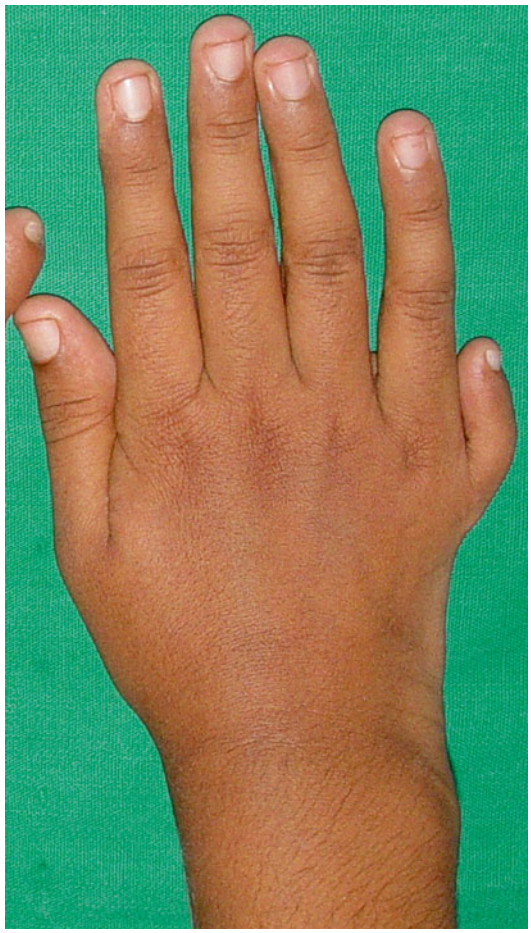

Fig. 12.2 Postaxial polydactyly

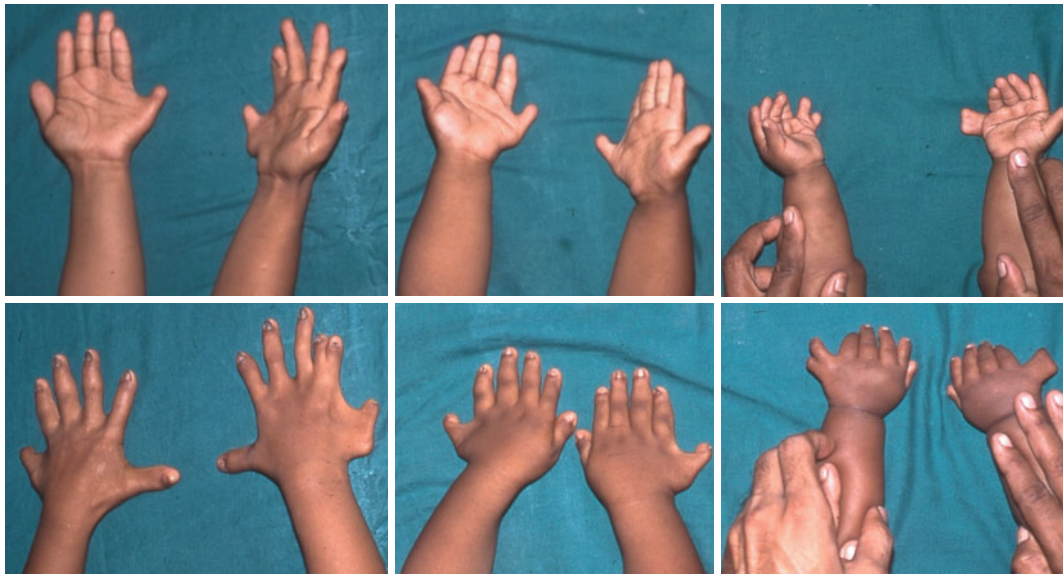

Fig. 12.3 Dorsal and volar views of the hands of three siblings with postaxial polydactyly associated with Ellis-van Creveld syndrome

tyly is one of the characteristic features of Ellis-van Creveld syndrome (Fig. 12.3); other anomalies include dystrophic nails, abnormal dentition and congenital cardiac abnormalities.

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Duplication of Toes

13

Benjamin Joseph

13.1 Introduction

Duplication of one or more toes may occur as an isolated anomaly (Bromley et al. 2000) or in association with other skeletal anomalies or be part of a generalized syndrome (Castilla et al. 1998). In 85 % of instances, polydactyly is an isolated anomaly, while the remaining 15 % are associated with other congenital anomalies (Castilla et al. 1998). As many as 310 clinical entities associated with polydactyly have been identified, and 80 mutations in 90 genes have been implicated in the causation of syndromes with this anomaly (Biesecker 2011). This emphasizes the need for the clinician to be aware that a newborn infant with polydactyly must be evaluated to exclude the more serious of the numerous syndromes associated with this seemingly trivial anomaly. The location, number, pattern, and symmetry of the duplicated toes may help in differentiating some of these conditions.

13.2 Questions to Establish a Diagnosis

- **Is the supernumerary toe preaxial, central, or postaxial?**
- **What is the proximal extent of the duplication?**
- **Is the duplication present in only one limb or is it bilateral and symmetrical?**
- **Are there associated limb anomalies?**
  - **Is the rest of the foot normal?**
  - **Are there deformities, deficiencies, or duplication of the bones of the leg?**
- **Are there extra-skeletal anomalies or defects?**

Is the supernumerary toe preaxial, central, or postaxial?

The duplication may be preaxial (duplicated first digit), central, or postaxial (duplicated fifth digit); postaxial duplication is the commonest form of toe duplication. Preaxial polydactyly of the toes is more frequently encountered (Fig. 13.1) than central duplication which is rare.

What is the proximal extent of the duplication?

Patterns of toe duplication were documented and comprehensively classified (Blauth and Olason 1988; Watanabe et al. 1992). The authors recognized that the duplication may only involve the terminal phalanges in some instances, while the more proximal bony structures of the affected ray (middle or proximal phalanx, metatarsals, and tarsals) are also often duplicated (Fig. 13.2). It is important to establish the proximal extent of duplication to plan appropriate treatment.

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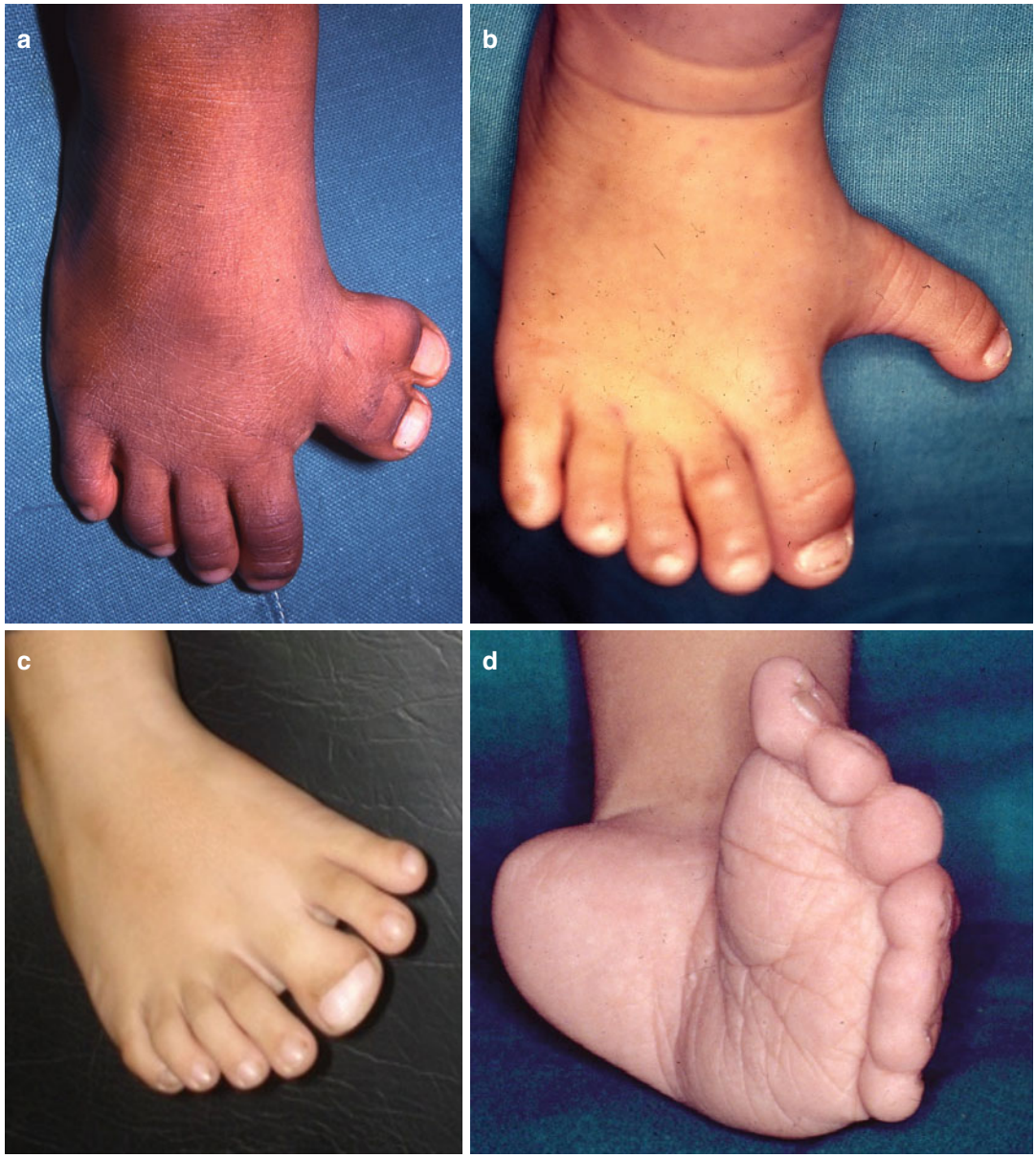

Fig. 13.1 Examples of preaxial polydactyly (**a**–**d**). The great toe may be duplicated with associated hallux varus (**a**). The supernumerary toe may have characteristics of a lesser toe (**b**) or there may be mirror duplication (**c**, **d**)

Is the duplication present in only one limb or is it bilateral and symmetrical?

The duplication may be bilateral and symmetrical and may also involve the hands (Fig. 13.3).

Are there associated limb anomalies?

The frequency of isolated polydactyly (i.e., without any associated limb or visceral anomaly) varies with the location of duplication (Castilla et al. 1998). The proportion of children with associated anomalies in addition to polydactyly is least with postaxial polydactyly (duplication of the fifth digit; 11.8 %), twice as frequent with preaxial polydactyly (duplicated biphalangeal first digit; 20 %), and five times as frequent with rarer forms of polydactyly (including central polydactyly, triphalangeal

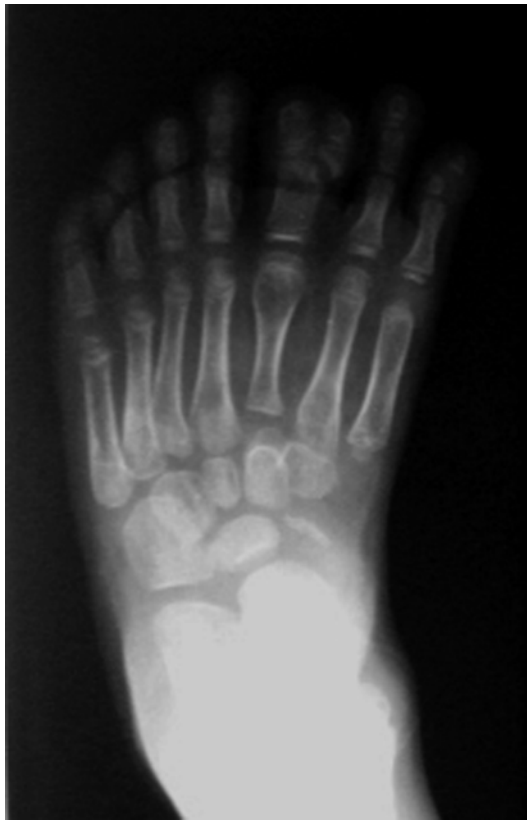

Fig. 13.2 Radiograph of the foot shown in Fig. 4.7b shows that the duplication of the great toe only involves the terminal phalanx. One of the two preaxial supernumerary toes shows duplication up to the respective tarsal bones (Courtesy: Dr.Ranjit Deshmukh, Pune, India)

thumb, and preaxial mirror polydactyly 54.9 %). This knowledge should warn the clinician to anticipate other anomalies frequently in children with rare forms of polydactyly.

Are there extra-skeletal anomalies or defects?

Presence of extra-skeletal anomalies suggests that the polydactyly is part of a syndrome.

13.3 Examination

Systematic examination of the foot, leg, and thigh should be done to identify associated skeletal anomalies.

A careful examination of the head, face, eyes, mouth, palate, neck, chest, heart, abdomen, rectum, genitalia, hair, and nails is essential to exclude anomalies in each of these regions that may point to a specific syndrome. In addition to the initial examination of the newborn, these children need to be followed up into childhood as some features such as intellectual impairment would only manifest in early childhood. Referral to a pediatrician is needed for the initial evaluation and follow-up.

13.4 Differential Diagnosis

13.4.1 Isolated Polydactyly

Isolated polydactyly is usually postaxial in location; it may be unilateral or bilateral.

13.4.2 Polydactyly Associated with Other Anomalies of the Limb

Syndactyly

Syndactyly is the commonest anomaly seen in association with polydactyly; this may not result in major disability in the foot.

Hallux Varus

Hallux varus is commonly seen in association with preaxial polydactyly (Belthur et al. 2011) (Fig. 13.1a). The first metatarsal may be short and dysplastic if hallux varus is present (Joseph et al. 1984, 1987). Surgery is likely to be needed in early childhood.

Equinovarus Deformity

A severe equinovarus deformity may indicate a partial or complete tibial deficiency (Verghese et al. 2007) (Figs. 13.4 and 13.5).

Tibial Deformity or Deficiency

Anterolateral bowing of the tibia has been described in association with preaxial polydactyly (Bressers and Castelein 2001) (see Chap. 4). The tibia may be deficient in children with mirror polydactyly; the extent of tibial deficiency may vary from mild hypoplasia to total absence (Verghese et al. 2007) (Fig. 13.4).

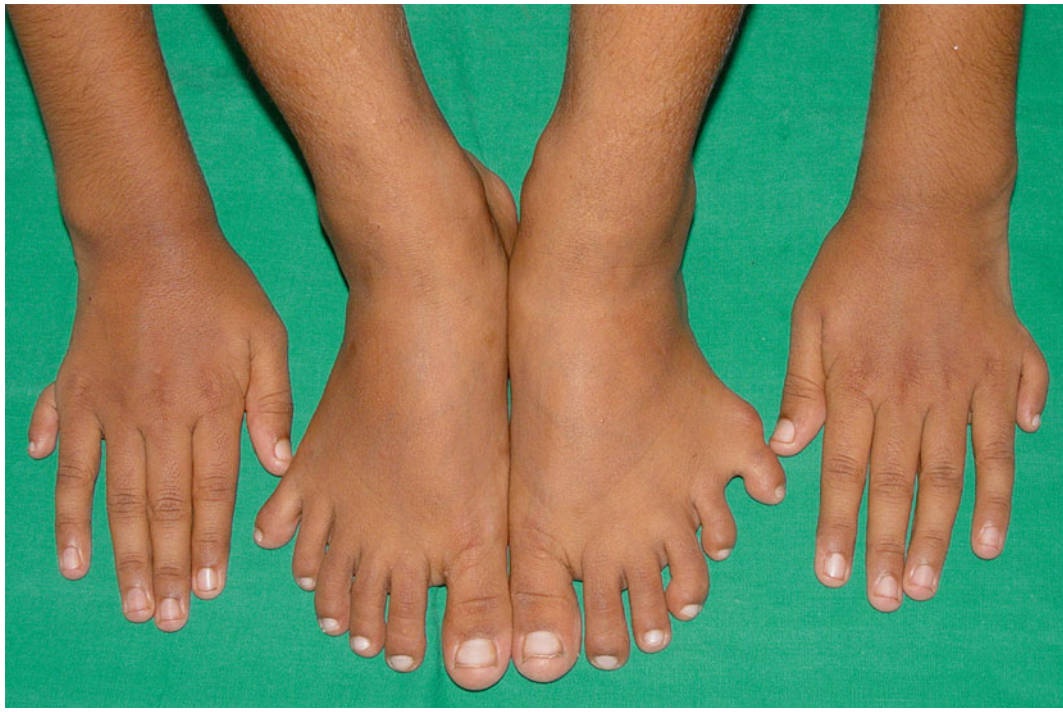

Fig. 13.3 Postaxial polydactyly of the feet and hands

Fibular Dimelia

Duplication of the fibula (fibular dimelia) may also be seen in association with preaxial mirror toe duplication and complete tibial agenesis (Fig. 13.5a) (Verghese et al. 2007; Bayram et al. 1996; Ganey et al. 2000). The duplication of the skeleton in the foot extends proximally up to the calcaneus, and this is seen as a double heel (Fig. 13.5b). Children with this unusual anomaly may have identical anomalies of the upper limbs with ulnar dimelia and mirror hands. This pattern of upper and lower limb anomalies with associated nasal defects is known as the Laurin-Sandrow syndrome (Sandrow et al. 1970).

13.4.3 Polydactyly Associated with a Syndrome

Several syndromes are associated with polydactyly of the foot; some of these are listed in Tables 13.2, 13.3, and 13.4.

13.5 Establishing the Diagnosis

An outline of process of establishing the diagnosis of the condition associated with polydactyly of the toes in the newborn is presented in Table 13.1. A plain radiograph of the affected limb would be needed to formulate a working diagnosis.

A definitive diagnosis may be made in several of the syndromic conditions only after a detailed evaluation of the child with appropriate imaging and other diagnostic investigations pertaining to the system that appears to be affected on clinical examination. Genetic studies may also be needed to confirm the diagnosis. If it is established that the polydactyly is isolated, treatment of polydactyly can be planned (Al-Qattan et al. 2002; Giorgini and Aquino 1984; Morley and Smith 2001).

Three classes of syndromes are associated with polydactyly: those with limb anomalies warranting major orthopedic intervention apart

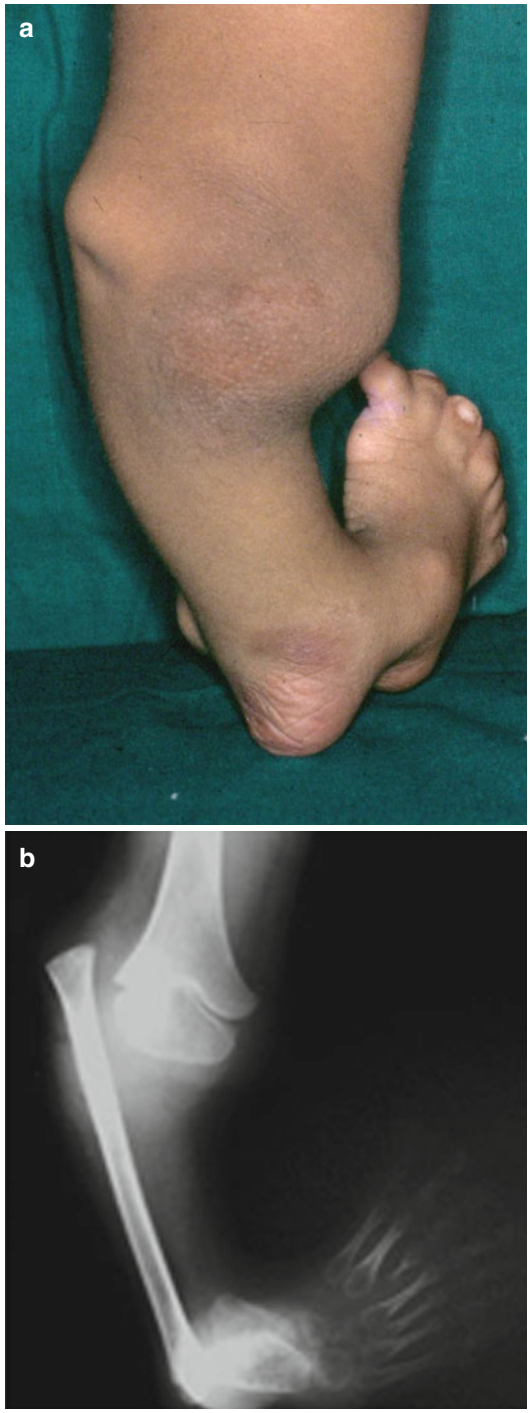

Fig. 13.4 Severe equinovarus deformity with complete absence of the tibia seen in a child with preaxial mirror polydactyly

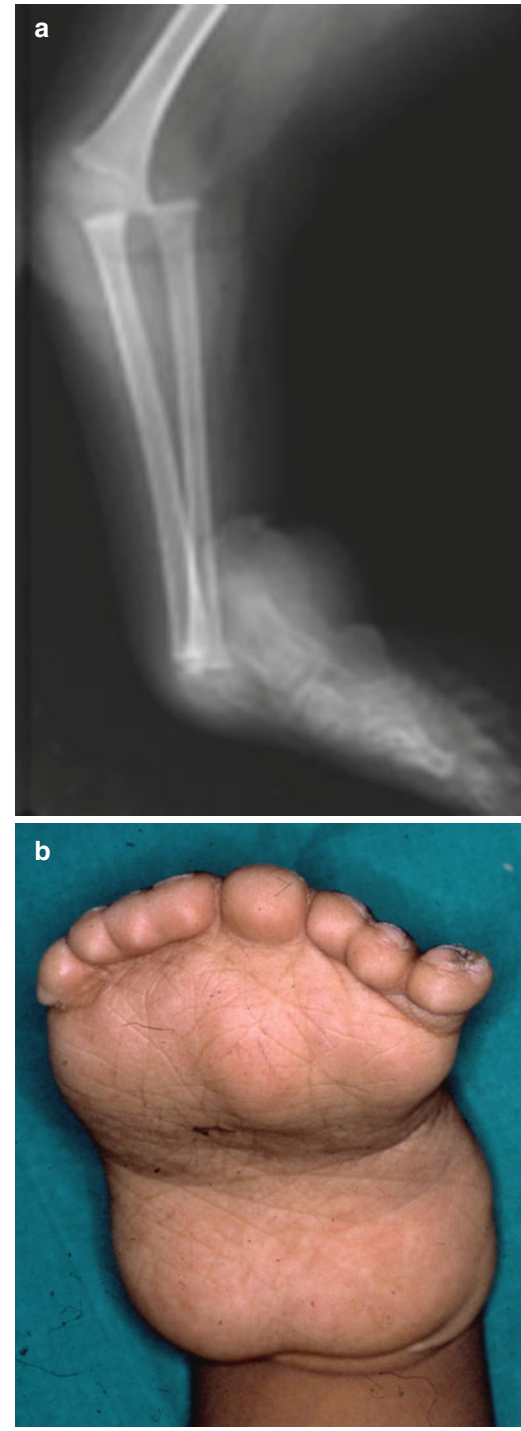

Fig. 13.5 Fibular dimelia, total agenesis of the tibia, and preaxial mirror polydactyly. The ulna was not duplicated, and there were no facial abnormalities

Table 13.1 Features facilitating the diagnosis of conditions associated with polydactyly of the toes

	Isolated polydactyly	Preaxial polydactyly with anterolateral bowing of the tibia (Bressers and Castelein 2001)	Spectrum of mirror foot and tibial aplasia (Verghese et al. 2007)			Polydactyly as minor part of syndrome
Position of the supernumerary toe	Postaxial or preaxial (occasionally central)	Preaxial	Preaxial mirror polydactyly			Preaxial/postaxial or central
Proximal extent of duplication[a]	Variable	Proximal phalanx	Tarsal (talar duplication)	Metatarsals	Tarsals including calcaneum	Variable
Associated limb anomalies or deformities	NIL	Anterolateral bowing of the tibia	Equinovarus foot Tibial dysplasia	Equinovarus foot Total tibial agenesis	Equinovarus with double heel Total tibial agenesis Fibular dimelia	Variable
Bilaterality and upper limb involvement	May be bilateral Upper limbs may be affected	May be bilateral Upper limbs normal	May be bilateral Upper limbs normal	May be bilateral Upper limbs normal	May be bilateral Upper limbs may show identical anomalies	Variable
Presence of extra-skeletal anomalies	NIL	NIL	NIL	NIL	Anomaly of the nose may be present[b]	YES

[a]Determined from plain radiographs

[b]Mirror polysyndactyly of the upper and lower limbs with anomaly of the nose is the Laurin-Sandrow syndrome

Table 13.2 Examples of some syndromes with polydactyly that are likely to require orthopedic intervention (apart from dealing with the polydactyly)

Syndrome/reference	Major characteristic features	Nature of orthopedic intervention likely to be needed
Fuhrmann et al. (1980)	*Polysyndactyly* Hypoplasia of the fibula and pelvis, bowing of the femur	Correction of femoral bowing, correction of foot and ankle deformities that may occur with fibular hypoplasia
Ellis-van Creveld syndrome (Goldblatt et al. 1992)	*Postaxial polydactyly* Sparse hair, abnormal nails and teeth, and congenital cardiac anomalies	Correction of genu valgum that develops later on in childhood in many children with this syndrome (the cardiac anomaly would need attention much earlier)
Lamb et al. (1983)	*Preaxial polydactyly* Five-fingered hand, partial or complete tibial absence	Pollicization, correction of problems associated with tibial agenesis
Borg et al. (1999)	*Preaxial mirror polydactyly* Unilateral tibial hypoplasia	Management of limb length inequality
Agarwal et al. (1996)	*Polysyndactyly* Five-fingered hand, tibial agenesis, agenesis of the lower end of the radius	Pollicization, correction of problems associated with tibial and radial agenesis
Laurin-Sandrow syndrome (Sandrow et al. 1970; Pilkington et al. 2000)	*Preaxial mirror polydactyly of feet and hands* Ulnar dimelia, fibular dimelia, agenesis of radius and tibia, nasal abnormality	Improving function of the hands and lower limbs

Table 13.3 Examples of syndromes with polydactyly as a minor component of the syndrome

Syndrome/reference	Characteristic features
Trisomy 13	*Postaxial polydactyly* Structural anomalies of the brain, cleft lip and palate, cardiac defects, renal abnormalities, omphalocele, undescended testes in boys, and bicornuate uterus in girls, congenital vertical talus
Acrocallosal syndrome (Aykut et al. 2008)	*Polydactyly of fingers and toes* Mental retardation, agenesis of the corpus callosum; Prominent forehead, broad nasal bridge, short nose and mandible, hypertelorism, epicanthic folds, large anterior fontanelle and tapered fingers, omphalocele, and inguinal hernia are some other common findings
Greig cephalopolysyndactyly (Debeer et al. 2007)	*Preaxial or postaxial polysyndactyly* Craniofacial abnormalities
WAGR (Bremond-Gignac et al. 2005)	*Preaxial polydactyly* *W*ilms' tumor, *a*niridia, *g*enitourinary abnormalities, and *g*rowth and mental *r*etardation (*WAGR*)
Lawrence-Moon-Bardet-Biedl syndrome (Castle et al. 1993; Gershoni-Baruch et al. 1992)	*Postaxial or central polydactyly* Rod–cone dystrophy with childhood-onset visual loss, truncal obesity, learning disabilities, male hypogenitalism and complex female genitourinary malformations, and renal dysfunction
Orofacial digital syndrome (Doss et al. 1998)	*Postaxial polydactyly* Oral (cleft palate, bifid tongue), dental, and facial abnormalities, with various associated anomalies of other organs depending on the type (at least 13 types have been described)
McKusick-Kaufmann syndrome	*Polydactyly* Hydrometrocolpos, congenital cardiac defects

Table 13.4 Some lethal syndromes which include polydactyly as one of the characteristic features

Syndrome	Characteristic features
Hydrolethalus syndrome	*Polydactyly* Hydrocephalus, micrognathia, cardiac and respiratory anomalies
Short rib, polydactyly syndrome – Majewski type	*Polydactyly* Short ribs, lung hypoplasia, dwarfism
Meckel syndrome	*Postaxial polydactyly* Occipital encephalocele, renal cystic dysplasia, hepatic ductal proliferation, fibrosis, and cysts
Hall syndrome	*Postaxial polydactyly* Hypothalamic hamartoblastoma, hypopituitarism, imperforate anus

from dealing with the polydactyly (Table 13.2), those with anomalies of important visceral organs of which the treatment take precedence over the limb anomalies (Table 13.3), and a group of lethal syndromes (Table 13.4).

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Deformities of Fingers and Toes in the Newborn

14

Benjamin Joseph

14.1 Introduction

Congenital malformations of fingers and toes include duplication, failure of differentiation (syndactyly and symphalangism), brachydactyly, deficiencies, and deformities. Deficiencies of digits have been discussed in Chap. 11, and duplication of fingers and toes has been discussed in Chaps. 12 and 13; other common malformations are discussed in this chapter. Most of these anomalies can be diagnosed easily on the basis of clinical examination and plain radiographs. The few situations where diagnosis may be confusing will be highlighted. Characteristic finger or toe deformities that aid the diagnosis of a generalized syndrome will be emphasized.

14.2 Questions to Establish a Diagnosis

- **Is the anomaly of the hand or foot a failure of differentiation, deficiency, brachydactyly, or a deformity?**
- **Is the deformity an important diagnostic criterion of a generalized condition?**

Is the anomaly of the hand or foot a failure of differentiation, deficiency, brachydactyly, or a deformity?

The exact nature of the anomaly needs to be identified. This will facilitate planning for treatment if it is required.

Is the deformity an important diagnostic criterion of a generalized condition?

Some deformities of the hand or foot are pathognomonic of generalized syndromes, and recognition of these deformities can facilitate the diagnosis of the syndrome.

14.3 Physical Examination

14.3.1 Look

Note the number of toes and fingers, and note their relative lengths. Look at the interdigital web spaces, and note if they are normal. Look at the alignment of the digits. Identify the location and plane of deformities that are present in the fingers and toes. Note if the nails are well formed or if they are dysplastic. Observe if the flexor creases over the interphalangeal joints are present. Note if the infant forms a fist normally.

14.3.2 Feel

Palpate the intermetacarpal and interdigital spaces to exclude bony fusion of the adjacent metacarpals or metatarsals and phalanges.

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14.3.3 Move

Attempt to move the metacarpophalangeal joints and the interphalangeal joints of each of the digits.

Abduct the thumb, and note if the first web space is normal or contracted.

14.4 Investigations to Confirm the Diagnosis

14.4.1 Radiography

Plain radiographs are necessary to determine if the skeleton of the hand and foot is normal or malformed.

14.5 Differential Diagnosis: The Fingers and Thumb

Malformation of the digits includes syndactyly, brachydactyly, polydactyly, symphalangism, agenesis, and a combination of these anomalies (Fig. 14.1). Different patterns of these anomalies have been described and classified (Fig. 14.2).

14.5.1 Syndactyly

Syndactyly, a common anomaly, is an abnormal interconnection between adjacent digits that may be an isolated anomaly or part of a syndrome (Entin 1976; Malik 2012). The interconnection may be

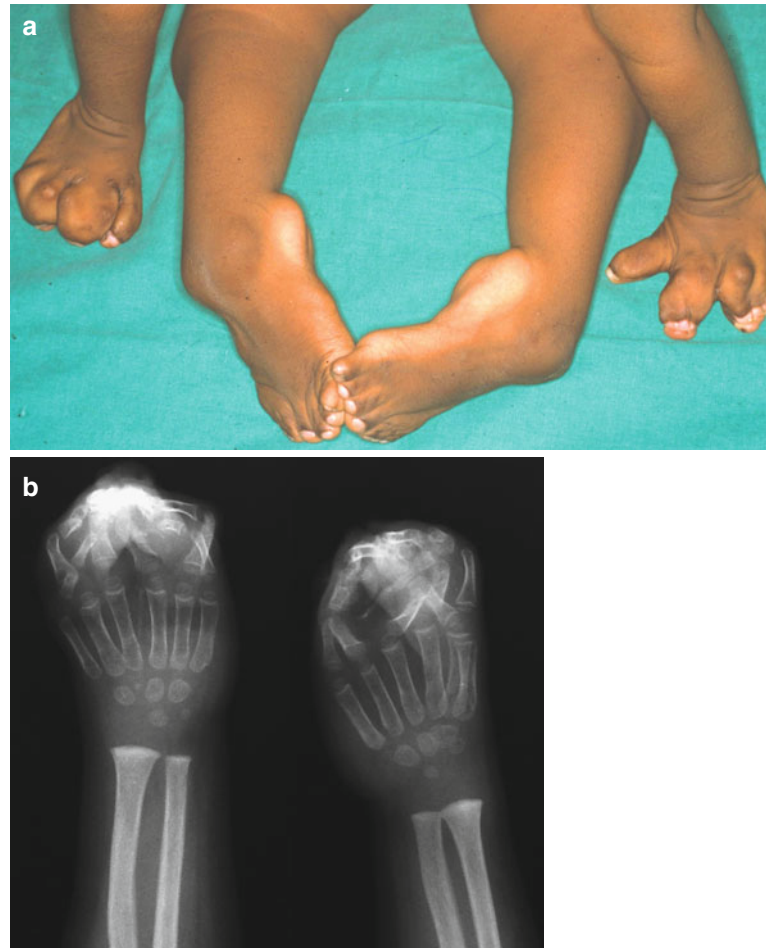


Fig. 14.1 Anomalies of the hand and feet in a child with fibular dimelia, syndactyly, polydactyly, and brachydactyly, are all present (**a**); polysyndactyly involving both hands in another child (**b**)

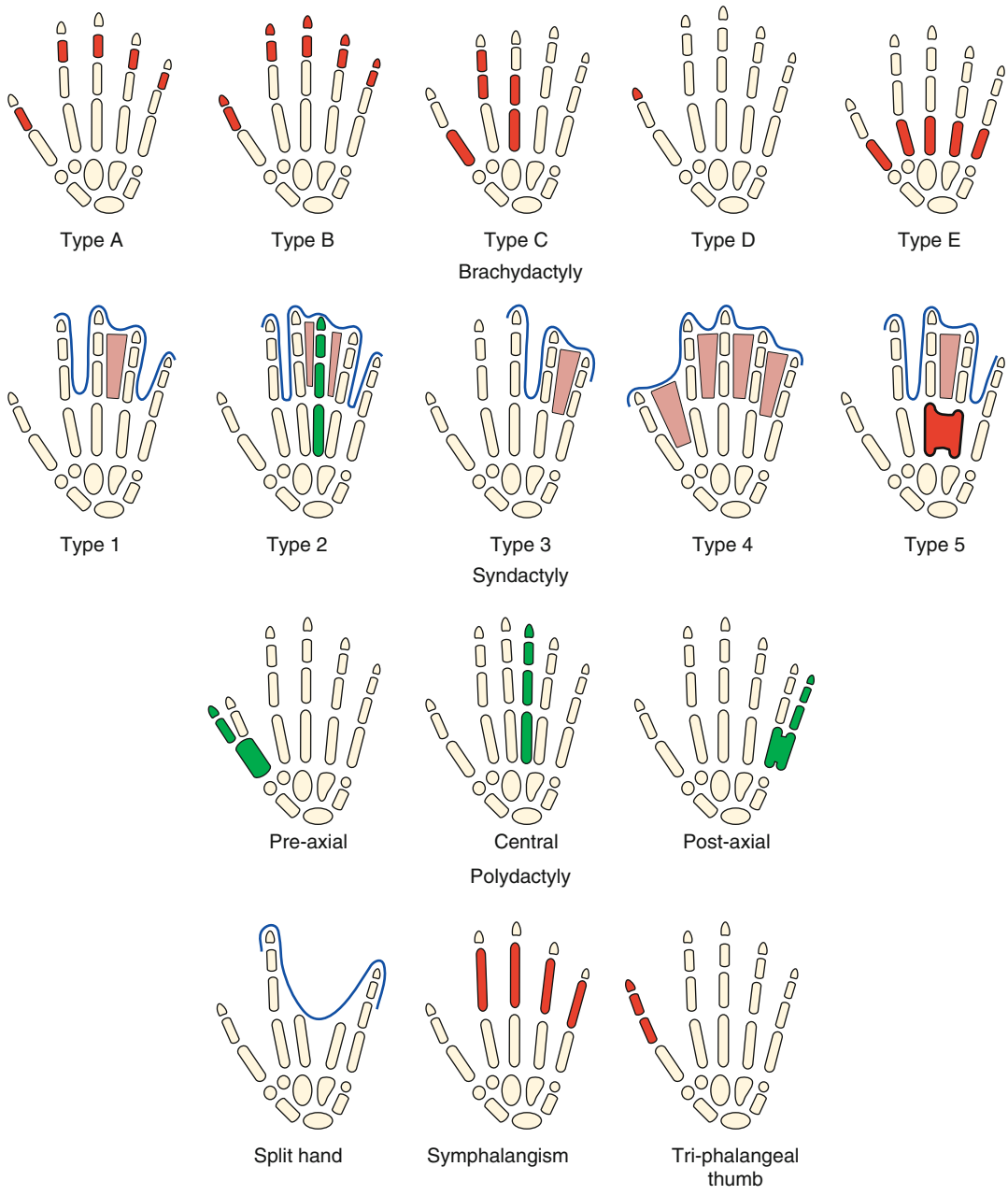


Fig. 14.2 Diagram showing malformations of the fingers (Modified from Schwabe and Mundlos 2004)

complete (full length of the digit) or incomplete (not right up to tip of the digit) and may involve only the soft tissues (simple syndactyly) or may include the adjacent bones (complex syndactyly). Rarer forms of syndactyly involving multiple soft tissue and bony abnormalities are referred to as complicated syndactyly; they are usually associated with syndromes. Isolated syndactyly has been classified according to the interdigital spaces affected (Table 14.1 and Fig. 14.3).

Complicated syndactyly is often associated with syndromes such as Apert syndrome (acro-

Table 14.1 Types of isolated syndactyly

Type	Characteristics
Type I	Most common type
	Involves syndactyly of 3rd and 4th digits of the hand and 2nd and 3rd toes
	Syndactyly may be partial or complete
	Autosomal dominant inheritance
Type II	Soft tissue syndactyly between 3rd and 4th fingers and 4th and 5th toes with supernumerary digit in the syndactylous web
	Brachydactyly, clinodactyly, or camptodactyly may also be present
	Autosomal dominant inheritance
Type III	Syndactyly between 4th and 5th fingers
	Middle phalanx of the 5th finger is rudimentary or absent
Type IV	Extremely rare
	Complete syndactyly of all fingers
	No bony fusion
Type V	Extremely rare
	Metacarpal fusion of 4th and 5th or 3rd and 4th digits

cephalosyndactyly), Poland syndrome, and amniotic constriction band syndrome.

Syndactyly in Apert Syndrome

Children with Apert syndrome or acrocephalosyndactyly have cranial, midfacial, and hand anomalies (Fig. 14.4). In addition, there may be airway obstruction. The severity and complexity of the hand deformity vary, and descriptive terms such as spade, spoon, or rosebud hands have been used to categorize them (Table 14.2). In the most severe form, all four fingers and the thumb are involved, and the nails are fused into a single broad nail (Journeau et al. 1999; Upton 1991).

Syndactyly in Amniotic Band Syndrome

Syndactyly in amniotic constriction band syndrome differs from other forms of syndactyly (Fig. 14.5). Some fingers may be missing on account of intrauterine amputation. Constriction bands will be present in the fingers, wrist, calf, or foot. The pattern of involvement is asymmetric, and small clefts may be present in the region of the original web spaces. Clubfoot may be an associated deformity. Prenatal diagnosis is possible, and fetoscopic release of the constriction band has been successful in some instances (Richter et al. 2012).

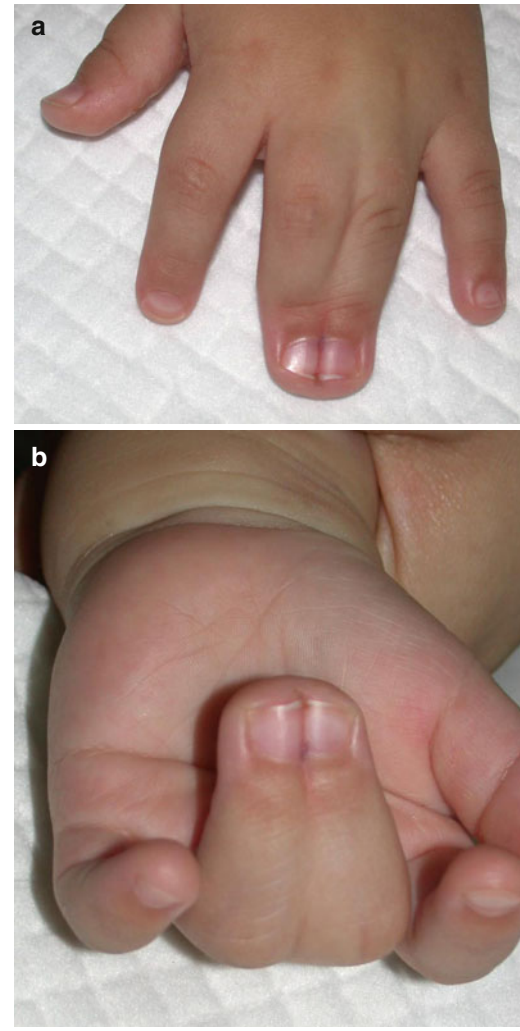


Fig. 14.3 (**a**, **b**) Isolated syndactyly

14.5.2 Brachydactyly

Brachydactyly or "short digits" may occur in isolation or as part of a generalized malformation syndrome (Nguyen and Jones 2009; Temtamy and Aglan 2008). Brachydactyly has been classified on the basis of the pattern of affected bones (Table 14.3).

Some examples of brachydactyly that is part of generalized malformation syndromes are shown in Table 14.4.

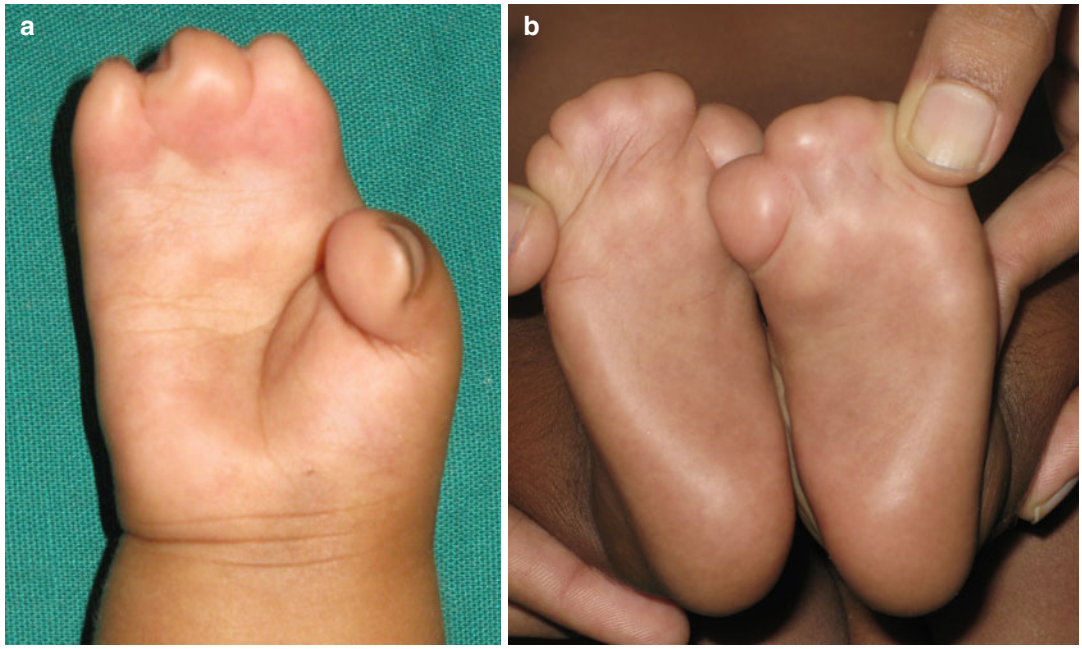


Fig. 14.4 Syndactyly in a child with Apert syndrome; the spade hand (**a**). Syndactyly of the feet is also present in this child (**b**)

Table 14.2 Patterns of syndactyly in Apert syndrome

Type	Characteristics
Type I: The spade hand	The most common variety
	The least severe variety
	The thumb is separated from the index finger by a narrow web space
	4th web syndactyly is simple
Type II: The spoon hand	The first web has a simple syndactyly
	Bony fusion of 2nd, 3rd, and 4th digits at terminal phalanges
	4th web syndactyly is simple
Type III: The rosebud hand	Most severe variety
	Least common variety
	Thumb, 2nd, 3rd, and 4th digits form complex syndactyly with a single nail
	4th web syndactyly complete and often complex with fusion of metacarpals

14.5.3 Symphalangism

Absence of interphalangeal joints or symphalangism most frequently involves the proximal interphalangeal joint (Ensink et al. 1999; Baek and Lee 2012). Less common is absence of the distal interphalangeal joint. The flexor finger creases will be absent, and the child cannot form a fist normally.

Symphalangism may be associated with carpal and tarsal coalition or be part of multiple synostosis syndromes such as the Nievergelt-Pearlman syndrome or the Fuhrmann type of multiple synostosis syndrome. Clubfoot that may occur in these syndromes will require surgery on account of the multiple or massive tarsal coalitions.

14.5.4 Deformities

Clinodactyly

Clinodactyly is a finger deformity in the coronal plane most commonly involving the middle phalanx of the little finger (Fig. 14.6). Occasionally, clinodactyly may involve other digits. Clinodactyly is very common in Down syndrome, Klinefelter syndrome, and Turner syndrome; a frequency as high as 79 % has been reported in Down syndrome (Sureshbabu et al.

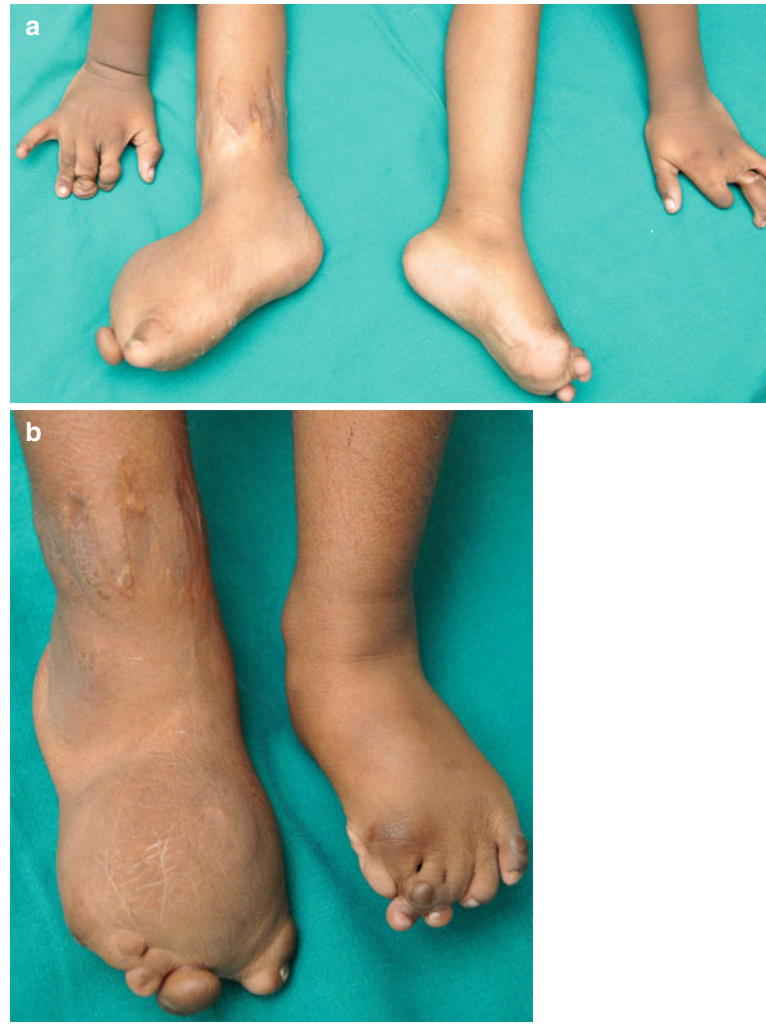


Fig. 14.5 The features of syndactyly in amniotic band syndrome are seen in the fingers and toes of this child (**a**); small clefts in the region of the original web spaces are visible (**b**)

Table 14.3 Classification of brachydactyly

Type	Characteristics
Type A	Hypoplasia or aplasia of middle phalanges
Type B	Hypoplasia of distal phalanges, nail dysplasia
	Hypoplasia of middle phalanges
Type C	Hypoplasia of middle phalanges of index and middle finger
	Short first metacarpal
	Fourth finger is the longest digit
Type D	Distal phalanx of the thumb is short
Type E	Hypoplasia of the metacarpals

2011). Familial clinodactyly is an isolated abnormality without any other skeletal or systemic abnormality.

Thumb clinodactyly is characteristically seen in Apert syndrome, Rubinstein-Taybi syndrome (Fig. 14.7), and diastrophic dwarfism. Thumb clinodactyly also occurs in triphalangeal thumbs.

Camptodactyly

Camptodactyly is a flexion deformity of the proximal interphalangeal joint; it typically involves the little finger though less frequently, other fingers may also be affected (Engber and Flatt 1977; Poznanski et al. 1969). The deformity which may be bilateral tends to progress. Three types have been described: type I involves the little finger only and becomes apparent in infancy (Fig. 14.8); type II manifests in preadolescence; and type III

Table 14.4 Brachydactyly that is part of generalized syndromes

Pattern of brachydactyly	Syndrome associated with brachydactyly
Generalized brachydactyly	Skeletal dysplasias (achondroplasia, diastrophic dysplasia, multiple epiphyseal dysplasia, spondyloepiphyseal dysplasia)
Hypoplasia of distal phalanges	Pyknodysostosis
	Cleidocranial dysostosis
	Fanconi anemia
	Progeria
Hypoplasia of distal phalanx of thumb (stub thumb)	Rubinstein-Taybi syndrome
	Robinow syndrome
Hypoplasia of first metacarpal	Holt-Oram syndrome
	Fanconi anemia
	Fibrodysplasia ossificans progressiva
Hypoplasia of middle phalanx of little finger	Down syndrome
	Russell-Silver syndrome
Brachydactyly with polydactyly	Ellis-van Creveld syndrome
	Orofaciodigital syndrome
Brachydactyly with syndactyly	Apert syndrome
	Cornelia de Lange syndrome

involves multiple digits and is typically seen in arthrogryposis and other syndromes.

The Clasped Thumb

The adducted clasped thumb (Fig. 14.9) must be differentiated from a trigger thumb and thumb-in-palm deformity of cerebral palsy. The deformity in trigger thumb is flexion at the interphalangeal joint, while in the clasped thumb, there is a flexion deformity of the metacarpophalangeal joint (Mih 1998; Tsuyuguchi et al. 1985). The primary deformity in cerebral palsy is adduction at the carpometacarpal joint. Congenital clasped thumb may be associated with arthrogryposis, Freeman-Sheldon syndrome, and other rare syndromes. Often there is muscle imbalance with weakness of the thumb extensors.

The Windblown Hand

A combination of an adducted and flexed thumb, flexion deformity of the metacarpophalangeal joints of the fingers, and ulnar deviation of the fingers at the metacarpophalangeal joints are the characteristic deformities in the windblown hand (Fig. 14.10). The deformity is seen in Freeman-

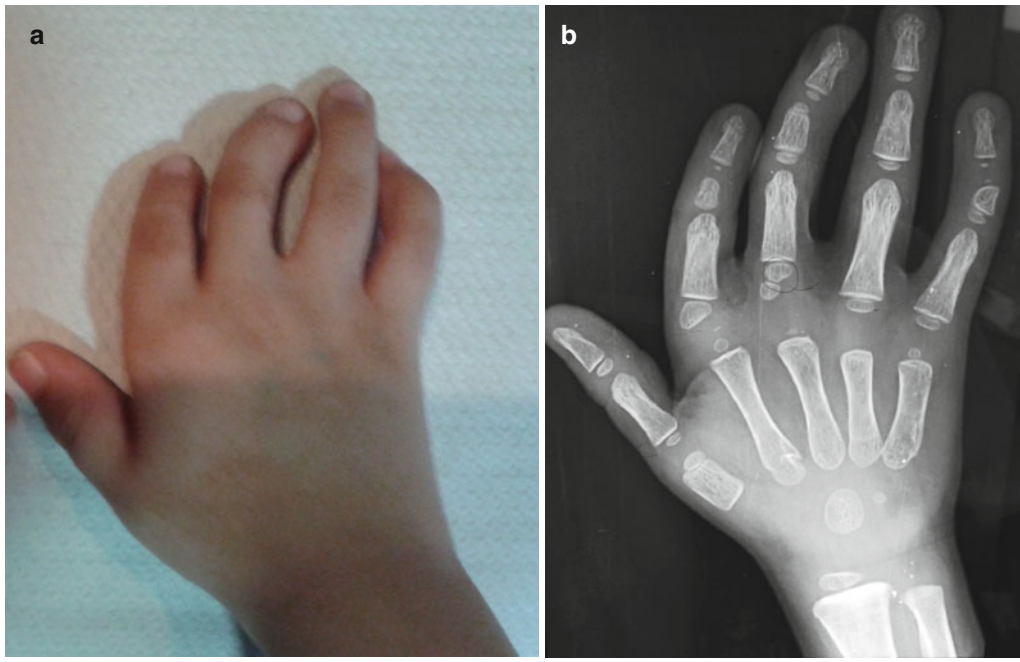


Fig. 14.6 (**a**, **b**) Clinodactyly

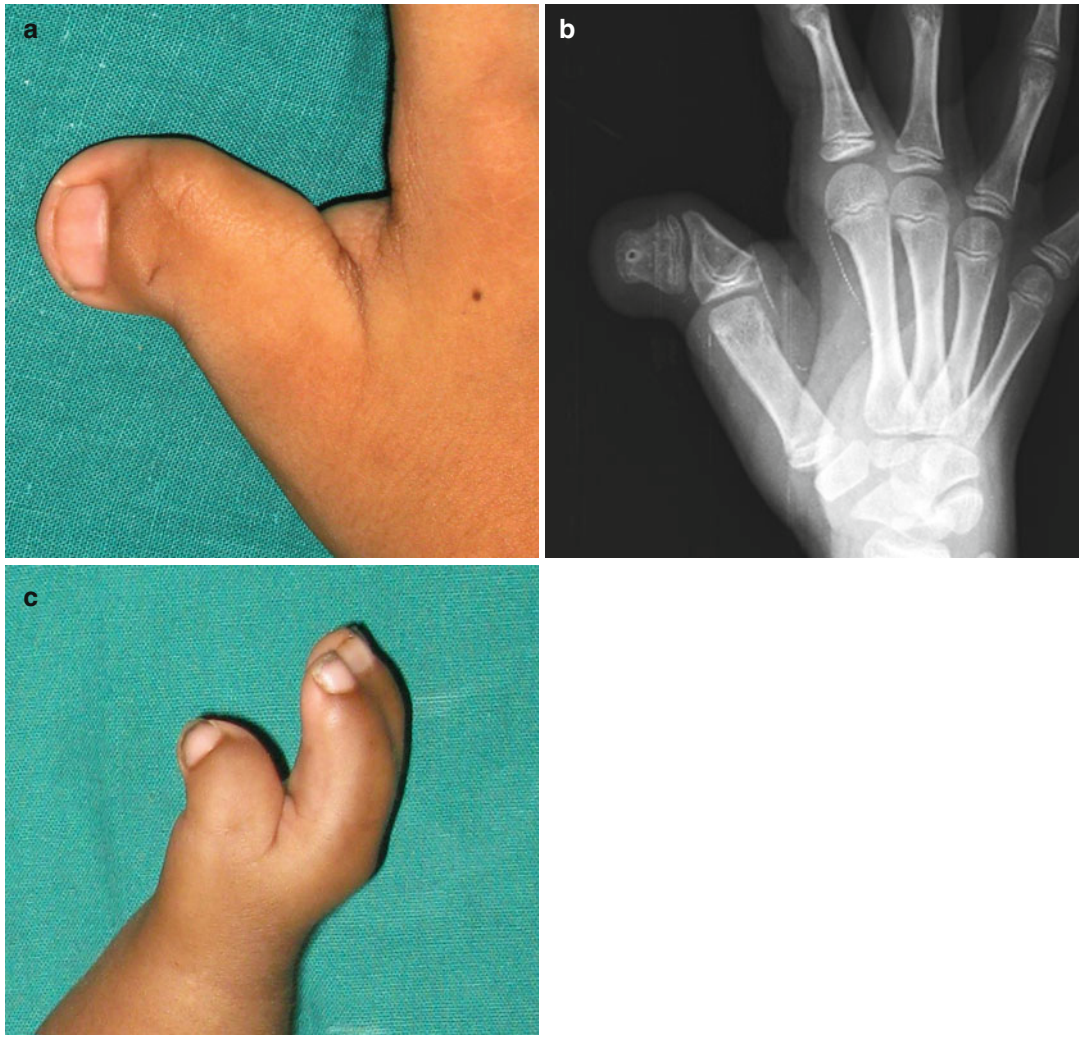


Fig. 14.7 Short, stubby thumb with clinodactyly in Rubenstein-Taybi syndrome (**a**); the proximal phalanx is a delta phalanx (**b**). Thumb clinodactyly in a child with Apert syndrome (**c**)

Sheldon syndrome and arthrogryposis (Wood 1994; Grunert et al. 2004). Clubfoot or congenital vertical talus deformities may be present in the feet in children with Freeman-Sheldon syndrome.

14.5.5 Arachnodactyly

Arachnodactyly or unusually long fingers may be seen in Marfan syndrome. When associated with flexion contractures of the fingers, elbows, and knees, the likely diagnosis is Beal syndrome (congenital contractural arachnodactyly).

14.6 The Toes

14.6.1 Brachydactyly

The Short Great Toe

The great toes are short and may have only a single phalanx in children with fibrodysplasia ossificans progressiva (FOP), a debilitating condition. Diagnosis of this condition can be made early in

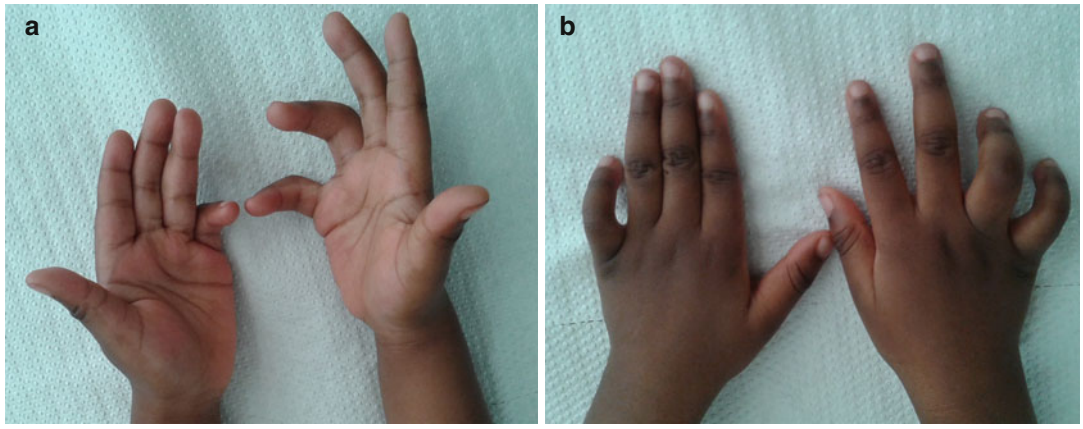


Fig. 14.8 (**a**, **b**) Camptodactyly of the little finger of the left hand and of the ring and little fingers on the right hand of a child

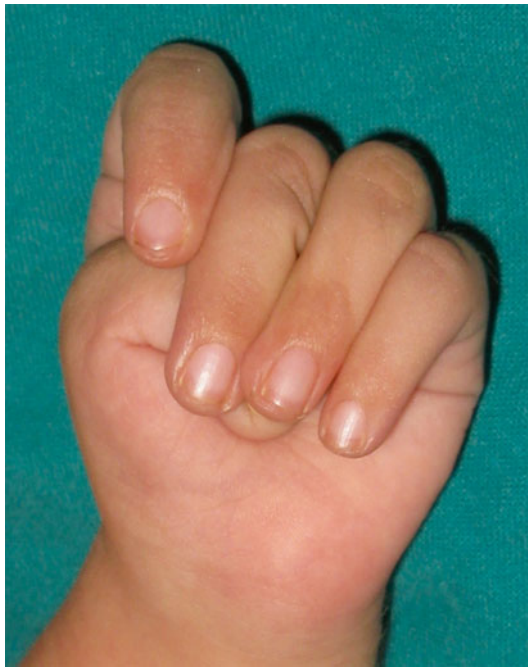


Fig. 14.9 The clasped thumb

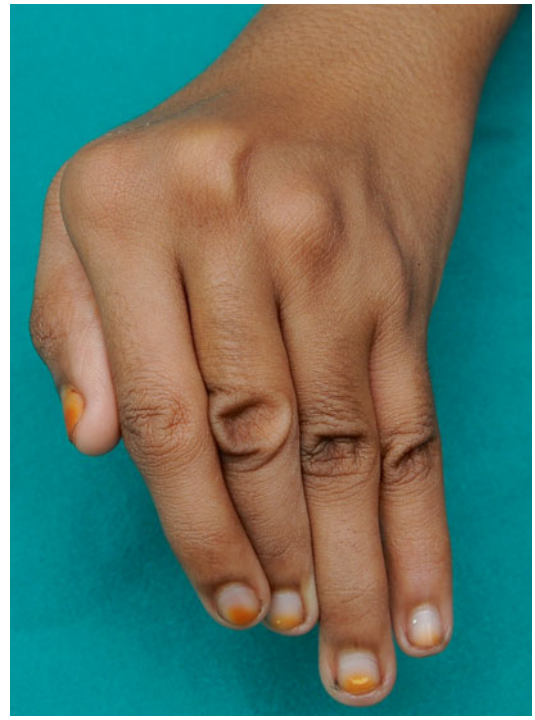


Fig. 14.10 Windblown hand

life by doing genetic analysis if cognizance is taken of a short great toe that is evident at birth (Kaplan et al. 2008). Unfortunately, the diagnosis is often delayed (Fig. 14.11). In the interim period, the child may be subjected to ill-advised surgery (biopsy or resection of soft tissue swelling that precedes heterotopic ossification) which hastens the progression of the disease. It is estimated that 90 % of children with FOP are misdiagnosed, and 67 % undergo dangerous and unnecessary diagnostic procedures that lead to permanent harm (Kitterman et al. 2005). Since it is now known that the condition is caused by a missense mutation of the gene encoding activin

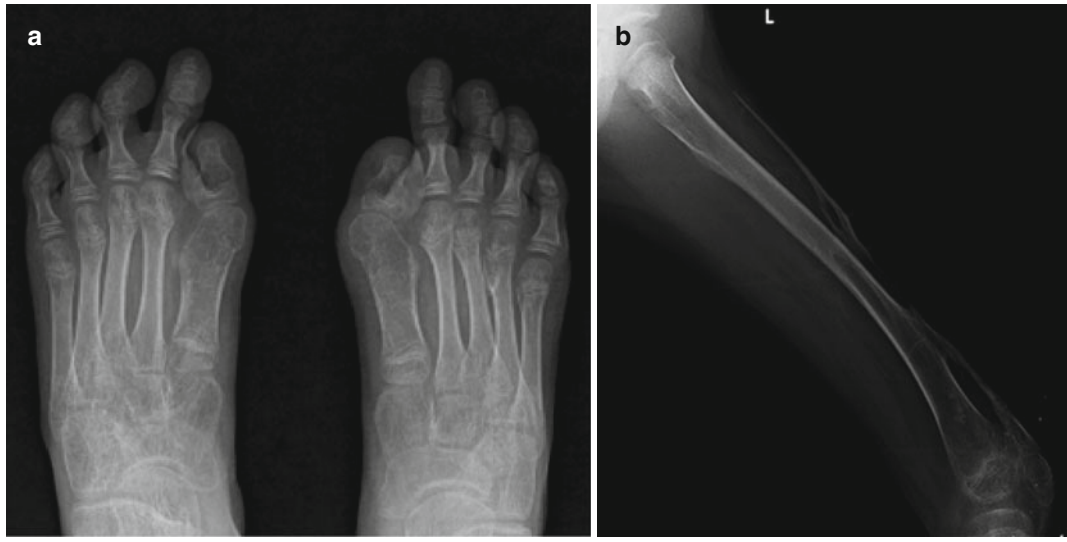


Fig. 14.11 Brachydactyly of the great toe in FOP is evident on the radiograph of the feet of a girl in whom the diagnosis was delayed and inappropriate surgery had been undertaken on the knee (**a**). The heterotopic ossification that developed following the surgery is seen in the thigh (**b**)

receptor IA (ACVR1), genetic testing can establish the diagnosis as soon as the condition is suspected on the basis of brachydactyly of the great toe (Haupt et al. 2014; Kaplan et al. 2010). In addition to the short great toe, children with FOP may have proximal tibial osteochondromas (~90 % of patients), fusion of the posterior elements of the cervical spine (~80 % of patients), and brachydactyly of the thumbs (~50 % of patients). However, the most evident feature is the short toe, and genetic testing on the basis of this alone is recommended (Kaplan et al. 2008).

Brachymetatarsia

Short metatarsals will give the impression that the corresponding toes are short (Fig. 14.12) (Baek and Chung 1998). The fourth metatarsal is most frequently affected. Brachymetatarsia is seen in Down syndrome and pseudohypoparathyroidism.

14.6.2 Deformities

Hallux Varus

Hallux varus is a medial deviation deformity at the metatarsophalangeal joint of the great toe; it must be distinguished from metatarsus primus varus and metatarsus adductus where the deformity is at the tarsometatarsal joint. Different forms of congenital hallux varus have been recognized (Joseph et al. 1984, 1987). The mildest form occurs in isolation and is easy to treat. Hallux varus associated with reduction or duplication anomalies of the foot is usually rigid and requires surgical correction (Fig. 14.13a–d).

Hallux Varus Interphalangeus

Here, the medial deviation is at the interphalangeal joint of the great toe (Fig. 14.14).

Lesser Toe Deformities

Curly toes which underlap or overlap the adjacent toe are fairly common deformities; the fifth toe is most frequently affected (Hamer et al. 1993; Smith et al. 2007). These deformities are often very responsive to nonoperative treatment (Smith et al. 2007) suggesting that there is no significant underlying structural anomaly.

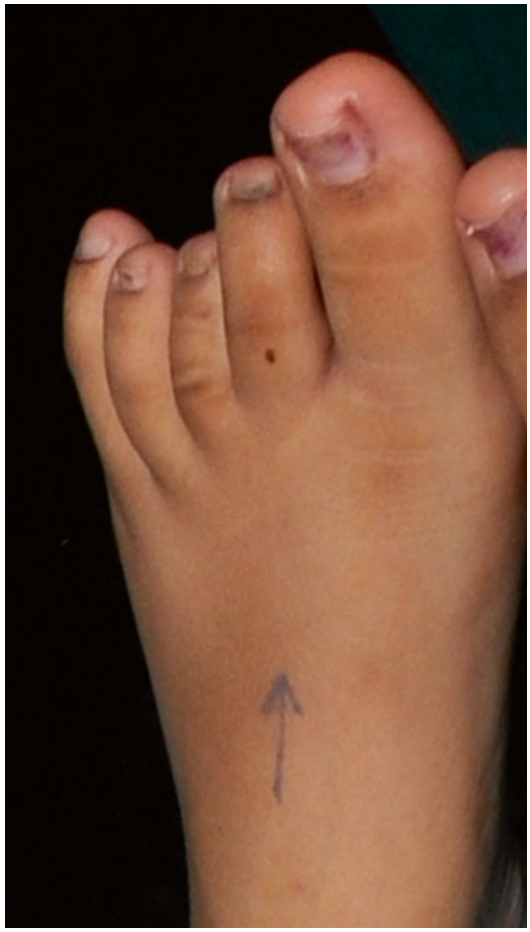


Fig. 14.12 Brachymetatarsia involving the third and fourth toes

14.7 Abnormalities of the Nails of Fingers and Toes

Nail changes in nail-patella syndrome may be varied; the nail may be hypoplastic, ridged, discolored, or pitted. Very similar changes may be seen in children with Ellis-van Creveld syndrome (Fig. 14.15).

14.8 Disorganized and Nonfunctional Digits

Grebe Chondrodysplasia

Bulbous digits that are nonfunctional are a feature of the homozygous form of this rare acromesomelic dwarfism. Agenesis or severe hypoplasia of several phalanges, metacarpals, and metatarsals results in this degree of severe malformation of the fingers and toes (Rao and Joseph 2002).

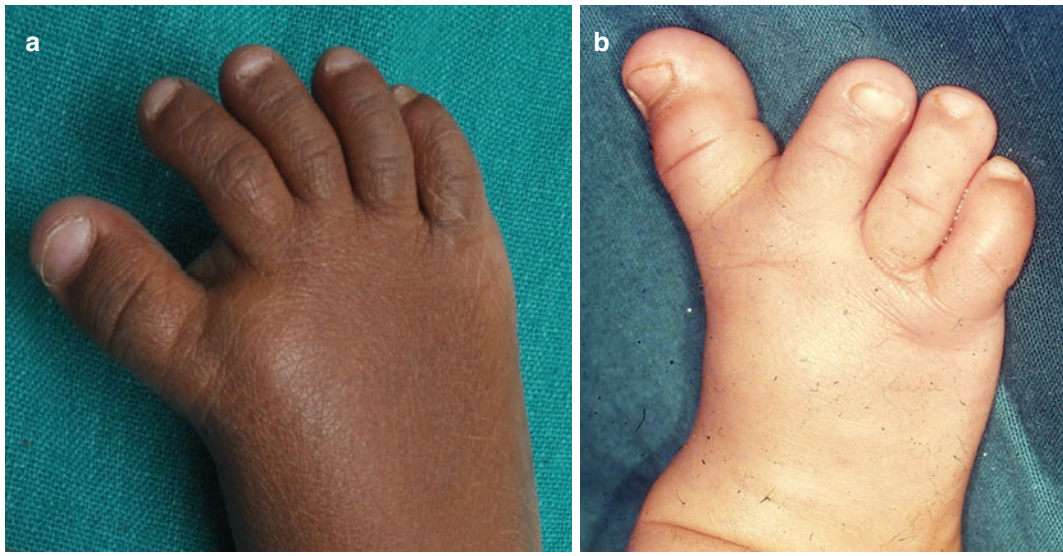
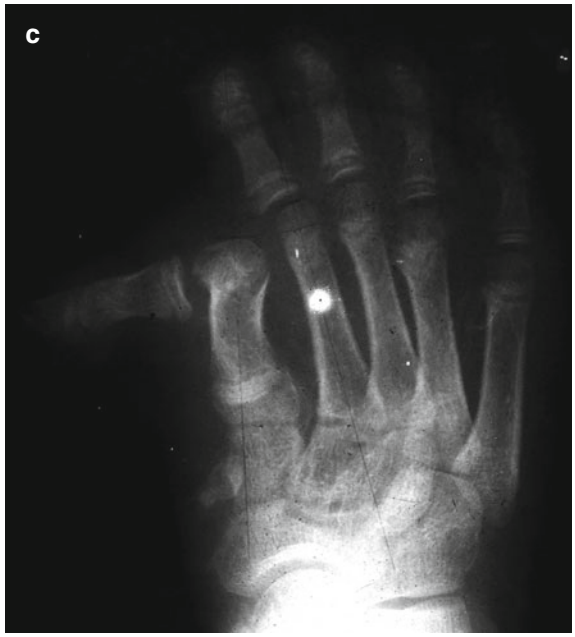


Fig. 14.13 Isolated hallux varus (**a**); hallux varus in a child with fibular hemimelia with aplasia of the lateral rays of the foot (**b**); radiograph of a child with hallux varus (**c**); hallux varus associated with preaxial polydactyly (**d**)



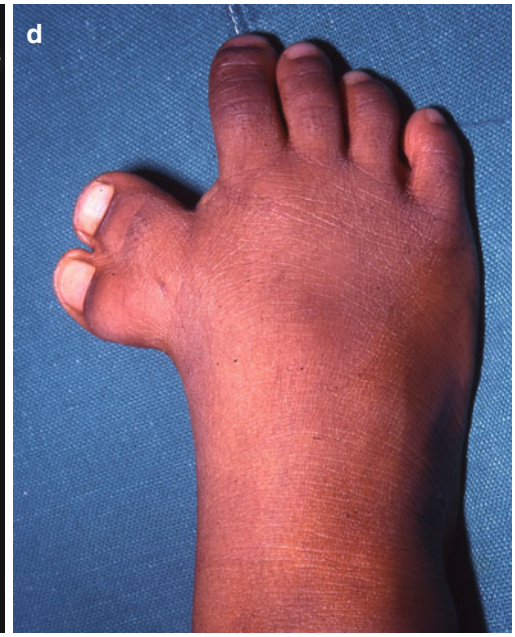


Fig. 14.13 (continued)

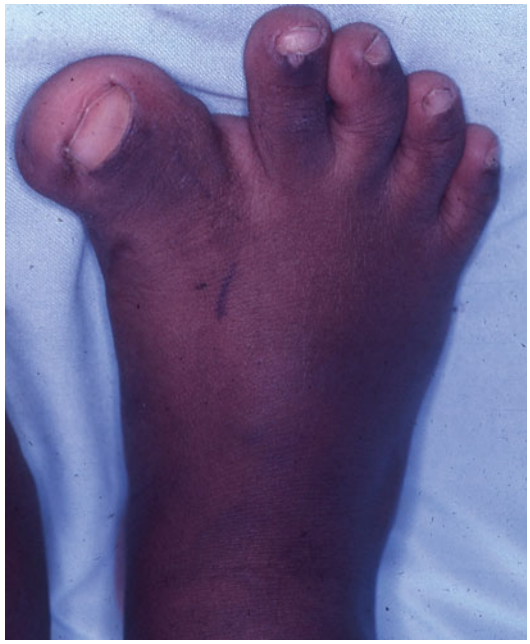


Fig. 14.14 Hallux varus interphalangeus

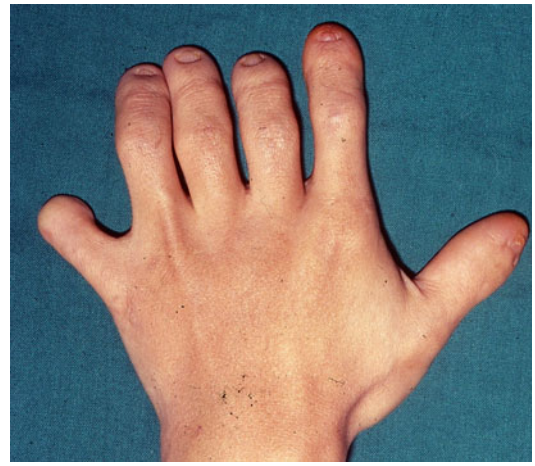


Fig. 14.15 Dystrophic nails in Ellis-van Creveld syndrome; postaxial polydactyly and brachydactyly are also present

14.9 Finger and Toe Abnormalities that may Facilitate Diagnosis of a Generalized Disorder

Some of the more important examples of finger or toe abnormalities that can facilitate prompt diagnosis of a generalized condition are shown in Table 14.5.

Table 14.5 Characteristic deformities of the fingers or toes that facilitate a diagnosis of syndromes

Finger or toe anomaly	Possible underlying syndrome	Comment
Clinodactyly of little finger	Chromosomal abnormality like Down syndrome	Very frequently seen in Down syndrome
Thumb clinodactyly	Rubinstein-Taybi	In addition to the angular deformity, the thumb is broad and stubby
Thumb clinodactyly	Diastrophic dysplasia	Thumb is also abducted (the hitch-hiker thumb). Dwarfism and deformities of the feet are also present. The external ear may be deformed
Great toe brachydactyly	Fibrodysplasia ossificans progressiva (FOP)	A child with a short great toe must be assumed to have FOP unless proved otherwise Surgical procedures must be avoided unless essential
Symphalangism	Carpal-tarsal coalition or multiple synostosis syndrome	If clubfoot is present, exclude tarsal synostosis before embarking on treatment
Arachnodactyly	Marfan syndrome or congenital contractural arachnodactyly (Beal syndrome)	Additional features of a crumpled pinna, flexion contractures of the finger, elbows, and knees will help to distinguish Beal syndrome from Marfan syndrome
Dysplastic nails	Nail-patella syndrome Ellis-van Creveld syndrome	Nail-patella syndrome will have associated abnormalities of the patella Postaxial polydactyly is an associated feature of Ellis-van Creveld syndrome

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Limb Length Inequality at Birth

15

Randall T. Loder and Benjamin Joseph

15.1 Introduction

Limb length inequality that is evident at birth is most frequently seen in children with congenital malformation of one limb (Fig. 15.1) or in children with bilateral asymmetric malformations (Fig. 15.2).

15.2 Questions to Establish a Diagnosis

- **What is the severity of limb length inequality?**
- **Is the limb well formed and are the thigh and leg segments of normal relative proportions?**
- **If the limb is not well formed, is there shortening in the thigh segment and the leg segment, or are both segments short?**
- **Is there a visible angular or bowing deformity of the short segment?**

What is the severity of limb length inequality?

A severe degree of limb length inequality to the extent that the foot of the short limb is at or above the level of the normal knee almost invariably indicates that there is a major deficiency of the femur.

Is the limb well formed and are the thigh and leg segments of normal relative proportions?

If the limb is well formed with the segments of near-normal proportions, conditions like unilateral teratological congenital hip dislocation, unilateral congenital coxa vara, or minor form of congenital short femur are possible likely diagnoses.

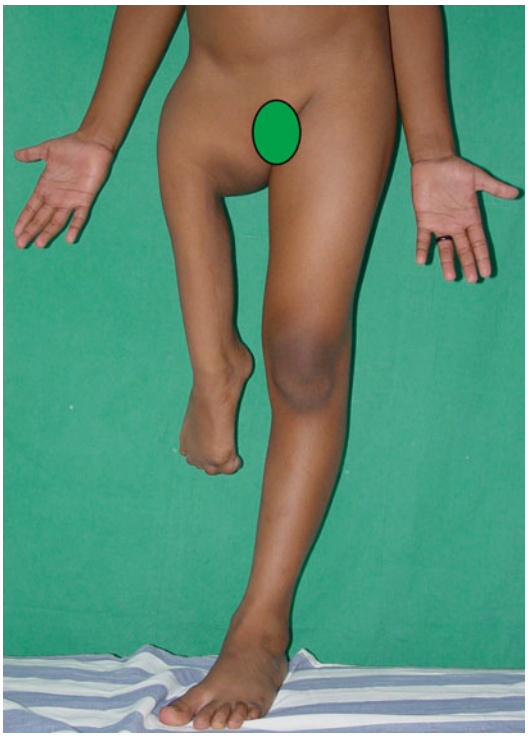

Fig. 15.1 Severe shortening of one lower limb in a boy with distal femoral deficiency

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_15

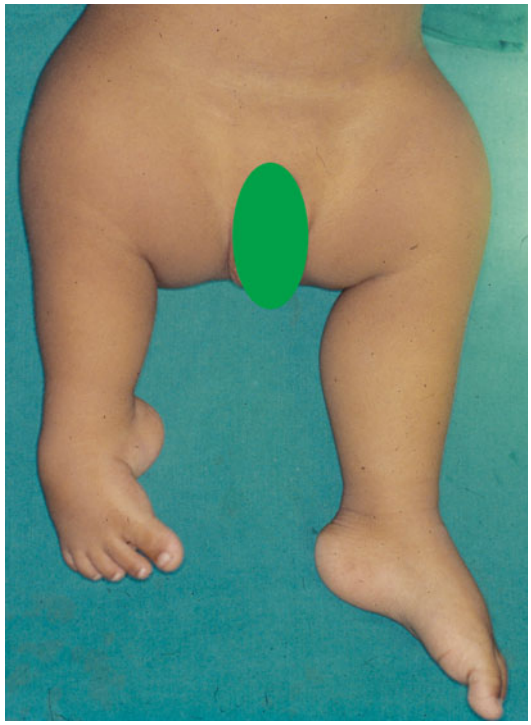

Fig. 15.2 Limb length inequality in a child with bilateral proximal focal femoral deficiency. The severity of the deficiency is asymmetric

If the limb is not well formed, is the shortening in the thigh segment, the leg segment, or are both segments short?

If the thigh segment is severely short, the conditions that need to be considered are congenital short femur, proximal or distal focal deficiency, and femoral agenesis. Modest shortening of the supratrochanteric part of the femur will be seen in children with coxa vara and hip dislocation. Shortening of the leg segment is seen in tibial and fibular hemimelia and congenital posteromedial bowing of the tibia.

Is there a visible angular or bowing deformity of the short segment?

Angular or bowing deformities of a short tibia are seen in association with fibular hemimelia and congenital posteromedial bowing of the tibia.

15.3 Physical Examination

15.3.1 Look

Note if the thigh and leg segments are well formed, hypoplastic, or deformed. Identify each segment of limb that is short.

15.3.2 Feel

If the leg is short, palpate the tibia and fibula to determine if they are formed.

15.4 Investigations to Confirm the Diagnosis

Radiography

A plain radiograph of both the lower limbs that include the hips and feet is often sufficient to identify the site and degree of shortening.

Ultrasound

Ultrasound of the hip is useful to delineate parts of bone that are yet to ossify (e.g., the proximal femur or tibia). Children with limb deficiency should have a cardiac and abdominal ultrasound to exclude visceral anomalies that may be associated with the limb anomalies.

15.5 Differential Diagnosis

15.5.1 Deficiencies

Major deficiencies that involve the femur include congenital short femur (Gillespie 1998; Gillespie and Torode 1983; Pappas 1983), proximal femoral focal deficiency (Gillespie 1998; Lange et al. 1978; Panting and Williams 1978; Pappas 1983), and distal femoral focal deficiency (Gilsanz 1983; Taylor et al. 2009) and total agenesis of the femur. The degree of shortening can be very severe in these conditions (see Chap. 1).

Deficiencies affecting the leg include tibial hemimelia (Jones et al. 1978) and fibular hemi-

melia (Rodriguez-Ramirez et al. 2010). The degree of shortening is not as severe as that seen in femoral deficiencies but is often quite significant. Associated deformities of the knee and foot are common, and the leg may be bowed.

15.5.2 Deformities

Unilateral congenital coxa vara will produce mild shortening of the limb (Beals 1998; Weinstein et al. 1984). Adduction of the hip is characteristically increased, and the range of abduction is proportionately reduced. Congenital bowing of the tibia (anterior, anterolateral, and posteromedial) may often be associated with moderate degrees of shortening (see Chap. 4).

15.5.3 Dislocation

Dislocation of the hip that is present at birth is usually a teratogenic dislocation and not the typical developmental dysplasia of the hip (DDH) as hip dislocation in DDH is not present at birth but occurs postnatally. Unilateral hip dislocation will result in shortening which is mild and usually associated with reduced passive hip abduction.

15.5.4 Hemihypertrophy

Hemihypertrophy may manifest at birth with limb length inequality. The girth of the limb and the size of the foot are also increased on the affected side (see Chap. 34).

15.5.5 Other Causes of Limb Length Inequality at Birth

Mild shortening may be seen in children with unilateral congenital idiopathic clubfoot (Little and Aiona 1995; Spiegel and Loder 2003; Shimode et al. 2005). Limb length inequality may be present in children with asymmetric paralysis of the lower limbs associated with spina bifida and sacral agenesis.

15.6 Establishing the Diagnosis

An outline of establishing the diagnosis of the cause of limb length inequality in the newborn is shown in Table 15.1.

Table 15.1 Establishing the diagnosis of the cause of limb length inequality at birth

Physical examination				
Short limb not well formed ± deformity			Both limbs well formed No externally visible deformity	
Severe shortening	Moderate shortening	Mild to moderate shortening	Mild shortening	Mild shortening
Thigh segment very short	Leg segment short	Leg segment short	Thigh and leg segment of near-normal proportions Supratrochanteric shortening	Thigh and leg segment of normal proportions
–	–	–	Thigh and calf girths on both limbs are equal	Thigh and calf girth on the longer limb is greater
Leg not bowed	Leg may be bowed	Leg bowed	Leg not bowed	Leg not bowed
Foot normal	Foot often deformed	Foot may be deformed	Foot normal	Foot larger in the longer side
Working diagnosis: Femoral deficiency	Working diagnosis: Tibial or fibular deficiency	Working diagnosis: Shortening associated with tibial bowing	Working diagnosis: Teratologic congenital hip dysplasia	Working diagnosis: Hemihypertrophy
Investigations				
Plain radiograph: Femoral deficiency and limb length inequality	Plain radiograph: Deficiency of the femur/tibia/fibula and limb length inequality	Plain radiograph: Deformity of the tibia and limb length inequality	Plain radiograph: Hip dislocation or coxa vara and limb length inequality	Plain radiograph: No deformity/dislocation/deficiency Only limb length inequality
Diagnosis				
Major femoral deficiency (PFFD/DFFD/agenesis of femur)	Major deficiency of tibia or fibula with or without minor femoral deficiency (Congenital short femur/tibial or fibular hemimelia)	Shortening associated with bowing of the tibia (Congenital posteromedial bowing/anterolateral bowing)	Teratologic hip dysplasia/coxa vara	Hemihypertrophy

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Abnormal Body Proportions and Dwarfism at Birth

16

Benjamin Joseph

16.1 Introduction

Dwarfism that is evident prenatally and in the newborn constitutes a small proportion of all the types of dwarfism that occur in children. Though the dwarfism per se is easily recognized on antenatal ultrasound scans, establishing a definitive diagnosis of the type of dwarfism is more difficult and needs the skills of an experienced radiologist. Despite this, the accuracy of prenatal diagnosis of dwarfism has improved in recent years; precise diagnoses have been reported with ultrasound (Wladimiroff et al. 1984; Trujillo-Tiebas et al. 2009; Jones et al. 1990) and CT scans (Miyazaki et al. 2012; Mace et al. 2013; Tamaru et al. 2009).

Dwarfism that is evident prenatally includes lethal and nonlethal conditions; only nonlethal forms of dwarfism are discussed in this chapter. It is useful to be aware of the more common nonlethal causes of dwarfism at birth as parents of these children are often distressed; they need reassurance and some information regarding the natural history of the condition.

16.2 Questions to Establish a Diagnosis

- **Is there a history of intrauterine growth retardation?**
- **Is the dwarfism proportionate or disproportionate?**

Is there a history of intrauterine growth retardation?

Intrauterine growth retardation (IUGR) due to placental insufficiency will result in stunted growth of the fetus, and the baby may then have short stature and low birth weight. The baby, however, will have relatively normal body proportions. IUGR may be of two types – asymmetrical and symmetrical. Asymmetrical IUGR is more common; the development during the first two trimesters of pregnancy is adequate, but there is retardation of growth during the last trimester. In this form of IUGR, neurological sequelae are less frequent as the brain development in the early part of gestation has progressed normally. In symmetric dwarfism, early brain development is also affected, and consequently, permanent neurological sequelae are often present. Maternal factors that predispose to IUGR include hypertension, diabetes, malnutrition, anemia, viral infections, and substance abuse. Chromosomal abnormalities and genetic mutations may also lead to IUGR.

A baby with short stature due to IUGR due to placental insufficiency must be differentiated from dwarfism of other causes.

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Is the dwarfism proportionate or disproportionate?

Distinguishing between proportionate dwarfism and disproportionate dwarfism is useful as it may help in reaching a diagnosis. Proportionate dwarfism is seen in primordial dwarfs, spondyloepiphyseal dysplasia, and spondyloepimetaphyseal dysplasia. Disproportionate dwarfism with short limbs occurs in achondroplasia and diastrophic dysplasia, while disproportionate dwarfism with a short trunk is seen in brachyolmia. Dwarfism in brachyolmia may not be evident at birth but usually manifests later in childhood.

16.3 Physical Examination

The body proportions must be assessed; a visual impression is often sufficient when disproportion is marked. If disproportion is doubtful, the upper and lower segments of the body should be measured (see Chap. 33). When disproportionate dwarfism is evident, note which segments of the limb are abnormally short. Note if the skull is large or if there is microcephaly. Observe if the bridge of the nose is depressed or if there is facial dysmorphism. Examine the mouth to exclude a cleft palate. Examine the spine to exclude spinal deformity.

16.4 Investigations to Confirm the Diagnosis

Radiography

Plain radiographs of the entire spine and limbs are essential when dwarfism is noted in the newborn baby (Fig. 16.1).

On the radiographs, note and document the features listed in Table 16.1.

Table 16.1 Features to note on the radiograph of the newborn infant

Region	Feature to look for	Abnormality
Spine (any region)	Height of the vertebrae	Platyspondyly
	Segmentation defect or failure of formation	Hemivertebrae or block vertebrae
Cervical spine	Craniovertebral anomaly	Basilar impression Odontoid hypoplasia Atlantoaxial dislocation
Thoracic spine	Deformity	Scoliosis
Thoracolumbar spine	Deformity	Kyphosis
Lumbar spine	Dimensions of the spinal canal	Progressive reduction of interpedicular distances from L1 to L5
Pelvis	Inclination of the acetabular roof	Horizontal acetabular roof
Long bones	Ossification of the epiphyses	Delayed appearance of epiphyses
	Epiphyseal abnormality	Punctate calcification in epiphyses
		Epiphyseal irregularity
	Metaphyseal abnormality	Metaphyseal irregularity
Metacarpals and phalanges	Length Metaphyseal abnormality	Abnormally short Metaphyseal irregularity

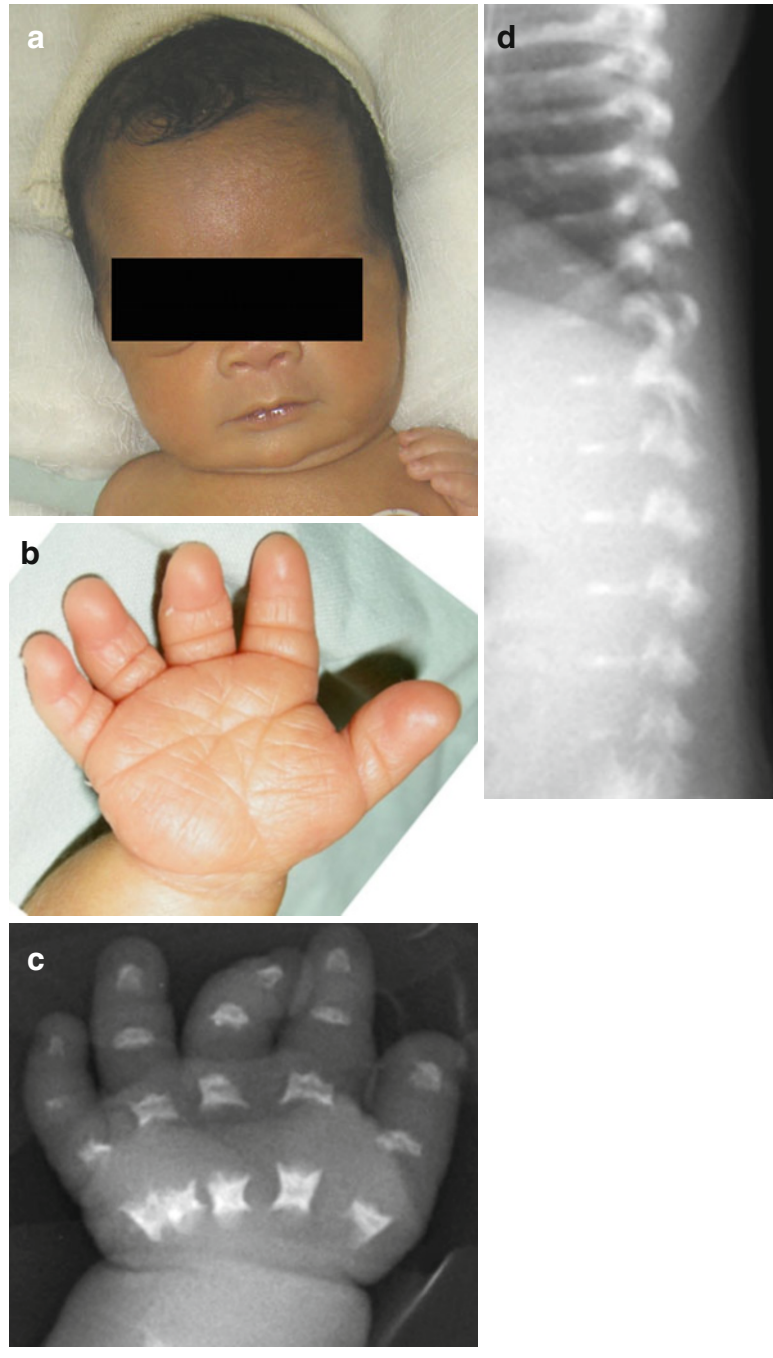

Fig. 16.1 Facial dysmorphism (**a**), short fingers with short phalanges and metacarpals (**b**, **c**), and marked platyspondyly (**d**) in a newborn with opsismodysplasia, a very rare form of dwarfism that was evident at birth (Courtesy Dr. Girish K, Manipal, India)

Table 16.2 Genetic abnormalities associated with some skeletal dysplasias with dwarfism that can be recognized at birth or prenatally

Genetic abnormality	Nonlethal forms of dwarfism	Lethal forms of dwarfism
Fibroblast growth factor receptor 3 (FGFR 3) mutations	Achondroplasia	Thanatophoric dwarfism
Sulfate transportation gene mutations	Diastrophic dysplasia	Achondrogenesis type IB Atelosteogenesis type II
Transient receptor potential cation channel, subfamily V, member 4 gene (TRPV 4) mutations	Brachyolmia Spondylometaphyseal dysplasia – Kozlowski type (SMDK) Metatropic dysplasia	Lethal metatropic dysplasia
Type II collagen gene mutations	Spondyloepiphyseal dysplasia (SED) congenita Spondyloepimetaphyseal dysplasia (SEMD) – Strudwick type	Achondrogenesis Hypochondrogenesis

16.5 Differential Diagnosis

16.5.1 Skeletal Dysplasia

The underlying genetic abnormality in several skeletal dysplasias is now known (Rossi and Superti-Furga 2001; Sulko et al. 2005; James et al. 2003; Cui et al. 2008), and this helps in classifying dwarfism recognizable at birth and prenatally (Table 16.2).

Disproportionate Dwarfism

Achondroplasia

Achondroplasia is the commonest and most well-known form of disproportionate dwarfism recognizable at birth.

The clinical features include short stature with rhizomelic shortening of the arms and legs, mild flexion deformity of the elbows, genu varum, and a trident hand. The head is large with frontal bossing, and the bridge of the nose is depressed. A gibbus may be present in the thoracolumbar region. Stenosis of the foramen magnum is present in some children with achondroplasia which may require surgical decompression to avert serious neurological compromise (Rimoin 1995; Bagley et al. 2006).

The typical clinical and radiographic features are shown in Fig. 16.2 and Table 16.3 respectively.

Fig. 16.2 Appearance of a newborn with achondroplasia. The rhizomelic shortening is evident (Courtesy Professor William MacKenzie, Wilmington, Delaware USA)

Diastrophic Dysplasia

Newborn infants with this form of dysplasia are short and have disproportionately short limbs. The thumbs are widely abducted (hitchhiker thumbs), and rigid foot deformities are characteristic features. Contractures of large joints may also be present. Scoliosis is common, and the deformity may become very severe. The skull is of normal size. Cleft palate is present in nearly half these children. Swelling of the pinna

Table 16.3 Radiographic features of achondroplasia

Region	Feature to be noted	Present/absent
Spine (any region)	Platyspondyly	Absent
	Segmentation defect or failure of formation	Absent
Cervical spine	Craniovertebral junction abnormality: foramen magnum stenosis	Often present
Thoracic spine	Scoliosis	Absent
Thoracolumbar spine	Kyphosis	Present
	Progressive reduction of interpedicular distances from L1 to L5	Present
Pelvis	Horizontal acetabular roof	Present
Long bones	Delayed appearance of epiphyses	Absent
	Punctate calcification in epiphyses	Absent
	Epiphyseal irregularity	Absent
	Metaphyseal irregularity	Absent
	Fibula longer than tibia	Present

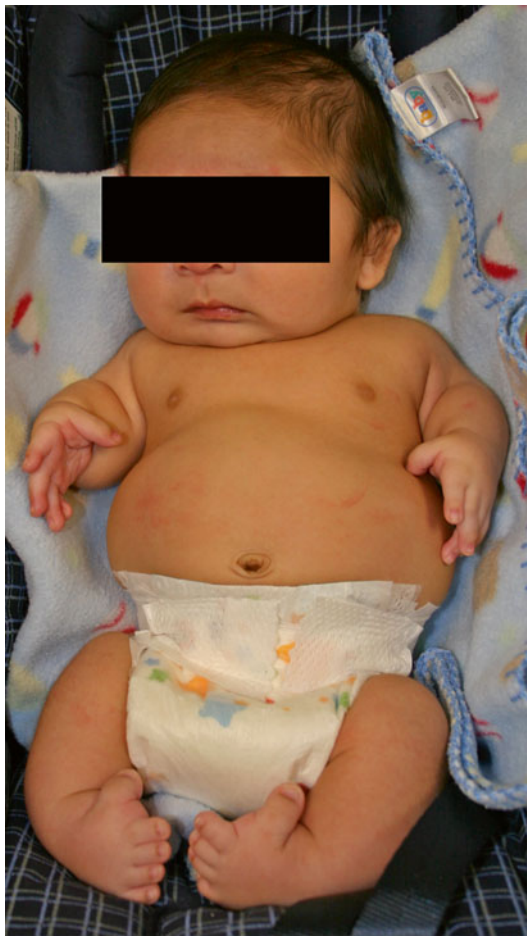

Fig. 16.3 Appearance of a newborn with diastrophic dysplasia. The hitchhiker thumbs and the deformed feet are seen along with the rhizomelic shortening (Courtesy Professor William MacKenzie, Wilmington, Delaware USA)

Table 16.4 Important radiographic features of diastrophic dysplasia

Region	Feature to be noted	Present/absent
Spine (any region)	Platyspondyly	Absent
	Incomplete ossification of upper thoracic vertebrae	Present
Cervical spine	Kyphosis Widening of cervical spinal canal ("cobra-like" appearance)	Present
Thoracic spine	Scoliosis	Present
Thoracolumbar spine	Coronal clefts in lower thoracic and lumbar vertebrae	Present
	Progressive reduction of interpedicular distances from L1 to L5	Present
Pelvis	Horizontal acetabular roof	Present
Long bones	Delayed appearance of epiphyses	Absent
	Punctate calcification in epiphyses	Absent
	Epiphyseal irregularity	Absent
	Metaphyseal flaring	Present
Patella	Fragmented or multilayered	Present
First metacarpal	Short	Present

is often seen at birth; later, the pinna gets crumpled and deformed (cauliflower ears).

The clinical and radiographic features of diastrophic dysplasia are shown in Fig 16.3 and Table 16.4 respectively.

Metatropic Dysplasia

The term metatropic dysplasia (metatropos – changing pattern) reflects the changing clinical course of the condition. The disorder appears relatively benign at birth but has a serious progressive course (Kozlowski et al. 1988). At birth, the features are of a short-limbed dwarfism. Characteristically, there is no facial dysmorphism. A taillike appendage is often present in the sacral region. In the first year of life, delayed motor development, contractures of joints, and severe and rapidly progressive kyphoscoliosis become evident. The progressive spinal deformity renders the trunk short relative to the limbs.

There is increasing support to the view that there is a spectrum of disease entities caused by TRPV4 mutations with lethal metatropic dysplasia at one end and autosomal dominant brachyolmia at the other and other forms of metatropic dwarfism and spondylometaphyseal dysplasia (Kozlowski type) in between (Kannu et al. 2007).

Proportionate Dwarfism

Spondyloepiphyseal Dysplasia Congenita (SED)

These children have very short trunks and limbs from birth though the hands and feet may be of average length (Spranger et al. 1994).

Spondyloepimetaphyseal Dysplasia (SEMD): Strudwick Type

At birth, these children have short limbs and a short trunk and a small chest. Cleft palate is a frequent occurrence. Facial dysmorphism is mild. High myopia that can lead on to retinal detachment is a feature of the condition. The hands and feet are relatively normal. Lung function may be impaired.

The radiographic features of SED and SEMD Strudwick are shown in Table 16.5.

Table 16.5 Radiographic features of SED and SEMD Strudwick type at birth

Region	Feature to be noted	Present/absent
Spine	Generalized platyspondyly Oval vertebral bodies (become pear shaped with posterior narrowing and rounded anterior borders in childhood)	Present
Cervical spine	Kyphosis Hypoplasia of the C3 or odontoid hypoplasia may be present	Absent at birth but may develop later
Thoracic spine	Scoliosis	Absent
Lumbar spine	L1 vertebra larger than L5 (anisospondyly)	Present
Pelvis	Horizontal acetabular roof	Present
Long bones	Markedly delayed appearance of epiphyses of the proximal and distal femur and pubic rami Very short femoral necks	Present
	Punctate calcification in epiphyses	Absent
	Epiphyseal irregularity	Absent
	Metaphyseal abnormality	Absent at birth[a]

[a]Metaphyseal changes appear later in childhood in SEMD Strudwick type in the form of flocculated fragmentation of the metaphysis with islands of sclerosis. These changes are more marked in the ulna and fibula than the radius and tibia

16.5.2 Primordial Dwarfism

Primordial dwarfism is an extremely rare group of disorders characterized by profound retardation of growth both in utero and in postnatal life. Different forms of primordial dwarfism have been described, and they include Seckel syndrome, osteodysplastic primordial dwarfism types I and II, Russell-Silver syndrome, and Meier-Gorlin syndrome. Microcephaly is a feature of the first three types. The dwarfism is proportionate, and the condition may be confused with other causes of intrauterine growth retardation (IUGR) as the prenatal ultrasound will demonstrate that the fetus is small for gestational age. Though the dwarfism is evident at birth, the diagnosis may not be made for a few years in some milder cases till it becomes clear that postnatal growth is also grossly retarded (Fig. 16.4).

16.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis of dwarfism at birth is shown in Table 16.6.

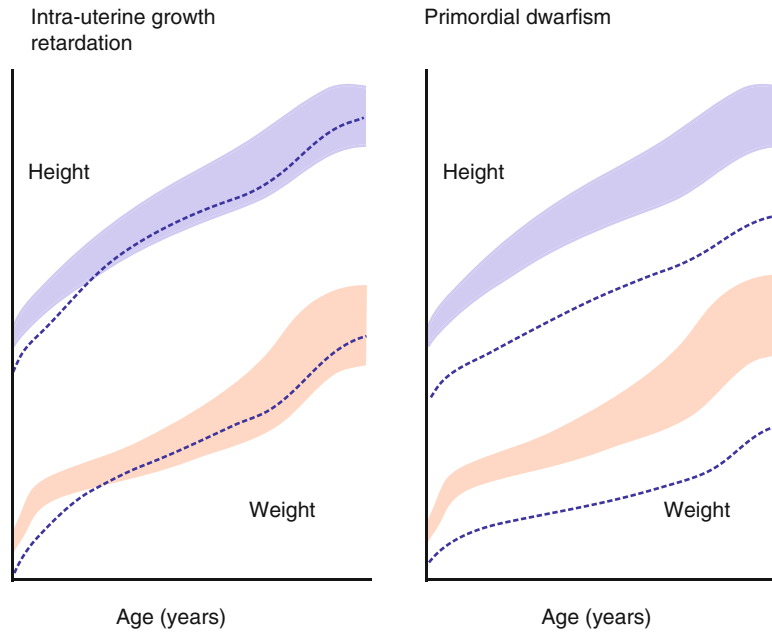

Fig. 16.4 Postnatal growth remains grossly retarded in primordial dwarfism unlike children with intrauterine growth retardation

Table 16.6 Establishing the diagnosis of dwarfism at birth

History			
Intrauterine growth retardation present	Intrauterine growth retardation present	Intrauterine growth retardation absent	Intrauterine growth retardation absent
Family history not present	Family history not present	Family history may or may not be present	Family history may or may not be present
No history of maternal illness during pregnancy	No history of maternal illness during pregnancy	No history of maternal illness during pregnancy	No history of maternal illness during pregnancy
Physical examination			
Proportionate dwarfism	Proportionate dwarfism often of a profound degree	Proportionate dwarfism	Disproportionate dwarfism
Head size normal	Microcephaly may be present	Head size normal	Head size may be normal or large
Spine normal proportion	Spine normal proportion Scoliosis may be present	Spine short Kyphosis may be present	Spine of normal proportion Thoracolumbar kyphosis or kyphoscoliosis may be present
Limbs normal	Limbs of normal proportions	Short limbs	Short limbs
Working diagnosis: Intrauterine growth retardation due to placental insufficiency	Working diagnosis: Primordial dwarfism	Working diagnosis: Skeletal dysplasia with proportionate dwarfism	Working diagnosis: Skeletal dysplasia with disproportionate dwarfism
Investigations			
Plain radiographs: Normal	Plain radiographs: No diagnostic features	Plain radiographs: Changes as noted in Table 16.5	Plain radiographs: Changes as noted in Tables 16.3 and 16.4
Diagnosis			
Short stature due to intrauterine growth retardation (placental insufficiency)	Primordial dwarfism	Spondyloepiphyseal dysplasia congenita or spondylometaphyseal dysplasia congenital (Strudwick type)	Achondroplasia or diastrophic dysplasia or metatropic dysplasia

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Part II

The Neonate and Infant

Decreased Movement of the Upper or Lower Limb in the Neonate

17

Benjamin Joseph

17.1 Introduction

Decreased spontaneous movement of a limb in a neonate is to be taken seriously as urgent intervention may be needed in some instances (Waseem et al. 2009). The two reasons for reduction in spontaneous movements are paralysis and pain, and it is the latter that should invoke a sense of urgency in the treating physician. Pain arising from the limb manifests as pseudoparalysis, and the two most common causes are birth trauma (Lam et al. 2002; Matsubara et al. 2008; Morris et al. 2002; Cebesoy et al. 2009; Nakazato et al. 2001; Jacobsen et al. 2009; Lindseth and Rosene 1971; Journeau et al. 2001) and osteoarticular infection (Bulbul et al. 2009; Dessi et al. 2008; Frederiksen et al. 1993). The commonest cause of paralysis in the newborn is obstetric brachial plexus palsy; rarer palsies have been reported in the literature (al-Qattan et al. 1996; Hayman et al. 1999; Goetz 2010). Occasionally, birth palsy may be associated with birth trauma (Journeau et al. 2001) and very rarely with osteoarticular infection (Estienne et al. 2005).

Electronic supplementary material The online version of this chapter (doi:10.1007/978-81-322-2392-4_17) contains supplementary material, which is available to authorized users.

17.2 Questions to Establish a Diagnosis

- **Was the delivery difficult?**
- **Was the child moving the limb after birth and then stopped moving it spontaneously?**
- **Was the reduction in movement preceded by fever?**
- **Does gentle passive movement of the limb cause the baby to cry?**

Was the delivery difficult?

It is important to elicit details of the delivery, including the birth weight of the baby. A history of difficult birth with shoulder dystocia may point to the likelihood of obstetric brachial plexus palsy or birth injury. Obstetric palsy is also more frequently encountered in large babies particularly of diabetic mothers.

Was the child moving the limb after birth and then stopped moving it spontaneously?

If the mother is certain that the baby was moving all four limbs to begin with and diminished movement of a limb was noted a day or two after birth, the diagnosis of a postnatal problem such as infection or trauma needs to be considered.

Was the reduction in movement preceded by fever?

The likelihood of bone or joint sepsis is high if the reduction of movement was preceded by fever or if the newborn is already receiving

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antibiotics in the neonatal intensive care unit for another focus of infection. However, in the neonate bone or joint infection may develop in the absence of fever.

Does gentle passive movement of the limb cause the baby to cry?

Pain on gentle passive movement of the affected limb suggests that there may be a birth injury or a focus of infection either in a joint or very close to the joint.

17.3 Physical Examination

Careful examination is most important for establishing the diagnosis.

17.3.1 Look

Look for fullness or redness over the clavicle, shoulder, and elbow if diminished movements of the upper limb have been noted. Similarly look for fullness around the knee and ankle if the lower limb is affected. It is important to remember that redness or swelling may not be seen often in the region of the hip as it is a deep-seated joint.

Watch the baby as it lies awake on its back and observe the spontaneous movement of the limbs. This requires some patience on the part of the physician as sometimes the baby may not move any limb for even up to a minute. Gently stroking the soles of the feet or the hands may be a stimulus for the baby to move the limbs. Diminished spontaneous movement of one limb can easily be appreciated when the baby moves the other limbs spontaneously (Fig. 17.1 and Video 17.1).

17.3.2 Feel

Begin by palpating areas that are not likely to be painful and then move to the area that is the probable site of pain. Systematically palpate gently each bone of the limb (clavicle, humerus, radius, and ulna in the upper limb and the femur, tibia, and fibula in the lower limb) and see if the baby cries when any particular point is palpated. Redo this examination to confirm the point of tenderness.

17.3.3 Move

Very gently move the joints of the affected limb. If the child does not cry when each joint is moved through a few degrees, gradually increase the arc of motion. If the joint is not inflamed and if the bones are intact, a full range of motion may be possible.

17.3.4 Special Tests

The Moro Reflex

If it is very clear that the baby does not have any pain on movement of the upper limb, elicit the Moro reflex.

The test: The head of the baby is gently lifted with enough support to just begin to raise the torso from the cot. The head is then released suddenly and allowed to fall for a very brief moment and quickly supported again before it comes in contact with the cot. The baby normally extends and abducts both shoulders and then adducts and flexes them.

Unilateral absence of the Moro reflex in the absence of local tenderness or pain on passive motion indicates that there is paralysis of the shoulder.

The Grasp Reflex

The test: A finger is placed in the palm of the baby; *the fingers of the baby grasp the examiner's finger.*

The reflex will not be present if the fingers are paralyzed.

The Plantar Grasp

The test: Stroking the sole causes the toes to flex or curl.

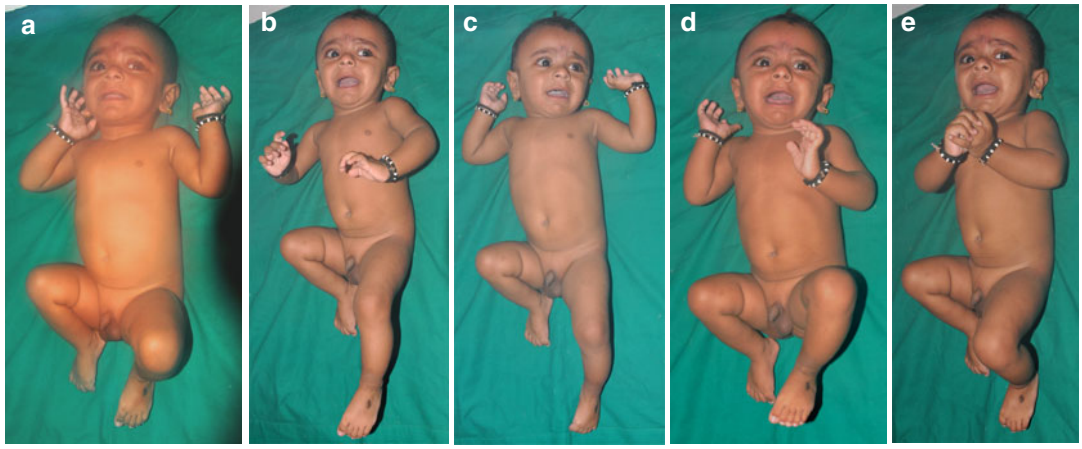

Fig. 17.1 (**a**–**e**) Photographs of an infant taken a few seconds apart (**a**–**e**) show spontaneous movements of the upper limbs and the left lower limb. The right lower limb has not been moved throughout this period. The infant had septic arthritis of the right hip

The reflex will be absent if the toes are paralyzed.

The Babinski Sign

The Babinski sign is usually positive in the normal newborn. Dorsiflexion of the great toe and fanning of the lesser toes will not be noted if the lower limb is paralyzed.

17.4 Investigations to Confirm the Diagnosis

17.4.1 Plain Radiographs

Plain radiographs of the affected limb are essential if birth trauma or infection is suspected. The radiograph should be centered on the region that is painful on movement or tender on palpation. Wherever possible, two orthogonal views of the region of interest should be obtained.

Radiographic features of birth trauma are usually quite evident with visible discontinuity of the bone with fractures of the diaphysis of long bones (Fig. 17.2a). However, epiphyseal separations may be more difficult to identify as the displaced epiphysis may not be ossified (Fig. 17.2b); the clinician must not be swayed by an unwary radiologist's report of a dislocation (epiphyseal separations may be demonstrated on the ultrasound scan).

The radiographic changes in osteomyelitis and septic arthritis will change and progress as the disease evolves (Offiah 2006; Jaramillo et al. 1995).

Sequential radiographic features of osteomyelitis include:

- Deep soft tissue swelling
- Osteoporosis
- Periosteal new bone formation (late feature)
- Bony destruction (late feature)

Sequential radiographic features of acute septic arthritis include:

- Widening of the joint space (Fig. 17.2c)
- Bulging of the soft tissue (e.g., distended capsular shadow)
- Subluxation
- Dislocation

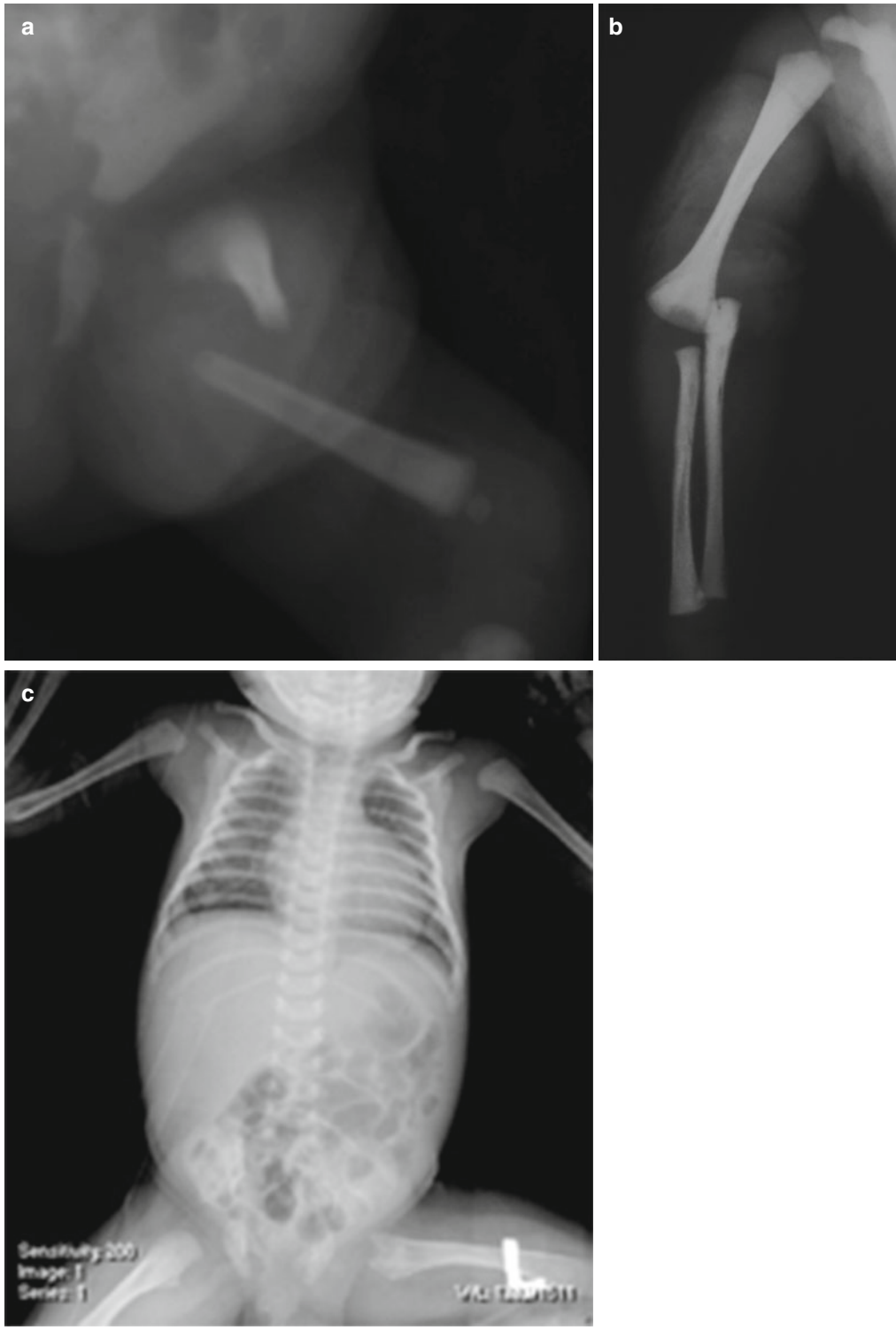

Fig. 17.2 (**a**) Fracture of the femoral shaft in a newborn following a breech delivery. (**b**) Traumatic separation of the distal humeral epiphysis noted soon after birth. The abnormal relationship between the humerus and the forearm bones suggests that the epiphysis is displaced. (**c**) Increased joint space and early bony destruction of the proximal femoral metaphysis in a neonate with osteomyelitis of the proximal femur with septic arthritis of the left hip. All three neonates had decreased spontaneous movements of the affected limb

17.4.2 Ultrasound

Ultrasound examination is operator dependent and this must be borne in mind. In experienced hands it can be very useful in the diagnosis of osteoarticular infection in the neonate (Mah et al. 1994).

Sequential ultrasonographic features of acute osteomyelitis include (Mah et al. 1994):

- Deep soft tissue swelling
- Elevation of periosteum by a thin layer of fluid
- Definite subperiosteal abscess
- Cortical erosion (late feature)

A normal ultrasound does not exclude osteomyelitis.

- Joint effusion (the nature of the effusion [purulent or otherwise] cannot be confirmed on the basis of the size of the effusion or the echogenicity)
- Subluxation
- Dislocation

17.4.3 Magnetic Resonance Imaging

Though MRI provides excellent soft tissue and bone contrast, areas surrounding the actual site of infection will also show signal changes if reactive or sympathetic inflammation is present (Offiah 2006).

The characteristic features of infection are:

- A low signal on T1-weighted images
- A high signal on T2-weighted and STIR sequences

Gadolinium enhancement gives better visualization of pus collection in the soft tissues and bone.

17.4.4 Isotope Scan

Isotope bone scan is more sensitive than radiography in the early stages of osteomyelitis, and it is particularly useful in diagnosing multifocal osteomyelitis (Offiah 2006).

High-quality images and pinhole collimation are needed to identify the pathology. A bone scan is recommended when clinical suspicion of infection is high, and radiographs and ultrasound are not conclusive (MRI may be preferred by some clinicians in this situation).

The characteristic features of acute osteomyelitis are:

- Increased uptake in all three phases (perfusion, blood pool, and bone metabolism phases) of the triple-phase bone scan (Aigner et al. 1996).

17.5 Establishing the Diagnosis

An outline of the process of establishing a diagnosis of the cause of diminished movements of a limb in the neonate is shown in Table 17.1.

Table 17.1 An outline of the process of establishing the diagnosis of diminished movement of a limb in the neonate

History			
History of difficult labor, large baby, or shoulder dystocia often present	–		History of difficult labor, large baby, or shoulder dystocia often present
–	Definite history of baby having moved the limbs normally soon after birth		–
–	History of fever preceding reduction in movement of limb may be present		–
Physical examination			
No tenderness over any joint or bone on palpation No pain on passive movement of joints of the affected limb	Tenderness over joint pain on passive movement	Tenderness over bone pain on passive movement	Tenderness over joint or bone Pain on passive movement

Table 17.1 (continued)

–	–	–	Palpable deformity or discontinuity of bone
Unilateral absence of primitive reflexes such as Moro and grasp (in the absence of pain on passive movement of the limb)			
Working diagnosis: Obstetric palsy	Working diagnosis: Septic arthritis	Working diagnosis: Acute osteomyelitis	Working diagnosis: Birth injury
Investigations			
Plain radiograph not indicated (unless physical examination features noted in column three are also present)	Plain radiograph: One or more of the following: Soft tissue swelling Distended capsular shadow Widening of joint space Joint subluxation or dislocation	Plain radiograph One or more of the following: Soft tissue swelling Periosteal reaction Bony erosion in the metaphysis	Plain radiograph: Discontinuity of bone or alteration of normal alignment of joint (in epiphyseal separation)
Ultrasound scan not indicated	Ultrasound scan: One or more of the following: Joint effusion Joint subluxation Joint dislocation	Ultrasound scan: One or more of the following: Deep soft tissue swelling Elevation of periosteum by a thin layer of fluid Subperiosteal abscess Cortical erosion	Ultrasound scan: indicated if epiphyseal separation is suspected and epiphysis is cartilaginous
MRI not indicated initially	MRI not indicated	MRI: indicated if ultrasound is equivocal Low signal on T1-weighted images High signal on T2-weighted and STIR sequences	MRI not indicated
Isotope bone scan not indicated	Isotope scan not indicated	Isotope scan: indicated only if radiograph and MRI are equivocal but clinical features suggest osteomyelitis Increased uptake perfusion, blood pool, and bone metabolism phases of the triple-phase bone scan	Isotope scan not indicated
–	White blood cell counts, ESR, and CRP may be elevated	White blood cell counts, ESR, and CRP may be elevated	–
–	Blood culture may be positive	Blood culture may be positive	–
Diagnosis:			
Birth palsy	Septic arthritis	Acute osteomyelitis	Birth injury

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18 Deformity of the Neck and Limitation of Movement of the Neck in the Neonate and Infant

Randall T. Loder

18.1 Introduction

Deformity of the neck and limited motion are commonly seen in pediatric orthopedics. The problem may be simply due to an intrinsic cervical issue or may be the manifestation of other underlying problems.

The important questions that need to be answered while trying to make a diagnosis of the nature and cause of neck deformity in the newborn are listed below.

18.2 Questions to Establish a Diagnosis

- **Was trauma involved?**
- **Was the onset gradual or abrupt?**
- **Are there associated constitutional symptoms (fever, chills, weight loss)?**
- **Are there associated neurologic symptoms or signs (bulbar or extremity involvement)?**
- **Is there torticollis?**
- **Is there shortening of the neck?**
- **Is the deformity continuous or intermittent?**
- **Are there any other associated medical conditions (e.g., gastroesophageal reflux) or syndromes (e.g., craniofacial)**

Was trauma involved?

Traumatic neck deformity typically indicates a fracture as the diagnosis. In the neonate or infant, cervical spine fractures can occur during a difficult birth. Prolonged labor and difficult delivery may indicate a diagnosis of congenital muscular torticollis.

Was the onset gradual or abrupt?

If abrupt, fractures and infections are likely diagnoses. If gradual, then inflammatory conditions (sepsis), neoplasms, and progressive congenital deformity are likely diagnoses.

Are there associated constitutional symptoms?

Fevers and chills are frequently associated with infections and weight loss with malignant neoplasms; both of these are rare in the neonate or infant.

Are there associated neurologic symptoms?

Bulbar (cranial nerve) involvement and upper extremity weakness point to an Arnold-Chiari malformation or CNS tumor as the underlying cause.

Is their associated pain?

Painless deformities are typically seen in congenital muscular torticollis, neoplasms, and progressive congenital anomalies. Pain is typically associated with infection and fracture.

Is their torticollis?

Torticollis is defined as the lateral deviation of the neck to one side and rotation to the opposite side. It may indicate a problem at the C1–C2 articulation.

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_18

Is their shortening of the neck?

Shortening of the neck is often seen in Klippel-Feil syndrome due to the fusion of the cervical vertebrae. It can also be seen in osteogenesis imperfecta or other causes of basilar impression.

Is the deformity continuous or intermittent?

Intermittent torticollis is often a sign of Sandifer syndrome or paroxysmal torticollis of infancy.

Are there any other associated medical conditions?

Children with gastroesophageal reflux may exhibit intermittent torticollis (Sandifer syndrome). Children with craniofacial syndromes (cleft lip/palate and synostoses) also exhibit deformities of the cervical spine.

18.3 Physical Examination

18.3.1 Look

A visual inspection will determine if there is true torticollis or a simple head tilt. Also look to see if the neck is of appropriate height or short. Elevation of the scapula points to a Sprengel deformity with or without associated Klippel-Feil anomalies. If the child is febrile and ill appearing, it points toward discitis or vertebral osteomyelitis.

18.3.2 Feel

Feel for tightness of the sternocleidomastoid – this will point toward congenital muscular torticollis. Tenderness to palpation points toward an infection or trauma (fracture, interspinous ligament strain).

18.3.3 Move

The most important part of the physical examination is the range of motion of the cervical spine. Is there torticollis, and is there tightness of the sternocleidomastoid muscle? If tightness is present, on which side is the tightness relative to the torticollis? Are there other congenital deformities (e.g., craniofacial deformity, scoliosis, hip instability)? Does the child demonstrate marked irritability with passive range of motion of the neck? If so, this points toward infection.

18.4 Investigations to Confirm the Diagnosis

18.4.1 Radiography

Standard AP and lateral radiographs are obtained. These may demonstrate fusions of the cervical vertebrae and other congenital vertebral anomalies in the Klippel-Feil syndrome. In children with a simple congenital muscular torticollis, imaging of the hip (either hip ultrasound or standard pelvis imaging – dependent upon the age of the infant) should be performed to ensure there is no developmental hip dysplasia.

If the anatomy cannot be readily seen on plain radiographs, then advanced imaging is performed. CT scans with our without three-dimensional reconstruction are invaluable to get a complete picture of the vertebral anatomy. This is especially so at the craniocervical junction (occiput to C2). The exact anatomic abnormality is not often delineated until several years of age due to the markedly cartilaginous nature of the vertebral bodies in infancy.

18.4.2 Special Imaging Studies

These studies, such as a bone scan and/or MRI, are needed if the above are negative, and there is a concern for an early osteomyelitis/pyomyositis or vertebral end-plate fracture not visualized on plain radiographs. CT scans with coronal, sagittal, and three-dimensional reconstructions are frequently needed to completely evaluate congenital deformities (e.g., hemivertebra, basilar invagination).

18.4.3 Laboratory Studies

Appropriate laboratory studies are also obtained where indicated (e.g., CBC with differential, ESR, CRP if concerned about infection).

18.5 Differential Diagnosis

18.5.1 Torticollis

Torticollis is a combined head tilt and rotatory deformity. Torticollis indicates a problem at C1–C2, because 50 % of the cervical spine rotation occurs at this joint. A head tilt alone indicates a more generalized problem in the cervical spine. The differential diagnosis of torticollis is large and can be divided into osseous and non-osseous categories. The non-osseous types include congenital muscular torticollis, Sandifer syndrome, and paroxysmal torticollis of infancy. The osseous types include occipitocervical synostosis, basilar impression, and odontoid anomalies (MacAlister 1983). Non-osseous, non-congenital muscular torticollis accounts for ~20 % of all childhood torticollis (central nervous system lesions such as Arnold-Chiari malformation, tumor or ocular disorders) (Ballock and Song 1996).

18.5.2 Congenital Muscular Torticollis

Congenital muscular torticollis is the most common cause of torticollis in the infant and presents at a median age of 2 months (Ho et al. 1999). The deformity is caused by contracture of the sternocleidomastoid muscle, with the head tilted toward the involved side and the chin rotated toward the opposite shoulder (Fig. 18.1). Many children have a history of a primiparous birth or a breech or difficult delivery although it can also be seen in children with normal births and those born by cesarean section (Ho et al. 1999; Ling 1976; MacDonald 1969). Plain radiographs of the cervical spine should be obtained to rule out associated congenital anomalies and are normal aside from the head tilt and spine rotation. Ultrasound is helpful to distinguish between congenital muscular torticollis from other pathologies in the neck (Chen et al. 2005; Dudkiewicz et al. 2005). If there is any concern regarding the hips, appropriate imaging (e.g., ultrasonography, radiography) is performed due to the increased incidence of DDH in these children (Weiner 1976). MRI is not needed for the diagnosis.

18.5.3 Sandifer Syndrome

This is a syndrome of gastroesophageal reflux, often from a hiatal hernia, with subsequent abnormal posturing of the neck and trunk, usually torticollis (Murphy and Gellis 1977; Ramenofsky et al. 1978). The torticollis is a way by which the child can decrease the esophageal discomfort resulting from the reflux. Most present in infancy where the incidence of gastroesophageal reflux is high (up to 40 %) (Darling et al. 1974). The diagnosis of gastroesophageal reflux is often overlooked in a child with torticollis. Physical examination demonstrates the absence of a tight and short sternocleidomastoid muscle which excludes congenital muscular torticollis. Plain radiographs of the cervical spine eliminate congenital anomalies or dysplasias; contrast studies of the upper gastrointestinal tract usually demonstrate the hiatal hernia and gastroesophageal reflux (Darling 1975). Esophageal pH studies may be necessary (Jolley et al. 1978).

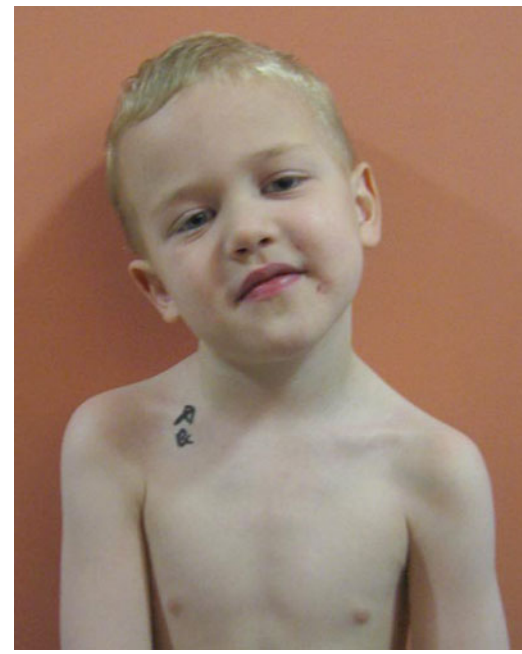

Fig. 18.1 A child with a typical congenital muscular torticollis

18.5.4 Paroxysmal Torticollis of Infancy

Paroxysmal torticollis of infancy is a rare, episodic torticollis, usually in girls, which lasts for minutes to days with spontaneous recovery (Drigo et al. 2000; Parker 1989; Snyder 1969). The average age of onset is 3 months. The attacks are usually in the morning and last from minutes to days from 1 to 3–4 episodes per month. The torticollis alternates from side to side which is instrumental in making the diagnosis. There may also be an associated scoliosis and abnormal eye movements. A positive family history of migraine headaches is often present (Al-Twaijri and Shevell 2002; Giffin et al. 2002). Physical examination is entirely normal when the episode is not present.

18.5.5 Klippel-Feil Syndrome

Klippel-Feil syndrome is a congenital fusion of the cervical vertebrae. Other associated anomalies are often present both in the musculoskeletal and other organ systems and include Sprengel deformity (Fig. 18.2) and/or omovertebral bar (Fig. 18.3), scoliosis (both congenital and idiopathic) (Hensinger et al. 1974), congenital limb deficiency (Thomsen et al. 2000), renal anomalies (Moore et al. 1975), deafness (Stark and Borton 1973), synkinesis (mirror movements) (Gunderson and Solitare 1968), pulmonary dysfunction (Baga et al. 1969), and congenital cardiovascular issues (Abbas et al. 2006; Kawano et al. 2008; Nora et al. 1961). Physical examination demonstrates a low posterior hairline and a short neck (Fig. 18.4), with variable degrees of reduced neck motion. In the infant, pas-

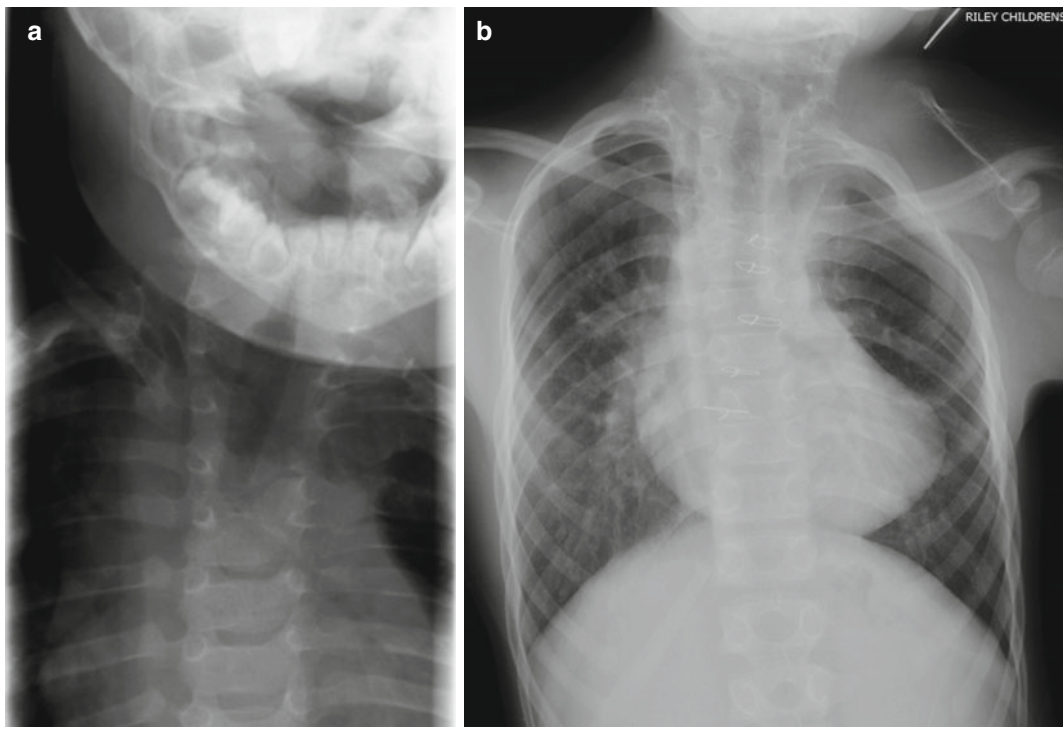

Fig. 18.2 A boy with significant congenital cardiac defects. (**a**) A cervical spine AP radiograph at 14 months of age demonstrates a lateral tilt to the right. (**b**) An AP chest radiograph at 8 years of age demonstrates a proximally positioned and hypoplastic left scapula. (**c**) At age 11 years, a 3D CT scan reconstruction was performed. The AP (*1*) and posterior (*2*) views of the scapula clearly demonstrate a proximally, high-riding scapula on the left. The AP (*3*) and posterior (*4*) views of the scapula demonstrate the tilt to the right along with multiple congenital vertebral anomalies

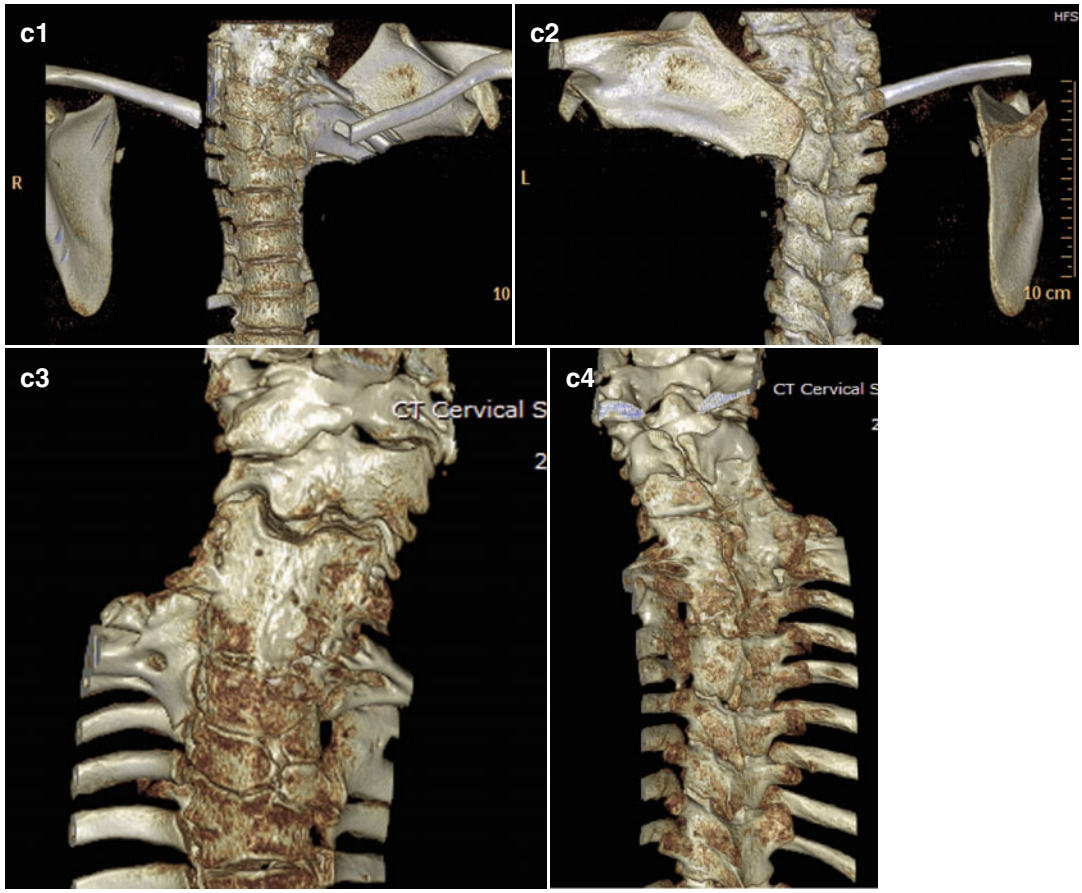

Fig. 18.2 continued

sive range of motion of the neck is often normal, making it more difficult to diagnose. Radiographs demonstrate a wide range of deformity, ranging from a simple block vertebrae to multiple and bizarre anomalies. The fusions become more apparent as the child ages (Samartzis et al. 2008). Children with Klippel-Feil syndrome should be further evaluated for other organ system problems. A general pediatric evaluation is performed to determine if there are congenital cardiac, neurologic, or other abnormalities. Renal ultrasonography should be done in all children (Drvaric et al. 1987). MRI is performed for concerns of neurologic issues (Ritterbusch et al. 1991). Klippel-Feil-like congenital anomalies are also seen in the fetal alcohol syndrome (Tredwell et al. 1982) and craniofacial syndromes (cleft lip and palate (Sandham 1986; Ugar and Semb 2001)) and craniosynostosis syndromes (Crouzon, Pfeiffer, Apert, Goldenhar, Saethre-Chotzen) which exhibit cervical vertebral fusion, atlanto-occipital fusion, and butterfly vertebrae (Anderson et al. 1997; Anderson et al. 1996; Hemmer et al. 1987; Louis and Argenta 1987; Moore et al. 1995; Sherk et al. 1982; Tsirikos et al. 2003) (Fig. 18.5).

18.5.6 Other Bony Causes of Torticollis and Short Neck

These rare conditions include basilar impression and various occipito-atlantal congenital anomalies. Basilar impression is skull floor indentation by the upper cervical spine. The odontoid migrates proximally and may eventually protrude into the foramen magnum. It can be primary or secondary.

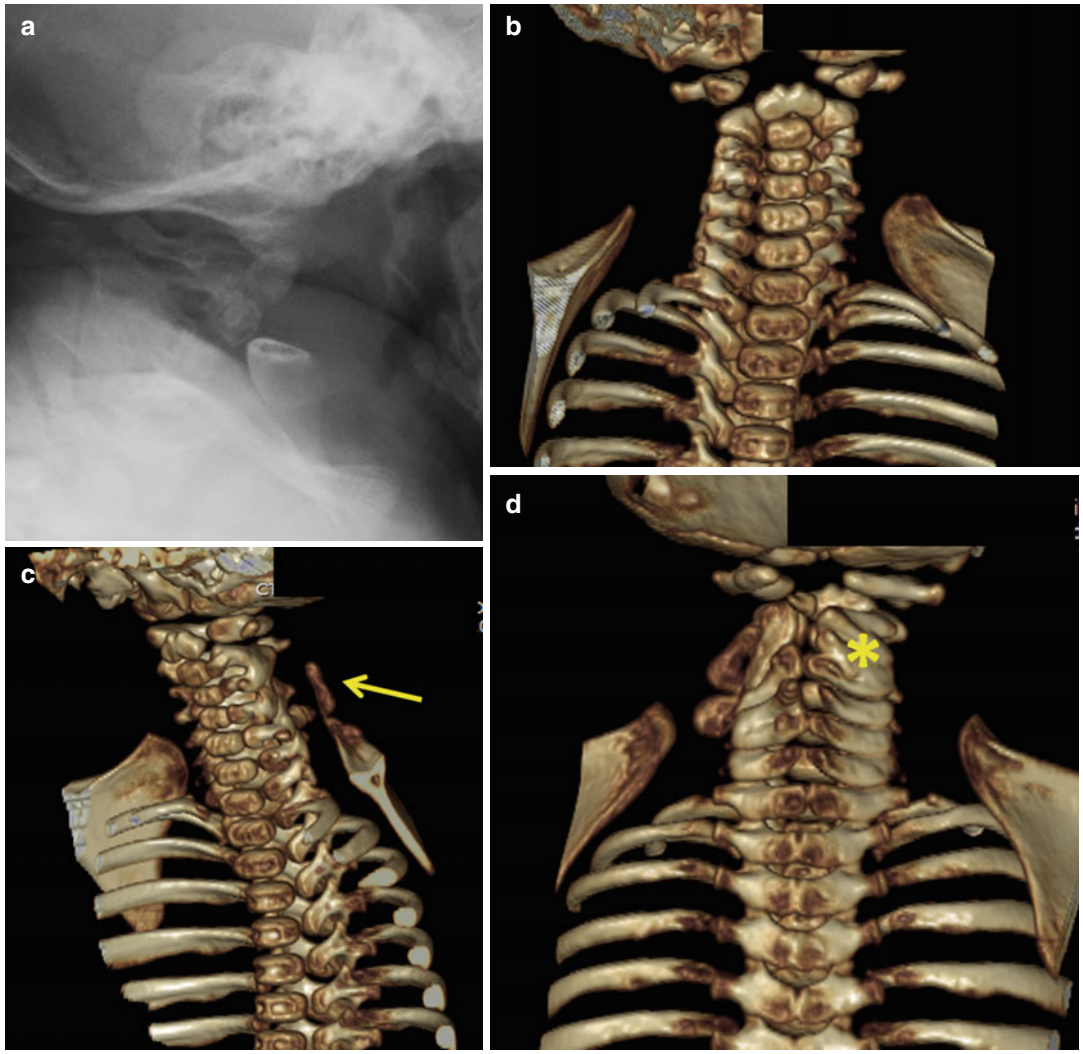

Fig. 18.3 A 3-month-old girl with shoulder asymmetry. (**a**) The lateral cervical spine radiograph is difficult to interpret, but concerning because a cervical vertebral anomaly is present. A CT scan with three-dimensional reconstructions was performed. (**b**) The AP 3D CT scan reconstruction demonstrates no anterior vertebral anomalies, but the left scapula is hypoplastic, rotated, and higher than the right scapula. (**c**) An oblique 3D CT scan reconstruction demonstrates that the right scapula is elevated along with an omovertebral bar (*yellow arrow*) between the scapula and the posterior elements of the cervical spine. (**d**) The posterior 3D CT reconstruction demonstrates fusion of the right-sided posterior cervical laminae and facet joints (*asterisk*) as well as spina bifida occulta of C1–C3

Primary basilar impression is a congenital abnormality often associated with other vertebral defects (e.g., Klippel-Feil syndrome, odontoid abnormalities, atlanto-occipital fusion, and atlas hypoplasia). Secondary basilar impression develops from softening of the basilar bone from metabolic bone disease (e.g., osteogenesis imperfecta (Harkey et al. 1990; Hayes et al. 1999; Ibrahim and Crockard 2007; Kovero et al. 2006; Pozo et al. 1984; Rush et al. 1989; Sawin and Menezes 1997)), dwarfisms (achondroplasia (Yamada et al. 1981), hypochondroplasia (Wong and Fung 1991)), or other dysplasias (e.g., neurofibromatosis (Isu et al. 1983)). These patients typically present with a short neck (de Barros et al. 1968). Torticollis may be present. Neurologic findings are often present

(Michie and Clark 1968) but are very difficult to notice in the very young child. The neurologic involvement can either be pyramidal-like (motor weakness with limb paresthesias) or cerebellar-like (in-coordination with ataxia, dizziness, nystagmus, and lower cranial nerve involvement due to an Arnold-Chiari malformation (Teodori and Painter 1984)). Plain radiographs are very difficult to interpret in the infant leading to difficulty in diagnosis. CT scan with sagittal plane reconstruction best demonstrates the bony anatomy, and MRI best delineates the neural anatomy.

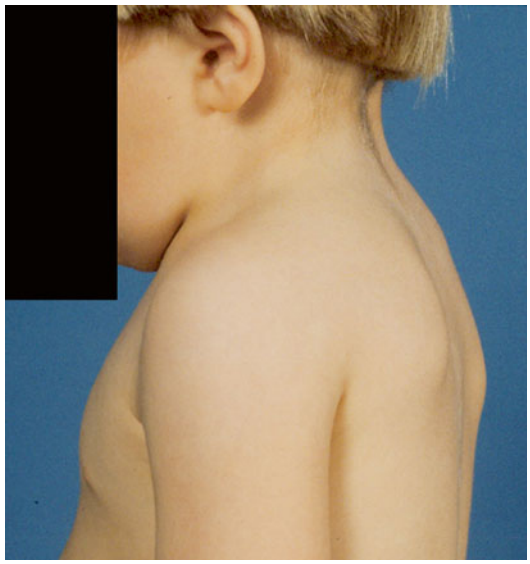

Fig. 18.4 A clinical photograph of a child with Klippel-Feil syndrome. Note the low hairline and short neck

Congenital bony anomalies at the atlanto-occipital junction (various degrees of atlanto-occipital synostoses with or without basilar impression) demonstrate many different deformities (Gholve et al. 2007; Hosalkar et al. 2008). These may be cartilaginous initially and not appear on plain radiographs until the child becomes more mature. Posterior congenital fusion of C2–C3 is a clue that occiput–C1 anomalies, or other more distal cervical fusions, may be present (Fig. 18.5). Physical examination is similar to infants with the Klippel-Feil syndrome: short, broad necks; restricted neck motion; low hairline; high scapula; and torticollis (Al Kaissi et al. 2006; Bharucha and Dastur 1964; Halanski et al. 2006; McRae and Barnum 1953). Again CT

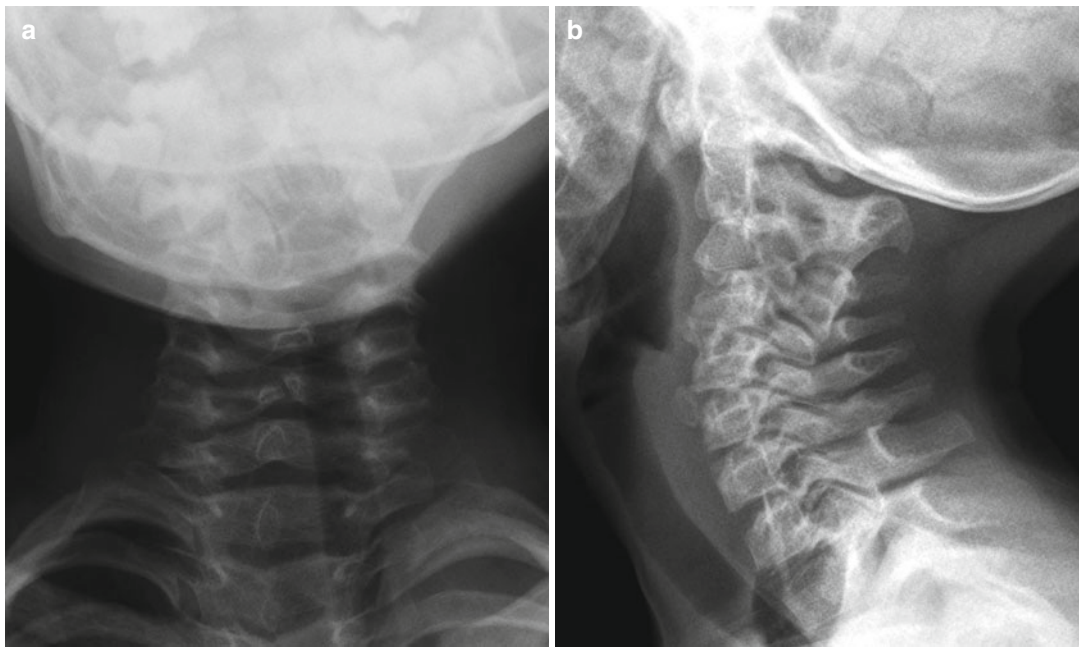

Fig. 18.5 A 5-year-old boy with known Goldenhar syndrome recently developed a slight tilt of the cervical spine. AP (1.8.5.1) and lateral (1.8.5.2) radiographs of the spine demonstrate a slight lateral tilt to the left, along with Klippel-Feil-like vertebral anomalies at the C2–C4 level along with fusion of the C1–C2 posterior elements

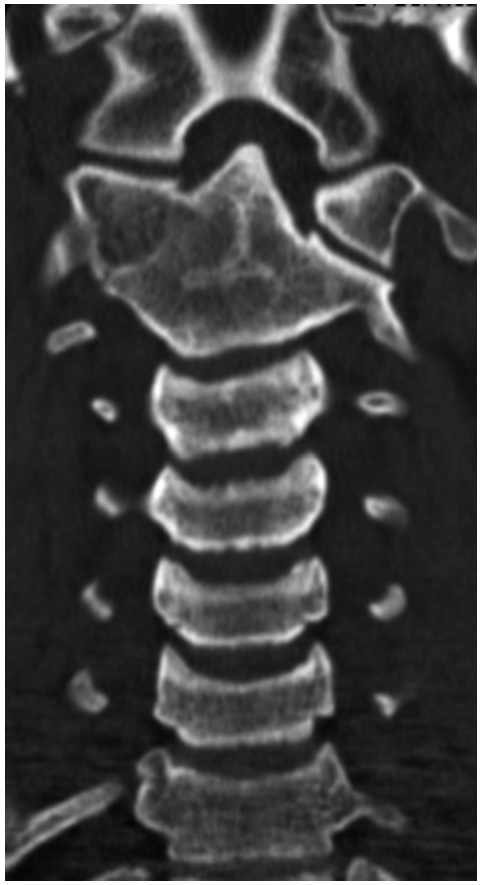

Fig. 18.6 A 12-year-old girl with a slowly progressive head tilt. Note the fusion of the right-sided C1 and C2 bodies and the absence of the left C2 vertebral body. This resulted in a slowly progressive head tilt due to the congenital vertebral anomaly

best defines the bony anatomy (Fig. 18.6). Another anomaly is unilateral absence of C1. This is a hemiatlas or congenital scoliosis of C1 (Doubousset 1986). They typically present as a torticollis, and most present at birth. The sternocleidomastoid muscle is not tight, although regional aplasia of the muscles in the nuchal concavity of the tilted side is noted. Neck motion is

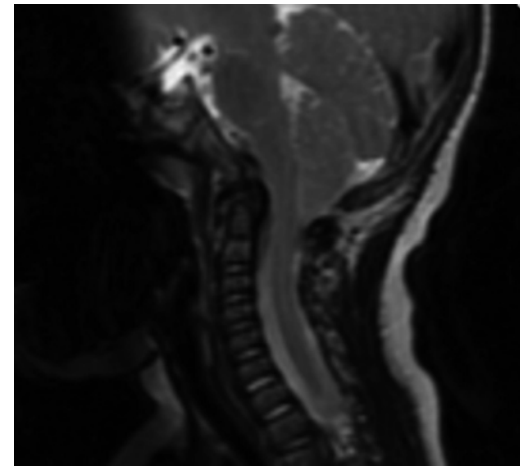

Fig. 18.7 A 4-month-old boy presented with torticollis but without tightness of the sternocleidomastoid muscle and having full cervical spine motion. The MRI scan demonstrates an Arnold-Chiari malformation with the cerebellar tonsils protruding into the foramen magnum and ending at the level of the C2 synchondrosis

variable. In all of these atlanto-occipital anomalies, plain radiographs rarely demonstrate the anomaly. CT scan is usually necessary to make the diagnosis.

18.5.7 Other Rare Conditions Causing Torticollis

Infants with posterior fossa and cervical cord tumors can present with torticollis (Dörner et al. 2007; Giuffrè et al. 1981; Kumandas et al. 2006; Marmor et al. 1990; O'Brien et al. 2001; Taboas-Perez and Rivera-Reyes 1984; Visudhiphan et al. 1982). The initial diagnosis is often congenital torticollis, obstetric birth palsy, muscular dystrophy, or cerebral palsy (Giuffrè et al. 1981). The often overlooked signs of the tumor are spinal rigidity, early spinal deformity, and spontaneous or induced vertebral pain. In infants, pain is often

expressed as irritability and restlessness (Rauch et al. 2001). Imaging of a child with a potential central nervous system tumor first includes radiographs of the skull and cervical spine followed by CT and MRI scans. Arnold-Chiari malformations (Dure et al. 1989; Wilkins and Brody 1971) may also present with torticollis (Fig. 18.7). The same for tumors is used (Dure et al. 1989). Finally, ocular pathology accounts for up to 1/3 of children with no obvious orthopedic cause of torticollis (Williams et al. 1996). These children typically present around 1 year of age with an atypical torticollis (Rubin and Wagner 1986). An ocular cause is likely if the head is tilted but not rotated or if the tilt changes when the child is lying versus sitting or standing up. The face and head exhibit many different and bizarre positions which optimize visual acuity for the child. Children with ocular torticollis have normal cervical spine motion without sternocleidomastoid muscle tightness as seen in congenital muscular torticollis. Ophthalmologic evaluation results in the diagnosis.

18.5.8 Other Rare Causes Resulting in Limitation of Neck Motion

Septic processes (e.g., cervical discitis/vertebral body osteomyelitis) are extremely rare in the young child (Ring, 1995 #85; Wenger, 1978 #182; Fang, 1983 #389; Hsu, 1984 #388; Govender, 2007 #1022). When present, the child will demonstrate varying degrees of limitation in active neck motion. Passive motion will result in considerable irritability to the child. The child may or may not be febrile. The cause may either be pyogenic bacteria or *Mycobacteria*. Plain radiographs may initially be normal, with MRI greatly assisting in the diagnosis. Cultures may or may not grow an organism.

18.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis of a deformity of the neck or limited motion of the neck in infancy is shown in Table 18.1.

Table 18.1 Establishing the diagnosis of neck deformity and limited motion in the infant

History							
No torticollis	May demonstrate torticollis	Intermittent torticollis	Intermittent torticollis	Intermittent torticollis	Persistent torticollis	Persistent torticollis	Persistent torticollis
No fever	No fever	No fever	No fever	No fever	No fever	No fever	History may be + for fever
		History of gastroesophageal reflux	Family history of migraine headaches				
Physical examination							
Not febrile	Not febrile	Not febrile	Not febrile	Not febrile	Not febrile	Not febrile	± febrile
± Short neck, low posterior hairline	± Short neck, low posterior hairline	Normal-appearing neck	Normal-appearing neck	Normal-appearing neck but held in bizarre positions	± Short neck, low posterior hairline	Tight sternocleidomastoid muscle	Normal-appearing neck or anterior fullness
Variable degrees of reduced neck motion	Variable degrees of reduced neck motion	Normal neck motion	Normal neck motion	Normal neck motion	Variable degrees of reduced neck motion	↓ External rotation on the side of the tight muscle and ↓ lateral flexion on the opposite side	Decreased neck motion with marked irritability
Sternocleidomastoid muscle usually normal	Sternocleidomastoid muscle usually normal	Sternocleidomastoid muscle normal	Sternocleidomastoid muscle normal	Sternocleidomastoid muscle normal	Sternocleidomastoid muscle normal	Short and atrophic sternocleidomastoid muscle	Sternocleidomastoid muscle normal
	Cleft lip/palate, hemifacial microsomia, craniosynostoses				Varying degrees of plagiocephaly	Varying degrees of plagiocephaly	
Investigations							
Plain radiograph of the cervical spine May show: Congenital fusions of the vertebral bodies and/or posterior elements	Plain radiograph of the cervical spine May show: Congenital fusions of the vertebral bodies and/or posterior elements	Plain radiograph of the cervical spine will be normal	Plain radiograph of the cervical spine will be normal	Plain radiograph of the cervical spine will be normal	Plain radiograph of the cervical spine May show: Anomalies at the occipitocervical junction	Plain radiograph of the cervical spine will be normal aside from lateral tilt and rotation Imaging of the hips may demonstrate various degrees of DDH	Plain radiograph of the cervical spine: May show: No abnormalities Decreased disk height and vertebral body erosions Anterior soft tissue fullness Varying amounts of kyphosis

Ultrasound not indicated	Ultrasound not indicated	Ultrasound not indicated	Ultrasound not indicated	Ultrasound not indicated	Ultrasound not indicated	Ultrasound will rule out other types of neck mass	Ultrasound may demonstrate an anterior cervical abscess
CT scan: Sagittal and coronal reconstructions will demonstrate osseous fusions of the cervical vertebrae ± other bony anomalies (hemivertebrae)	CT scan: Sagittal and coronal reconstructions will demonstrate osseous fusions of the cervical vertebrae ± other bony anomalies (hemivertebrae)	CT scan not indicated	CT scan not indicated	CT scan not indicated	CT scan: Sagittal and coronal reconstructions will demonstrate the anomalous vertebral anatomy (hemivertebrae, basilar impression)	CT scan not indicated	CT scan may be indicated to further study the bony anatomy
MRI not indicated to make the diagnosis May later demonstrate neural axis abnormalities (Arnold-Chiari, basilar impression)	MRI not indicated to make the diagnosis	MRI not indicated to make the diagnosis	MRI not indicated to make the diagnosis	MRI not indicated to make the diagnosis	MRI not indicated to make the diagnosis May later demonstrate neural axis abnormalities (Arnold-Chiari, basilar impression)	MRI not indicated to make the diagnosis	MRI will demonstrate disc and vertebral body involvement with or without anterior abscesses
Other special studies not usually necessary to make the diagnosis Renal ultrasound obtained after the diagnosis	Other special studies not usually necessary to make the diagnosis Renal ultrasound obtained after the diagnosis	GI studies will demonstrate gastroesophageal reflux and hiatal hernia	Other special studies not usually necessary to make the diagnosis	Ophthalmologic evaluation will demonstrate ocular nerve palsies/ anomalies	Other special studies not usually necessary to make the diagnosis Renal ultrasound obtained after the diagnosis	Other special studies not usually necessary to make the diagnosis	Other special studies not usually necessary to make the diagnosis

(continued)

Table 18.1 (continued)

Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis	Esophageal pH studies abnormal	Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis		Gram stain will demonstrate pyogenic bacteria or acid-fast bacilli WBC, ESR, and CRP often elevated Tissue/abscess cultures may be positive for either pyogenic bacteria or acid-fast bacilli
Diagnosis							
Klippel-Feil syndrome	Klippel-Feil-like deformities associated with Fetal alcohol syndrome Cleft lip/palate Craniosynostosis syndromes Hemifacial microsomia	Sandifer syndrome	Paroxysmal torticollis of infancy	Ocular torticollis	Atlanto-occipital anomalies Occipitocervical synostosis Hemiatlas of C1 Basilar impression	Congenital muscular torticollis	Discitis/osteomyelitis

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Part III

The Toddler and the Pre-school Child

The Child with Multiple Deformities

19

Benjamin Joseph

19.1 Introduction

The pediatric orthopedic surgeon is sometimes confronted with a child with multiple deformities, and before any treatment can be considered, it is important that the precise nature of the underlying condition is established.

Multiple limb deformities may be associated with major limb deficiencies (Fig. 19.1); the diagnosis is usually more straightforward in such children. This chapter will deal with multiple deformities that are not associated with limb deficiencies. Some of these conditions manifest as deformities at birth, while others develop deformities in infancy or childhood. The deformities may be at the joints, or the long bones of the limbs may be deformed; treatment will depend on the site of the deformity. In some conditions the deformities are associated with pain, while pain is not a feature in some conditions. These distinctions will also help in diagnosing the cause of the deformities (Table 19.1).

Establishing the specific diagnosis is also important to determine the natural history of the deformities. For example, some deformities seen at birth in congenital contractural arachnodactyly (Beals syndrome) may improve (Viljoen 1994), while deformities in classical arthrogryposis do not resolve and even have a propensity to recur following surgical correction.

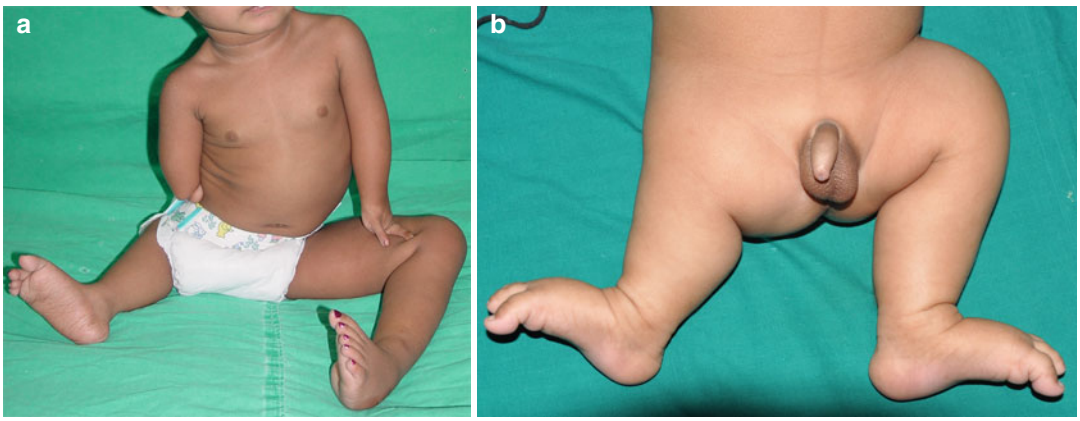

Fig. 19.1 Deformities of the lower limbs in a child with multiple congenital deficiencies of the upper and lower limb (**a**) and deformities of the lower limbs of another child with major congenital deficiencies (**b**)

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Table 19.1 Site, onset, and nature of deformities in children with multiple deformities

Onset	Site of deformities			
	Joint deformities		Bony deformities	
At birth	Painful: Very unusual	Painless: E.g., multiple congenital contractures	Painful: E.g., severe forms of osteogenesis imperfecta	Painless: E.g., campomelic dysplasia
Infancy or childhood	Painful: E.g., juvenile idiopathic arthritis	Painless: E.g., paralytic deformities	Painful: E.g., metabolic bone disease such as active rickets	Painless: E.g., milder forms of osteogenesis imperfecta

19.2 Questions to Establish a Diagnosis

- **Were the deformities present from birth, or did the deformities develop later?**
- **Did the deformities develop after a febrile episode, and what was the interval between the febrile episode and the onset of the deformities?**
- **Are the deformities symmetric?**
- **Are the deformities at the joint, or are they in the shaft of the bone of the limbs?**
- **If the deformities are in bones, are they mainly in the diaphysis of the bone or in the ends of the bone?**
- **If the deformities are at the joint, are the joints swollen, tender, or painful when moved?**
- **Are the stature and body proportions normal?**
- **Is there associated upper or lower motor neuron paralysis?**

Were the deformities present from birth, or did the deformities develop later?

This distinction is important as it may help to narrow down the possible diagnoses. Multiple joint deformities are seen at birth in children with multiple congenital contractures (arthrogryposis), while joint deformities of inflammatory arthritis develop only later.

Did the deformities develop after a febrile episode, and what was the interval between the febrile episode and the onset of the deformities?

Deformities following multifocal osteomyelitis or multifocal septic arthritis manifest after months following damage to the growth mechanism at the time of the acute illness. Paralytic deformities in polio that are secondary to muscle imbalance may develop some months after the onset of paralysis, while postural deformities may develop very soon after the onset of paralysis.

Are the deformities symmetric?

Symmetric deformities of the bone are more likely to be due to congenital or developmental abnormalities, while asymmetric deformities are more frequently acquired.

Are the deformities at the joint, or are they in the shaft of the bone of the limbs?

Identifying the site of the deformities will help in establishing the diagnosis; asymmetric deformities of the shaft of the long bones are seen in some children with osteogenesis imperfecta, while symmetric deformities of the long bones may be seen in some forms of skeletal dysplasia (e.g., campomelic dysplasia) or in children with vitamin D-resistant rickets. Occasionally there may be a combination of joint and bone deformities as in Bruck syndrome which has features of arthrogryposis and osteogenesis imperfecta (Leroy et al. 1998).

If the deformities are in the bones, are they mainly in the diaphysis of the bone or in the ends of the bone?

Diaphyseal deformities developing after birth suggest that there is a primary disease of the bone rendering the bone weak and susceptible to either bending or breaking as in metabolic bone disease or osteogenesis imperfecta.

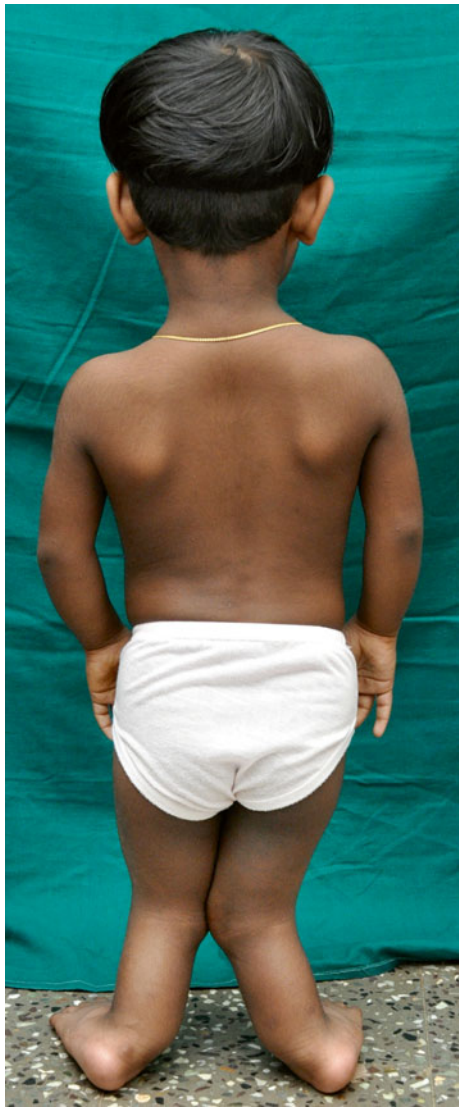

Fig. 19.2 This child has symmetric deformities of the knees and ankles, altered body proportions, and reduced stature. All these features suggest that she has a form of skeletal dysplasia

Metaphyseal deformities are more likely to be related to growth abnormalities.

If the deformities are at the joint, are joints swollen, tender, or painful when moved?

Painful or swollen joints are characteristic of inflammatory joint disease, the commonest of which is juvenile idiopathic arthritis; however, juvenile psoriatic arthritis, progressive pseudorheumatoid arthropathy of childhood, and the rare conditions such as carpal-tarsal osteolysis and neonatal-onset multisystem inflammatory disease (NOMID) may manifest with deformed painful joints (Kim et al. 2014; Choi et al. 1993; Ekbote et al. 2013).

Are the stature and body proportions normal?

Altered body proportions and reduced body stature suggest that the child has a form of skeletal dysplasia (Fig. 19.2).

Is there associated upper or lower motor neuron paralysis?

Joint deformities may develop in paralytic conditions as a consequence of muscle imbalance, due to static posturing or due to fibrosis of muscles.

19.3 Physical Examination

19.3.1 Look

Observe the pattern of deformities and document them. Some conditions have characteristic patterns of deformities that facilitate a diagnosis (Fig. 19.3). The pattern of deformity may also be diagnostic of underlying pathology. For example, bilateral symmetric rigid hyperextension deformities of the knees are invariably on account of contracture of the quadriceps muscle, and often there is an underlying neurological cause for the contracture. Note if there are skin dimples over the apex of deformities; this is characteristically seen in campomelic dysplasia and in deformities associated with limb deficiencies.

19.3.2 Feel

Palpate the long bones throughout their entire length and note the site of deformity. Palpate joints that are deformed to look for local warmth, effusion, and synovial hypertrophy.

19.3.3 Move

Put each joint through the full range of motion passively and document the range of motion and note if the movement is painful or associated with muscle spasm.

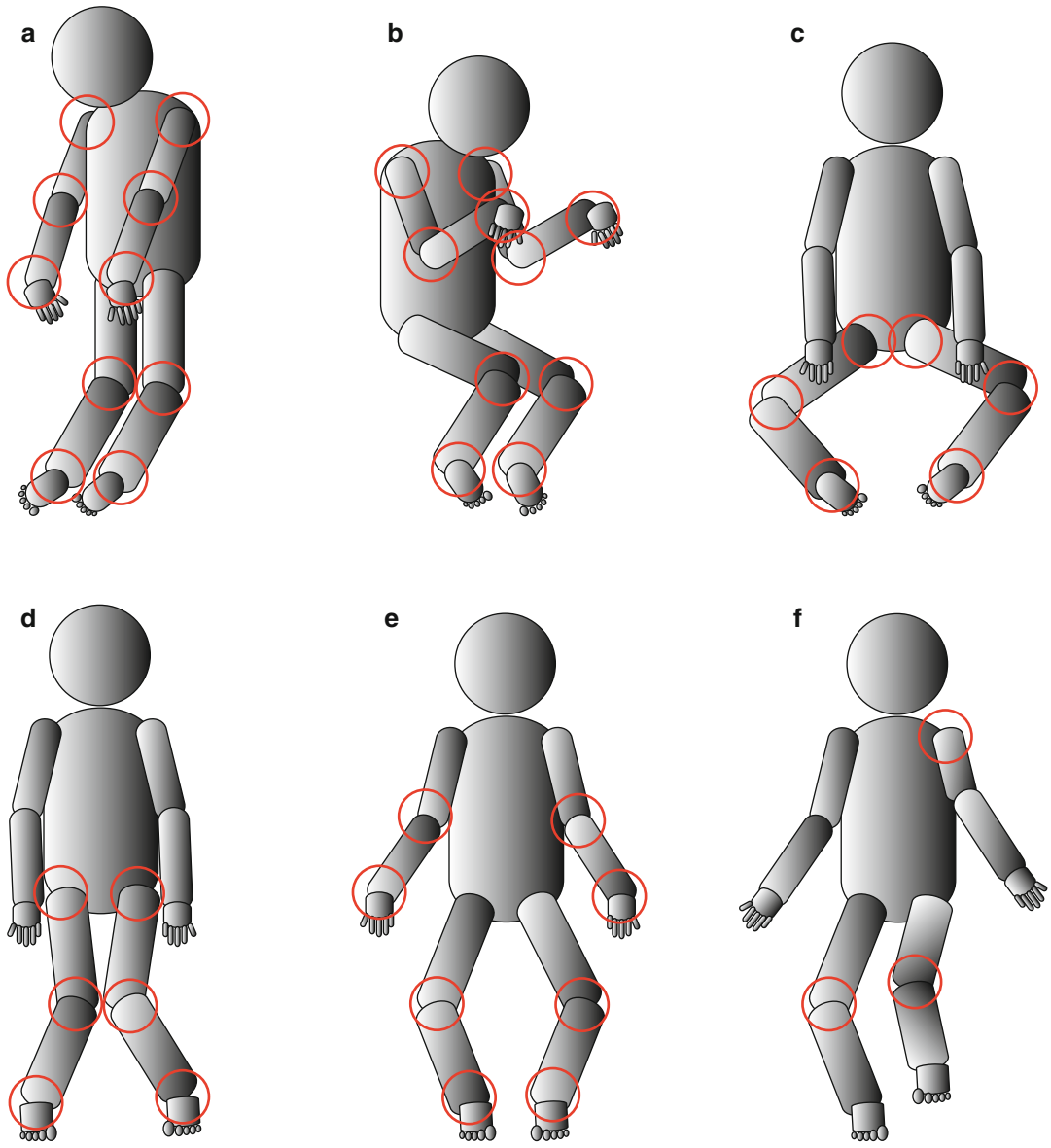

Fig. 19.3 Diagrams showing patterns of deformities in different conditions. Patterns of symmetric deformities in the sagittal plane (extension or flexion deformities) are typically seen in arthrogryposis (**a**, **b**). Symmetric deformities including flexion, abduction, and external rotation of the hips, flexion of the knees, and equinus of the ankles are characteristic of complete sacral agenesis (**c**). Symmetric coronal plane deformities (varus and valgus) involving the upper and lower limbs are seen in skeletal dysplasias (**d**, **e**). Asymmetric deformities in more than one plane with shortening of some segments of the bone are characteristically seen after growth plate damage at multiple sites following multifocal osteomyelitis or septic arthritis (**f**)

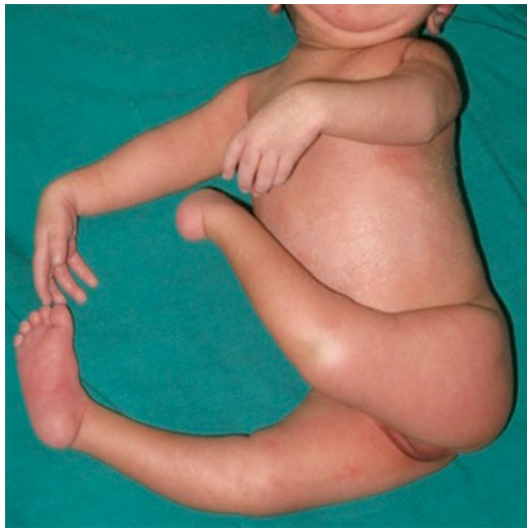

Fig. 19.4 Deformities of the joints of the upper and lower limbs in a child with classical arthrogryposis

19.4 Investigations to Confirm the Diagnosis

Plain Radiographs

Plain radiographs are essential if there are bony deformities.

If the joints are deformed, painful, or swollen, CRP, rheumatoid factor, antinuclear factor, and HLA B27 must be tested. Further help from a pediatrician and rheumatologist should be obtained if juvenile idiopathic arthritis (JIA) or other inflammatory disorder is suspected.

19.5 Differential Diagnosis of Conditions That Cause Multiple Deformities in Children

19.5.1 Multiple Congenital Contractures

A very large number of conditions manifest as multiple congenital contractures, the most well-known of which is classical arthrogryposis multiplex congenita or amyoplasia. Other conditions that present with multiple contractures in the newborn include distal arthrogryposis and congenital contractural arachnodactyly.

Classical Arthrogryposis

The patterns of deformities vary with the limbs being predominantly either in extension or flexion (Fig. 19.4). The upper and lower limbs are involved, and usually the deformities are symmetric.

Distal Arthrogryposis

This autosomal dominant, X-linked condition manifests as multiple joint contractures. Though the proximal joints may be affected to some degree, the predominant deformities involve the wrists, hands, and feet.

Congenital Contractural Arachnodactyly

Symmetric joint contractures of the large and small joints are present at birth. The knees and elbows are often flexed. Camptodactyly and arachnodactyly are typically seen; the external ear has a crumpled "cabbage leaf" appearance. Though there is some resemblance to Marfan syndrome, the distinguishing features are the conspicuous absence of joint laxity and ocular defects that are present in Marfan syndrome. The joint deformities tend to resolve with time (Kolble et al. 2002; Viljoen 1994).

19.5.2 Bone Disease

Osteogenesis Imperfecta

The deformities in osteogenesis imperfecta are on account of malunion of either complete fractures or micro-fractures; malunion of complete fractures results in angular deformities, while micro-fractures lead to bowing deformities. Typically, the deformities are in the diaphysis of the long bones (Fig. 19.5); frequently the femur tends to bow anteriorly and laterally, while the tibia tends to bow anteriorly.

Bruck Syndrome

In this genetic condition, there are features of arthrogryposis and osteogenesis imperfecta (Fig. 19.6) (Leroy et al. 1998). The condition is characterized by congenital contractures including popliteal or cubital pterygia. Fractures first occur in infancy or early childhood. Scoliosis and protrusion acetabulae may develop and progress rapidly (Fig. 19.7).

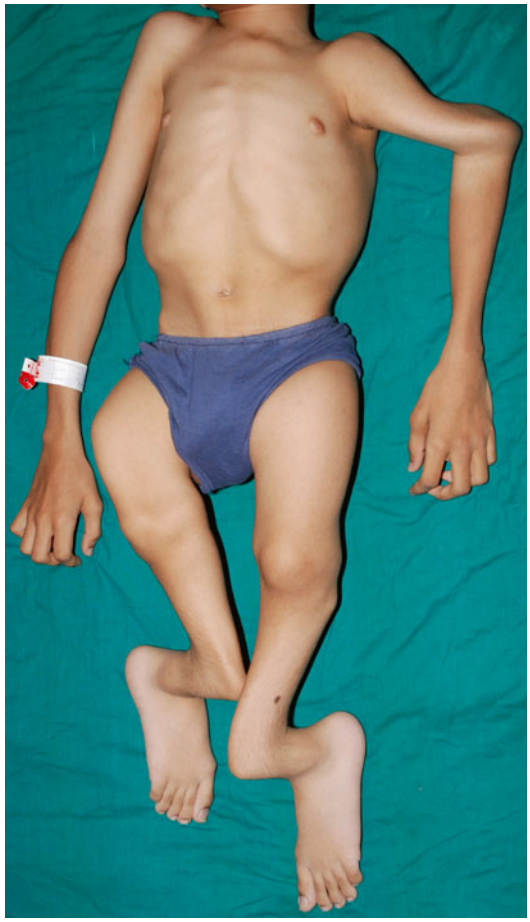

Fig. 19.5 Deformities of the shafts of the long bones of the upper and lower extremity in a child with osteogenesis imperfecta

Polyostotic Fibrous Dysplasia and Albright Syndrome
Multiple lesions develop in the skeleton where normal bone is replaced by fibrous tissue. The bone is prone to bend or break in the region of the lesions.

Multiple Enchondromatosis (Ollier Disease)
Benign cartilaginous lesions affect several bones; the distribution may affect only one side of the body. When both sides of the body are affected, the involvement is usually asymmetric. Palpable bony swellings develop when the lesions expand the cortices; limb-length inequality and deformities occur frequently.

Hereditary Multiple Osteochondromatosis
Bony swellings in the metaphyseal region of the long bones appear in early childhood and enlarge as the child grows. Growth abnormalities are common; they result in angular deformities and shortening of the affected bones.

19.5.3 Arthritis

Juvenile Chronic Arthritis
Flexion deformities of the hips and knees and elbows and wrists are frequently seen in juvenile chronic arthritis. In pauciarticular or oligoarticular JIA, fewer joints are involved (Meiorin et al. 2009). Joint swelling due to effusion and synovial hypertrophy is a feature of active disease.

Juvenile Psoriatic Arthritis
The pattern of joint involvement is different from JIA; the small joints of the hand are more frequently affected (Huemer et al. 2002).

19.5.4 Skeletal Dysplasia

Deformities and growth abnormalities of some forms of skeletal dysplasia are evident at birth, while others become evident as the child grows. A more detailed account of establishing a diagnosis of a skeletal dysplasia is described in Chap. 23. Hence, skeletal dysplasias are not included in this chapter with two exceptions: campomelic dwarfism, a dysplasia where the bones are bowed, and progressive pseudorheumatoid arthropathy in children which closely resembles JIA in its presentation merit consideration.

Campomelic Dysplasia
This dysplasia is recognizable at birth; the long bones are bowed with cutaneous dimples over the apices of the bowed bones. The deformities are symmetric. The scapula is hypoplastic. Cleft palate and respiratory distress in the newborn period are additional features of the condition.

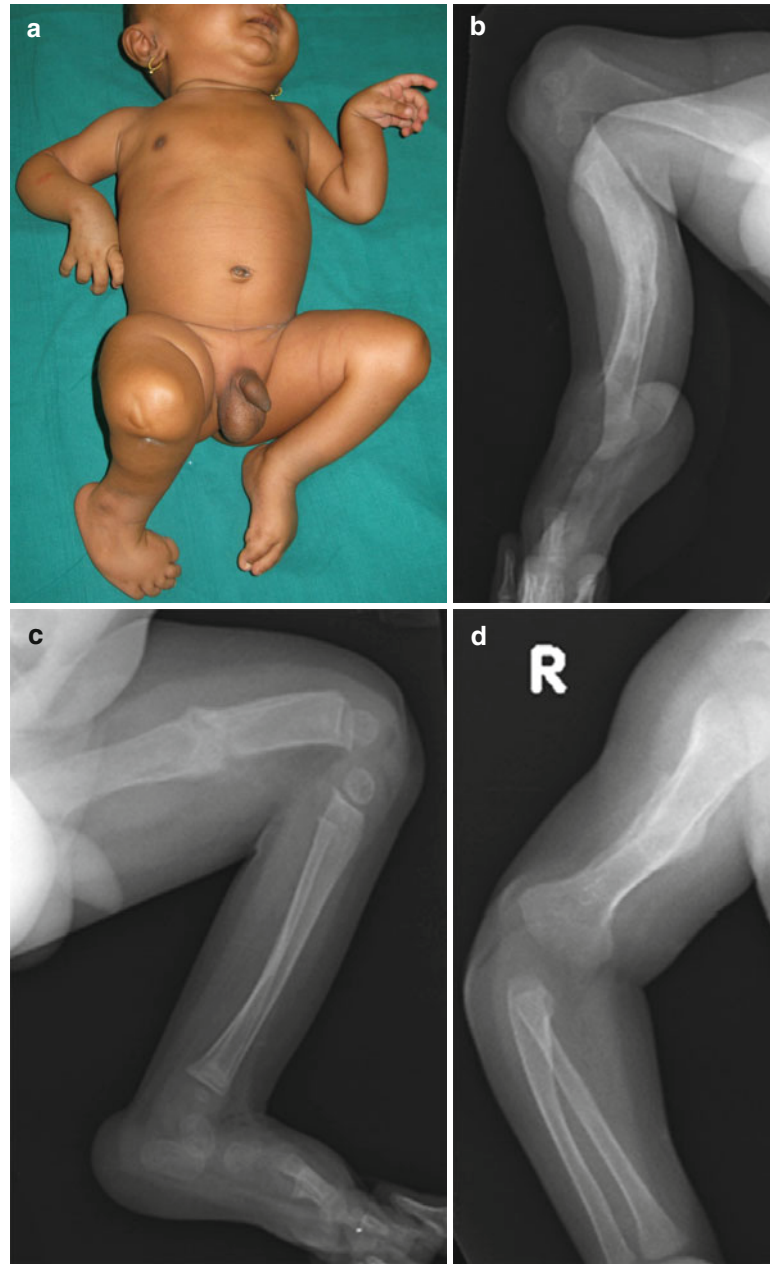

Fig. 19.6 Clinical appearance (**a**) and radiographs (**b**–**d**) of a child with Bruck syndrome; the joint deformities and the fractures of the limbs are characteristic

Progressive Pseudorheumatoid Arthropathy of Childhood

This condition which very closely resembles rheumatoid arthritis in its clinical presentation is known by other names such as spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA) and progressive pseudorheumatoid dysplasia (PPD). The symptoms which are progressive often begin around 3 years of age with joint stiffness affecting the hips. Morning stiffness and stiffness of the neck may also be present. The finger joints may develop swelling; this is not due to synovial hypertrophy but due to enlargement of the ends of the phalanges. The ESR is normal, the rheumatoid factor is negative, and the synovium does not show inflammatory changes of rheumatoid arthritis. Platyspondyly is a characteris-

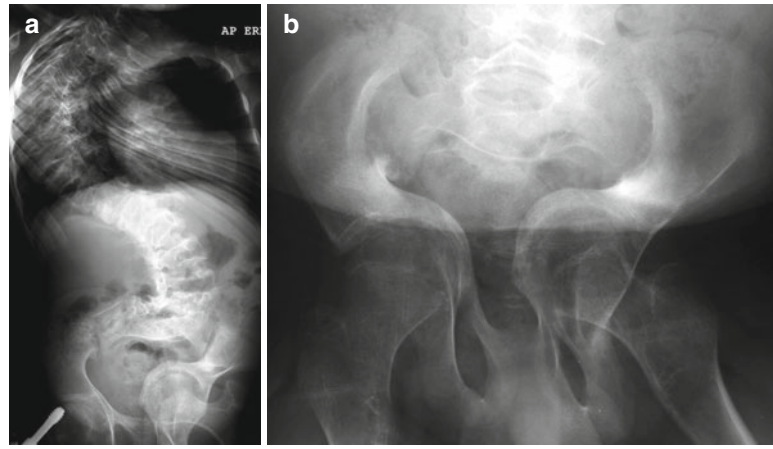

Fig. 19.7 Severe scoliosis (**a**) and protrusion acetabulae (**b**) in siblings with Bruck syndrome

tic feature of this condition. The condition does not respond to antirheumatic drugs; the importance of diagnosis is to avoid these potentially harmful drugs (Ekbote et al. 2013).

19.5.5 Sequelae of Multifocal Septic Arthritis and Osteomyelitis

Damage to the epiphysis and the growth plates will give rise to progressive deformities that may initially manifest a few months after the infective episode. The nature of the deformities will depend on the extent and site of damage of each bone.

19.5.6 Metabolic Bone Disease

Children with vitamin D-resistant rickets or other forms of untreated rickets often develop deformities of the weight-bearing long bones. Often the bones are bowed; recurrence of the deformity after correction is common.

19.5.7 Paralytic Conditions

Poliomyelitis
Multiple deformities may develop in a child following poliomyelitis. The deformities are either postural deformities (flexion, abduction, and external rotation deformities of the hips, flexion deformity of the knee, and equinus deformity of the ankle). If these deformities have been prevented by appropriate positioning during the acute phase of paralysis, any deformity that develops later will be on account of residual muscle imbalance.

Sacral Agenesis
Children with complete sacral agenesis are born with characteristic symmetric deformities of the hips, knees, and ankles. They include abduction and external rotation deformity of the hips, flexion of the knees, and equinus of the ankles. Associated spinal deformities, paralysis of the lower limbs, and neurogenic bladder make the management of these children difficult (Renshaw 1978).

19.5.8 Rare Conditions

Carpal-Tarsal Osteolysis
Idiopathic carpal-tarsal osteolysis is a rare disorder characterized by resorption of the carpals and tarsals with severe deformities of the wrist, hand, and feet; the more proximal joints may also be affected. There may be associated nephropathy (Carmichael et al. 2007; McDonald et al. 2007; Choi et al. 1993).

NOMID
Neonatal-onset multisystem inflammatory disease (NOMID) is a very rare disorder that affects the nervous system, joints, and skin. The joint manifestations include painful joint swelling and cartilage overgrowth that result in characteristic prominent knees with flexion

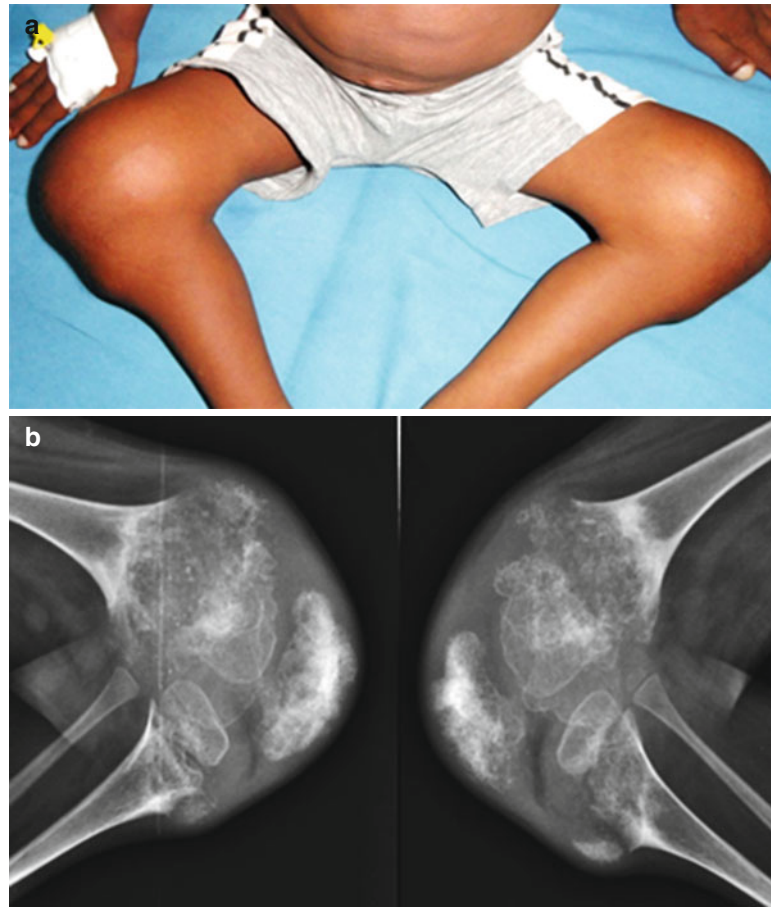

Fig. 19.8 Characteristic swelling and flexion deformities of the knees in a child with NOMID (**a**); the radiograph (**b**) shows the abnormal deposition of calcified cartilage in the region of the growth plate

deformities that progress with time (Fig. 19.8). Other joints may also get affected. The underlying cause is excessive release of interleukin-1β (Boschan et al. 2006; Kim et al. 2014; Hill et al. 2007).

19.6 Establishing the Diagnosis

Outlines establishing the cause of multiple deformities in a child are shown in Tables 19.2 and 19.3.

Table 19.2 Establishing the diagnosis of the cause of multiple bony deformities in a young child

History			
Onset of deformities: Either at birth or in early childhood	Onset of deformities: Antenatal	Onset of deformities: In early childhood	Onset of deformities: During childhood
History of repeated fractures following trivial trauma	History of respiratory problems in neonatal period may be present	Failure to thrive	May present initially as a pathologic fracture
A positive family history may be present		A positive family history may be present	
Natural history: Deformities may get worse if untreated, and fresh deformities may develop following fresh fractures	*Natural history*: Deformities remain static	*Natural history*: Deformities progress and have a propensity to recur after surgical correction	*Natural history*: Deformities may progress
Physical examination			
Blue sclera may be present	–	–	Café-au-lait spots may be present
Ligament laxity may be present	–	–	Precocious onset of puberty may be present
Diaphyseal bowing of the long bones of both the upper and lower limbs Femur: Anterior and lateral bow frequently at junction of proximal and middle third Tibia: Anterior bowing in mid-diaphysis common	Diaphyseal bowing of the long bones of both the upper and lower limbs Femur: Anterior and lateral bow frequently at junction of proximal and middle third Tibia: Anterolateral bowing in mid-diaphysis common Skin dimples often present over the apex of the bowed bones	Diaphyseal bowing of long bones predominantly of the lower limbs Femur: Coxa vara Lateral bowing at the junction of the middle and lower third of the Tibia: Anterior bowing in mid-diaphysis or at junction of the middle and lower third	Metaphyseal and/or diaphyseal bowing Femur: Bowing of the femoral neck with severe coxa vara (Shepherd crook deformity) frequently seen Tibia: Anterior bowing of mid-diaphysis
Working diagnosis: Osteogenesis imperfecta	Working diagnosis: Campomelic dysplasia	Working diagnosis: Vitamin D-resistant rickets	Working diagnosis: Fibrous dysplasia
Investigations			
Plain radiographs will confirm the presence of healing fractures and deformities Osteopenia Normal growth plates	Plain radiographs will confirm the presence of mid-diaphyseal bowing of the femur and tibia No osteopenia Normal growth plates	Plain radiographs will confirm the presence of the deformities Osteopenia Growth plates widened and irregular	Plain radiographs will show osteolytic lesions in the diaphysis and metaphysis of several bones. Bowing (frequently of the femoral neck) and thinning of the cortices of the long bones may be seen
		Serum calcium, phosphate, parathormone, and vitamin D levels will be deranged. Evidence of phosphaturia, glycosuria, and aminoaciduria may be present	
Diagnosis			
Osteogenesis imperfecta	Campomelic dysplasia	Vitamin D-resistant rickets	Fibrous dysplasia

Table 19.3 An outline of the process of establishing the diagnosis of multiple joint deformities in a young child

History						
Antenatal: History of akinesia in pregnancy	Antenatal: Nothing significant	Antenatal: Nothing significant	Antenatal: Nothing significant	Antenatal: Nothing significant	Antenatal: Nothing significant	Antenatal: Nothing significant
Onset of deformities: Present at birth	Onset of deformities: Present at birth	Onset of deformities: In early childhood associated with pain	Onset of deformities: In early childhood associated with pain	Onset of deformities: Months after septic episode	Onset of deformities: After febrile episode with acute flaccid paralysis	Onset of deformities: Present at birth
Natural history: Deformities remain static	*Natural history*: Resolution of deformities	*Natural history*: Progression of deformities	*Natural history*: Progression of deformities	*Natural history*: Progression of deformities	*Natural history*: Deformities remain static	*Natural history*: Deformities remain static
Physical examination						
Pattern of deformities: Symmetric	Pattern of deformities: Symmetric	Pattern of deformities: Symmetric (may be asymmetric in pauciarticular JIA)	Pattern of deformities: Symmetric	Pattern of deformities: Asymmetric	Pattern of deformities: Asymmetric	Pattern of deformities: Symmetric
No pain	No pain	Pain present	Pain present	No pain	No pain	No pain
No swelling of joints	No swelling of joints	Swelling of joints	Swelling of joints	No swelling of joints	No swelling of joints	No swelling of joints
No signs of inflammation	No signs of inflammation	Signs of inflammation	No signs of inflammation	No signs of inflammation	No signs of inflammation	No signs of inflammation
Normal muscle power	Normal muscle power	Normal muscle power	Normal muscle power	Normal muscle power	Lower motor neuron paralysis of the muscles of the deformed limbs	Lower motor neuron paralysis of the muscles of the deformed limbs
Other features: Stature: normal Spine: scoliosis may develop	Other features: Stature: normal Spine: scoliosis may develop Hand: arachnodactyly, camptodactyly	Other features: Stature: normal Spine: may be affected in disease	Other features: Stature: short Spine: platyspondyly	Other features: Stature: normal Spine: normal	Other features: Stature: normal Spine: paralytic scoliosis if spinal muscles are paralyzed	Other features: Stature: normal or short Spine: deformities may be present, sacrum absent

(continued)

Table 19.3 (continued)

Limb lengths equal	Limb lengths equal	Limb lengths equal	Limb lengths equal	Progressive shortening of affected limbs	Shortening of affected limbs may be present	Limb lengths equal
Investigations						
No investigations indicated needed to establish diagnosis	No investigations indicated to establish diagnosis Genetic analysis may be done for confirmation	ESR, rheumatoid factor, antinuclear antibody, synovial biopsy Plain radiograph of the affected joints will show periarticular osteoporosis and articular erosions	ESR, rheumatoid factor, and antinuclear antibody will be *negative* Plain radiograph of the spine: platyspondyly Plain radiograph of the affected joints may show reduction in joint space	Plain radiographs of the affected joints may show destruction of the epiphysis and physis, physeal bar formation, and deformity	No investigation required to establish the diagnosis	Plain radiograph of the pelvis will confirm the absence of the sacrum
Diagnosis						
Classic arthrogryposis/distal arthrogryposis	Congenital contractural arachnodactyly	Juvenile idiopathic arthritis	Progressive pseudorheumatoid arthropathy of childhood	Sequelae of septic arthritis or osteomyelitis with growth plate damage	Paralytic deformities	Sacral agenesis

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Acquired Deformities of the Knee

20

Benjamin Joseph

20.1 Establishing the Diagnosis of the Cause of Genu Varum or Valgum

20.1.1 Introduction

Acquired deformities of the knee are extremely common in young children; the vast majority are innocuous and will resolve spontaneously, but some require early intervention (Greene 1994). It is vital that the clinician differentiates those that can be ignored from those that require treatment. Coronal plane deformities of the knee (genu varum and valgum) are more frequently encountered than sagittal plane deformities (flexion or extension).

20.1.2 Questions to Establish a Diagnosis

- **When was the deformity first noted?**
- **Is the deformity unilateral or bilateral and symmetric?**
- **Is the deformity progressing, resolving, or remaining static?**
- **Is the deformity associated with shortening?**
- **Is it a uniplanar deformity, or are there deformities in more than one plane?**
- **Is the deformity in the femur or tibia?**
- **Is the deformity at the joint, epiphysis, physis, or metaphysis?**
- **Does the deformity change on standing or on flexing the knee?**

When was the deformity first noted?

Physiologic genu varum is often noted when the child starts to walk and may persist till the age of two. Thereafter the alignment of the knee tilts into valgus which gradually reduces after the age of 4 years, and by the age of 6 or 7 years, the normal adult alignment is achieved (Heath and Staheli 1993). It follows that genu varum that develops after the age of two or three is likely to be pathologic. Similarly genu valgum in a 1 year old is likely to be pathologic (Fulp et al. 1995).

Is the deformity unilateral or bilateral and symmetric?

Some bilateral symmetric deformities are physiologic (e.g., genu varum in an 18-month-old boy), while some are clearly pathologic (e.g., genu valgum in a child with Ellis-van Creveld syndrome, Fig. 20.1). Unilateral deformities are always pathologic (Fig. 20.2).

Is the deformity progressing, resolving, or remaining static?

Physiologic genu varum tends to gradually resolve from the point when it is first noted, while physiologic genu valgum initially progresses before it begins to resolve at around 4 years of age.

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_20

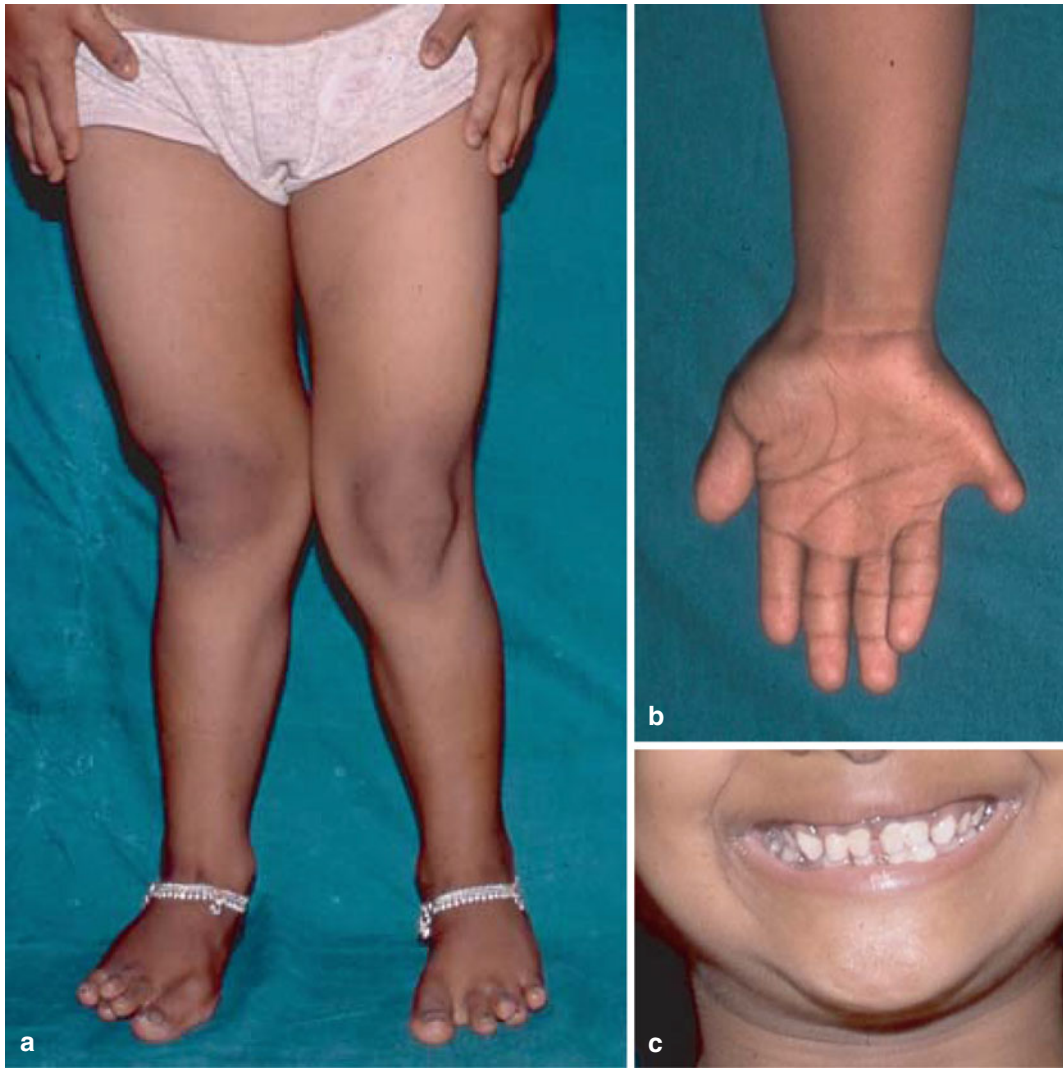

Fig. 20.1 Bilateral genu valgum in a young child with Ellis-van Creveld syndrome (**a**); the postaxial polydactyly (**b**) and abnormal dentition (**c**) are features of this form of skeletal dysplasia

Progression of pathologic deformity suggests that the growth plate may be affected with asymmetric growth inhibition or growth stimulation. This differentiation has a bearing on treatment planning. Mere correction of a progressive deformity due to a growth plate injury (without some form of physeal surgery) is not useful in the long run as the deformity will recur. On the other hand, correction of a static deformity is likely to be of long-standing benefit.

Is the deformity associated with shortening?

Deformity associated with shortening is likely to have developed due to damage to the growth plate especially if the deformity and shortening are progressive. Deformity associated with shortening may also be seen in children with long-standing lower motor neuron paralysis.

Is it a uniplanar deformity, or are there deformities in more than one plane?

It is important to identify deformities that are biplanar or multiplanar to facilitate appropriate treatment planning. A biplanar deformity that is common is genu varum associated with internal tibial torsion (Fig. 20.3).

Is the deformity in the femur or tibia?

It is essential that the surgeon identifies the bone that is deformed to plan appropriate treatment.

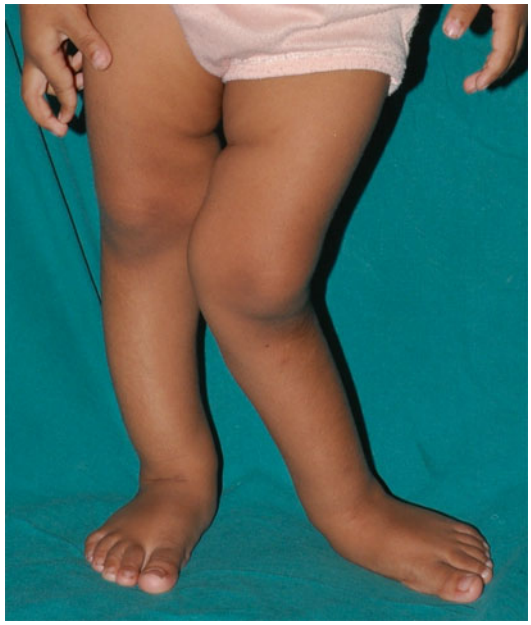

Fig. 20.2 Unilateral pathologic genu valgum in a young child; this was secondary to neonatal infection in the distal femur

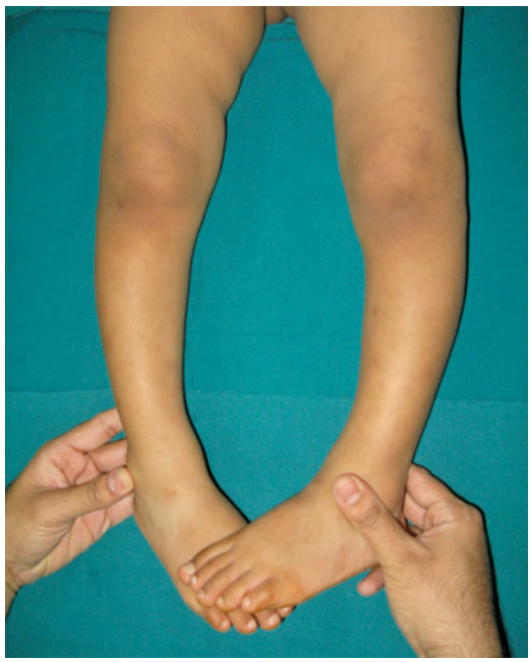

Fig. 20.3 Genu varum is often associated with internal tibial torsion

Genu varum and valgum can develop due to pathologic changes in the femur, tibia, or both, and on a cursory examination all may look identical. However, careful clinical examination can differentiate these three situations.

Is the deformity at the joint, epiphysis, physis, or metaphysis?

It is also essential that the exact site of the pathology in the bone is identified to facilitate proper treatment planning.

Does the deformity change on standing or on flexing the knee?

A varus or valgus deformity can get accentuated on standing if the collateral ligaments are lax. If genu valgum or varum deformity is due to a deformity in the distal femur, the deformity will get obliterated on flexing the knee, while the deformity will not get masked on flexing the knee if the deformity is in the proximal tibia (Fig. 20.4).

20.1.3 Physical Examination

Careful clinical examination is most important for establishing the diagnosis.

Look

Look at the knees of the child as the child is standing and when the child is recumbent with the knees extended and then with the knees flexed. Note if the deformity increases on standing and note if the deformity gets masked on flexing the knee. Observe if there is an associated torsional deformity. Note if there is a limb-length discrepancy. Look for features of rickets such as metaphyseal widening or rachitic rosary. Note if the stature, body proportions, and limb lengths are normal.

Feel

Palpate the distal femur and the proximal tibia and note if there is any bony irregularity of the metaphysis or epiphysis. Note if the metaphyses of the femur and tibia are widened or thickened. Palpate the patella and note if it is situated in the normal position or displaced laterally. Palpate and determine if there is partial or total fibular aplasia. Palpate the iliotibial band and note if it is taut.

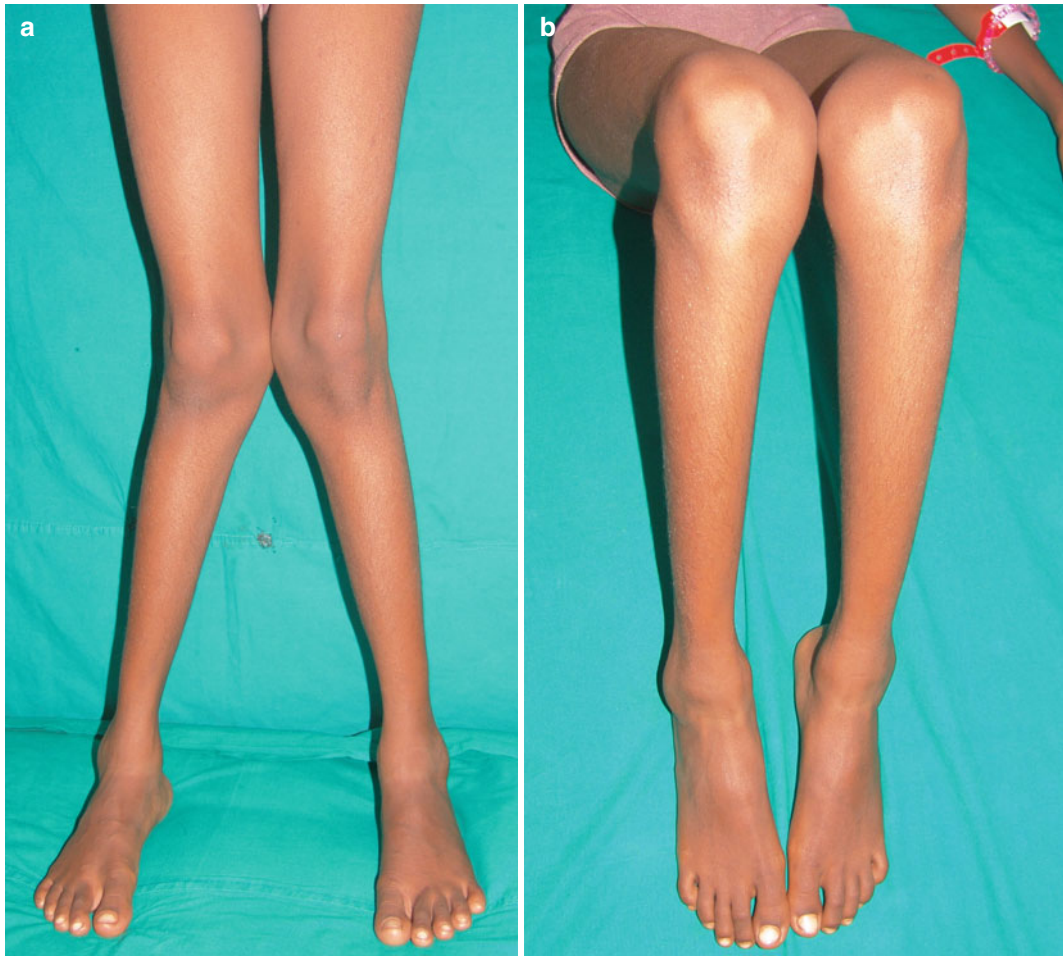

Fig. 20.4 Genu valgum that is clearly evident with the knees in extension (**a**) is masked when the knees are flexed (**b**)

Move

Check if the range of motion of the knee is normal.

Measure

If there is bilateral symmetric genu varum, measure the intercondylar distance when the child is recumbent with both limbs in neutral rotation (patellae facing upward) and again when the child is standing with both patellae facing forward.

If there is bilateral symmetric genu valgum, measure the intermalleolar distance when the child is recumbent with both limbs in neutral rotation (Fig. 20.5) and again when the child is standing; if the distance increases on standing, there may be laxity of the medial collateral ligaments of the knee.

Special Tests

Test for Collateral Ligament Laxity

Test for collateral ligament laxity by applying valgus and varus stress on the knee flexed to 20–30°.

The Cover-Up Test

This test has been found to be very useful to differentiate physiologic genu varum from infantile Blount's disease in children between 1 and 3 years of age (Davids et al. 2000). The lower two-third of the leg is covered leaving

Fig. 20.5 Measurement of the intermalleolar distance must be done with the patella facing forward

only the proximal third of the leg exposed. The alignment of the proximal third of the leg in relation to the thigh is noted (Fig. 20.6). In Blount's disease the proximal third of the leg is in neutral or varus alignment, while in physiologic genu varum the proximal third of the leg is valgus. Davids et al. noted that the test was 100 % sensitive with all children with Blount's disease having a positive cover-up test; the specificity was 0.86, the positive predictive value was 0.72, and the negative predictive value was 1.00.

Tests to Diagnose Coxa Vara

Exaggerated hip adduction with reduced hip abduction and a positive Trendelenburg test are suggestive of associated coxa vara.

Test for Iliotibial Band Tightness: The Ober Test

The patient lies on his/her unaffected side with the lower hip and knee flexed to 90°. The examiner stabilizes the pelvis with a hand on the patient's iliac crest throughout the test. The affected thigh is then abducted with the hip and knee flexed. The hip is then extended, and with the hip extended the thigh is slowly lowered toward the couch (the hip is adducted passively). The test is positive if the thigh cannot be brought parallel to the couch but remains abducted.

20.1.4 Investigations to Confirm the Diagnosis

Plain Radiographs

Anteroposterior and lateral views of the knee are obligatory, the only exception being symmetric bilateral genu varum in a child under 3 years of age with a negative cover-up test where a radiograph is not indicated. Apart from noting the alignment of the femur and tibia on these radiographs, abnormalities of the growth plates and the bones must be excluded.

An anteroposterior radiograph of the pelvis with both hips is required if coxa vara is suspected. If rickets is suspected, a radiograph of the wrist is useful to demonstrate growth plate changes at the distal radius and ulna. A skeletal survey is necessary if a skeletal dysplasia is suspected; this entails the radiographs mentioned in Table 20.1 at the very least.

Full-Length Standing Radiograph of the Lower Limbs

It is important to ensure that the child stands erect with both patellae facing forward with blocks under the foot of the shorter limb to compensate for shortening if there is limb-length inequality. On the radiograph the mechanical axis is drawn from the center of the femoral head to the center of the distal tibial articular margin (Fig. 20.7). The point at which the axis crosses the knee joint is noted and expressed as a percentage of the

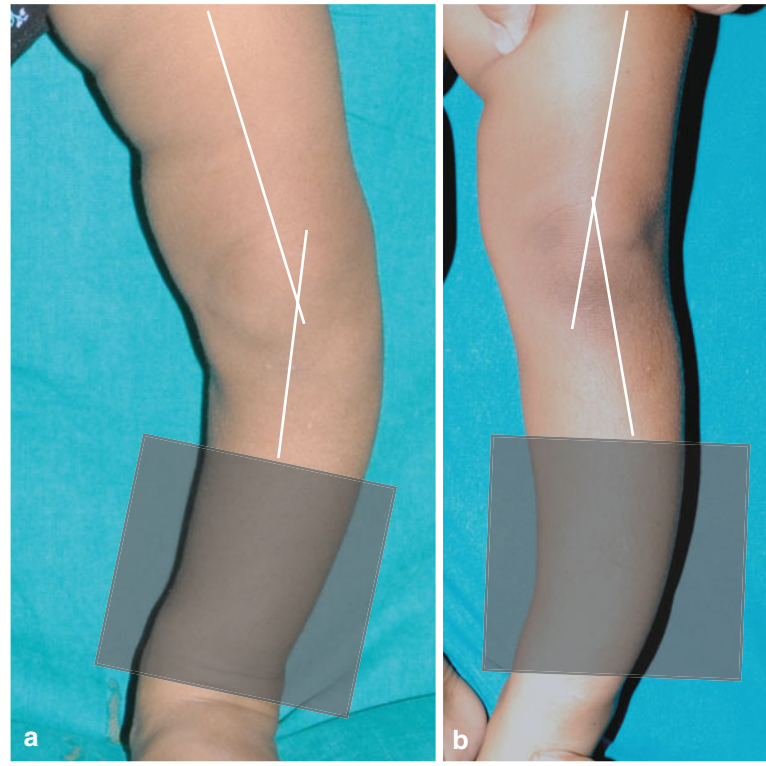

Fig. 20.6 The cover-up test: the middle third of the legs has been covered, and the alignment of the femur and the proximal third of the leg is assessed. Pathologic genu varum (due to Blount's disease) is noted in one limb, (**a**) while the bow leg is physiologic in the other limb (**b**)

Table 20.1 The list of radiographs to be included in a skeletal survey for establishing the diagnosis of skeletal dysplasia

Region	View	Comment
Skull	AP and lateral views	
Thoraco-lumbar spine	AP and lateral views	Very common site of characteristic abnormality
Chest	PA view	
Pelvis	AP view	Common site of characteristic abnormality
One upper limb	AP view	Enables to establish if the dysplasia is rhizomelic, mesomelic, or acromelic Also enables to establish if the involvement is epiphyseal, metaphyseal, or diaphyseal
One lower limb	AP view	
Left hand	AP	For assessing bone age

width of the tibial articular surface from the lateral limit of the articular margin. If there is deviation of the mechanical axis, the site of the deformity can be determined by measuring the lateral distal femoral angle and the medial proximal tibial angle.

Magnetic Resonance Imaging

MR imaging is useful if growth plate damage is suspected; the site and extent of damage can be mapped reliably.

Laboratory Investigation

Serum levels of calcium, phosphate, alkaline phosphatase, and vitamin D and renal function tests are indicated if there are knee deformities associated with metaphyseal widening and bowing of the tibia and femur or if the radiographs show widening of the growth plates.

20.1.5 Causes of Genu Varum or Valgum

Physiologic Genu Varum and Valgum

Physiologic genu varum is seen in healthy children under the age of 3 years. The deformity is usually noted soon after the child begins to walk.

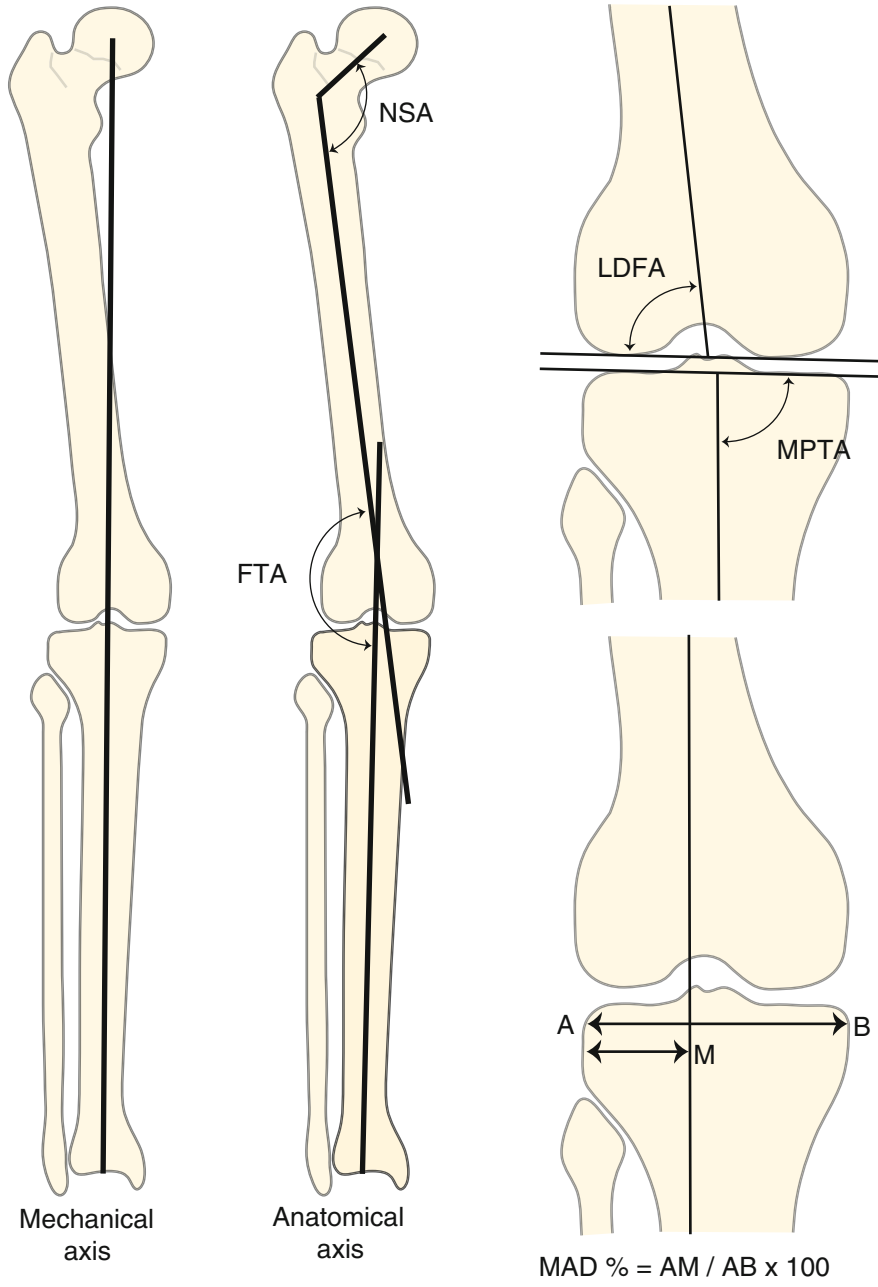

Fig. 20.7 The measurements that are made on full-length standing radiographs of the lower limbs taken with the patellae facing forward

The deformity is symmetric and may be associated with some internal tibial torsion. The cover-up test is typically negative. Physiologic genu valgum is seen in children between the ages of 2 and 7 years. The deformity is symmetric, and the site of the deformity is the distal femur.

Infantile Blount's Disease

Early Blount's disease may be clinically and radiologically indistinguishable from physiologic genu varum (Fig. 20.8a). Though the disease is frequently bilateral and symmetric, unilateral involvement may occur. The child may be

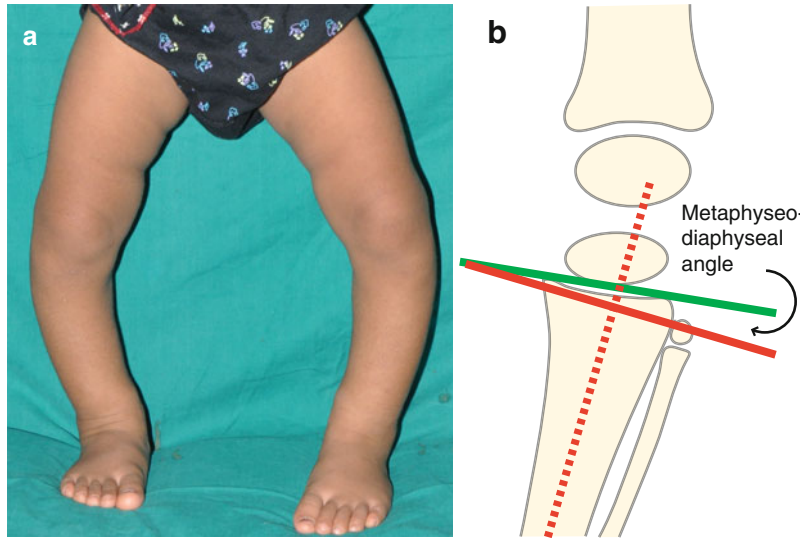

Fig. 20.8 Early infantile Blount's disease may be indistinguishable clinically from physiologic genu varum (**a**); a metaphyseo-diaphyseal angle greater than 11° is suggestive of Blount's disease (**b**)

overweight, and there may be a history of early walking; however, both these features are not present in several instances. The cover-up test becomes positive as the disease progresses, and radiographic changes of metaphyseal beaking and growth plate irregularity on the medial margin develop. The deformity can be demonstrated on the radiograph by measuring the metaphyseal-diaphyseal angle (Levine and Drennan 1982). This is a reproducible measurement (Lavelle et al. 2008), and the angle is typically greater than 11° in Blount's disease (Fig. 20.8b). However, a diagnosis must not be based on this measurement alone (Feldman and Schoenecker 1993).

Trauma

One of three mechanisms may contribute to a deformity at the knee following an injury in a young child. Firstly, an epiphyseal injury with growth plate damage can result in a progressive angular deformity. Secondly, malunion of a fracture of the femur or tibia may lead to genu valgum or varum; the deformity may decrease to some degree by remodeling but tends to remain static thereafter. Thirdly, following an undisplaced metaphyseal fracture of the tibia, a valgus deformity may develop; the deformity usually resolves over a period of time (Cozen 1953). The precise cause of this phenomenon first noted by Cozen is unclear.

Metabolic Causes

Genu valgum or varum may develop in nutritional and various forms of vitamin-D-responsive and vitamin-D-resistant rickets (Schnitzler et al. 1994; Petje et al. 2008; Bar-On et al. 2008; Bhagat et al. 2007). Widening or "flaring" of the metaphysis of the distal radius can often be clinically appreciated by carefully palpating the distal third of the radius and running the finger distally till the wrist joint. Rachitic rosary may be noted by palpating the costochondral junctions. The tibia and the femur are often bowed at the junction of the middle and lower thirds, and this aggravates the appearance of the bowing at the knee (Cheema et al. 2003). Radiographs may occasionally be normal (Feldman and Schoenecker 1993) but more characteristically will show cupping and flaring of the metaphysis which is best seen in the distal radius and ulna. Genu varum and valgum may also be seen in fluorosis (Krishnamachari and Krishnaswamy 1973; Khandare et al. 2005).

Infection: Sequelae of Septic Arthritis or Osteomyelitis

Septic arthritis in infancy can destroy a condyle of the tibia or femur (Fig. 20.9) with severe varus or valgus deformity (Tercier et al. 2012). Varus or valgus deformity may result from a metaphyseal focus of infection on the adjacent growth plate; mild infection may cause hyperemia and

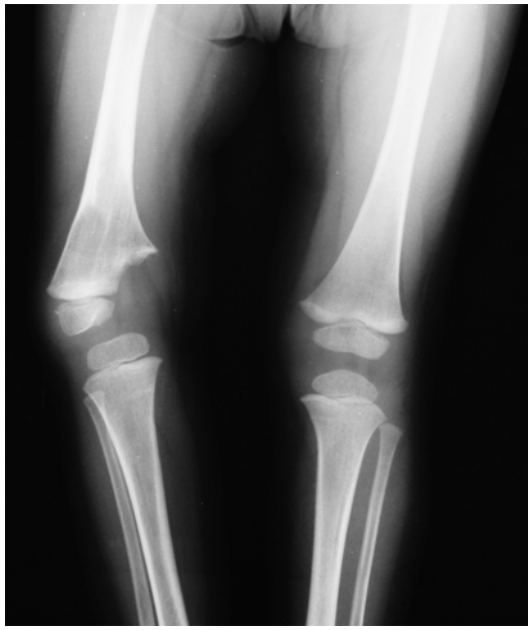

Fig. 20.9 Genu varum secondary to loss of a condyle of the femur following septic arthritis in infancy

asymmetric stimulation of growth, while more virulent infection may damage the growth plate. Angular deformities due to growth plate damage are usually associated with variable degrees of shortening of the limb.

Focal Fibrocartilaginous Dysplasia

Focal fibrocartilaginous dysplasia is a localized cortical defect most frequently noted in the medial aspect of the proximal tibial metaphysis (Fig. 20.10). The defect is surrounded by an area of sclerosis, and there is angulation at the site of the defect (Albinana et al. 1997). Biopsies of tissue curetted from the defect have shown fibrocartilaginous tissue. Less frequently the lesion may be localized in the medial aspect of the distal femur (Albinana et al. 1997). Very infrequently the lesion is located on the lateral aspect with resultant genu valgum (Ruchelsman et al. 2004). Biopsy of the lesion is not recommended as the radiographic appearance is so typical and spontaneous resolution occurs frequently. Curettage of the lesion may be curative if spontaneous resolution does not occur; corrective osteotomy is seldom required.

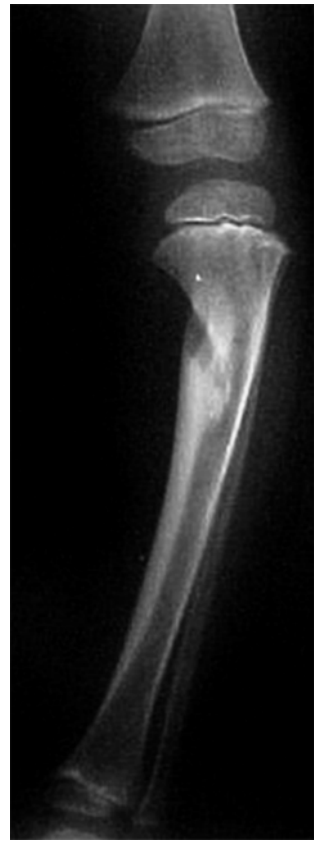

Fig. 20.10 Varus deformity of the proximal tibia due to a focal fibrocartilaginous defect

Fibular Hemimelia

The lateral femoral condyle is often hypoplastic in children with fibular hemimelia, and consequently they develop genu valgum.

Skeletal Dysplasia

Genu varum or genu valgum is very frequently seen in various different forms of skeletal dysplasia (Greene 1994; Lee et al. 2007; Ain et al. 2006; Amirfeyz et al. 2006). In achondroplasia the characteristic deformity at the knee is genu varum (Fig. 20.11a) (Ain et al. 2006; Lee et al. 2007). One reason attributed for the varus deformity is the relative overgrowth of the fibula noted in children with achondroplasia (Lee et al. 2007) though this view is not universally accepted (Ain et al. 2006). In Ellis-van Creveld syndrome (chondroectodermal dysplasia), the deformity at the knee is genu valgum (Fig. 20.1). The lateral tibial

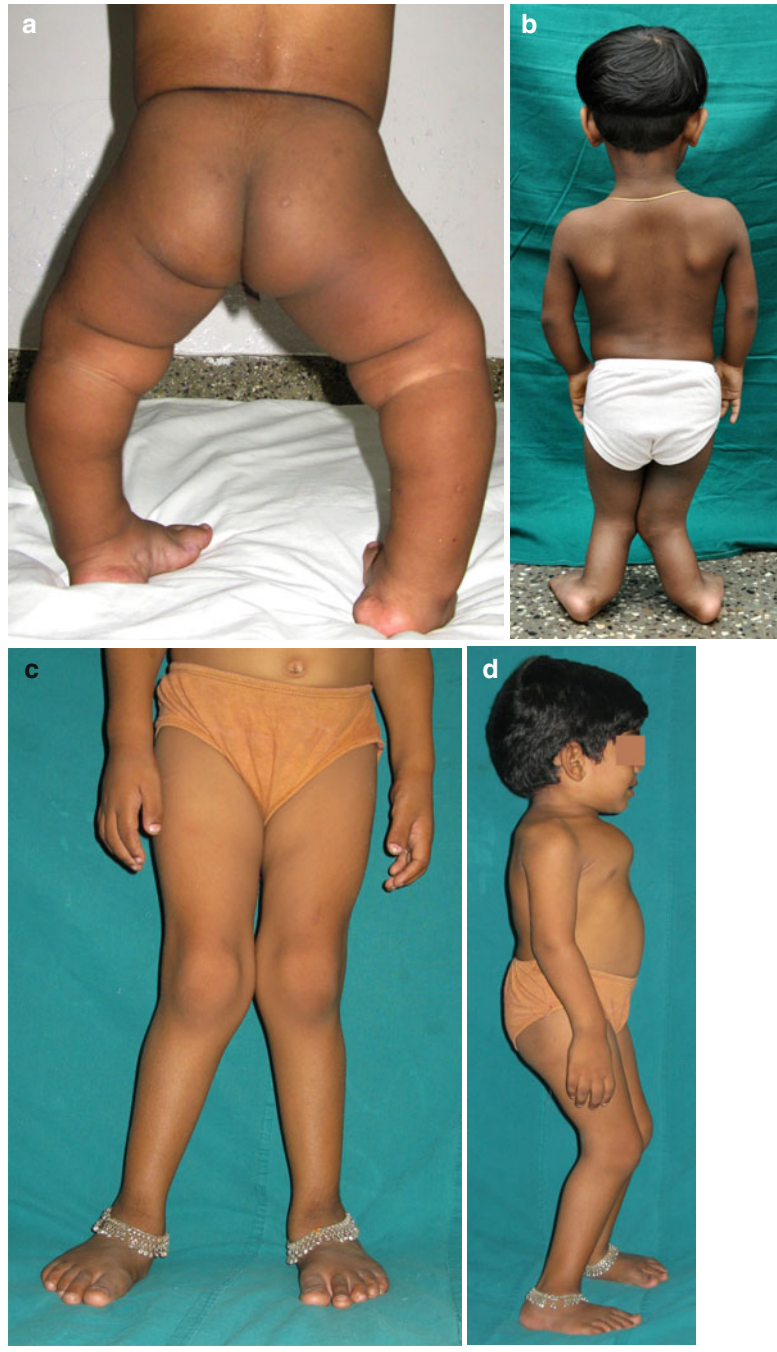

Fig. 20.11 Genu varum in achondroplasia (**a**), genu valgum in mesomelic dysplasia (**b**), and genu valgum in Morquio disease (**c**, **d**)

condyle is hypoplastic, and growth of the lateral part of the physis is inhibited in this condition. Genu valgum is a feature of Morquio-Brailsford syndrome, a form of mucopolysaccharidosis (MPS IV), and is also seen in forms of mesomelic dysplasia (Fig. 20.11b–d).

Dysplasia Epiphysealis Hemimelica, Enchondromatosis, and Osteochondromatosis Syndromes

Children with abnormalities of the cartilage and bone growth such as dysplasia epiphysealis hemimelica (Perl et al. 2009), enchondromatosis, and osteochondromatosis may develop angular deformities at the knee. Genu valgum may be seen in Down syndrome and some other rare syndromes (Herrera-Soto and Price 2008).

Soft Tissue Contractures

Contracture of the iliotibial band and the lateral intermuscular septum is commonly seen in polio, and this leads to a valgus deformity at the knee. Lower motor neuron paralysis with wasting will be evident, and the Ober test for iliotibial band contracture will be positive. Quadriceps contracture especially involving the vastus lateralis may also be associated with iliotibial band contracture and genu valgum (Bose and Chong 1976).

"Compensatory" Deformity

Shim et al. (1997) reported instances of genu valgum in children with coxa vara and hypothesized that the genu valgum is the result of altered weight-bearing stresses brought about by the coxa vara. A long-term follow-up study of children who had undergone a proximal femoral varus osteotomy for Legg-Calvé-Perthes' disease, however, did not demonstrate a propensity for clinically appreciable genu varum (Tercier et al. 2013).

20.1.6 Establishing the Diagnosis

An outline of the process of establishing a diagnosis of the cause of bilateral symmetric genu valgum or genu varum is shown in Table 20.2. Table 20.3 shows an outline of establishing the diagnosis of unilateral genu varum or valgum.

Table 20.2 An outline of the process of establishing the diagnosis of bilateral symmetric genu varum or valgum in the young child

History				
Child under 3 years of age	Child between 3 and 7 years of age	Child over 3 years of age	Any age	Any age
Deformity noted at around 1 year of age	Deformity noted around 3 years of age	Deformity noted at 1 year or later	Onset variable	Onset at birth in some conditions; variable age of onset in others
Some spontaneous resolution of deformity	Some progression between 3 and 5 years and spontaneous resolution between 5 and 7 years of age	Progression of deformity noted	No spontaneous resolution	No spontaneous resolution
			Positive family history of similar deformities may be present	Positive family history of similar deformities may be present
Physical examination				
Bilateral symmetric deformity in well-nourished child	Bilateral symmetric deformity in well-nourished child	Bilateral symmetric deformity	Bilateral symmetric deformity	Bilateral symmetric deformity

(continued)

Table 20.2 (continued)

Stature for age normal	Stature for age normal	Stature for age normal Obesity may be present	Stunting of growth and short stature may be present Body proportions maintained	Stunting of growth and short stature may be present Dwarfism may be proportional or disproportional (body proportions altered with predominant rhizomelic, mesomelic, or acromelic shortening of the limbs)
Cover-up test negative	*Not applicable*	Cover-up test positive	*Not applicable*	*Not applicable*
Deformity in the tibia (does not get masked on flexion of the knee)	Deformity in the femur (deformity gets masked on flexion of the knee)	Deformity in the tibia (does not get masked on flexion of the knee)	Deformity may be in the femur or the tibia	Deformity may be in the femur or the tibia
Associated with internal tibial torsion	No torsional abnormality	Associated with internal tibial torsion	Internal tibial torsion may or may not be present in association with genu varum	Internal tibial torsion may or may not be present in association with genu varum
No metaphyseal broadening of distal radius and no prominence of costochondral junctions	No metaphyseal broadening of distal radius and no prominence of costochondral junctions	No metaphyseal broadening of distal radius and no prominence of costochondral junctions	Metaphyseal broadening and prominence of the costochondral junctions present	Metaphyseal broadening may be present (in metaphyseal dysplasia) but often not present
Investigations				
No investigation warranted	No investigation warranted	Full-length standing radiograph of both lower limbs with patella facing forward Mechanical axis deviation is present Metaphyseal-diaphyseal angle of the tibia usually greater than 11°	Full-length standing radiograph of both lower limbs with patella facing forward Anteroposterior radiograph of the wrist	Skeletal survey
			Serum calcium, phosphorus, alkaline phosphatase, renal function tests, vitamin D and parathormone levels	Genetic analysis
Diagnosis				
Physiologic genu varum	Physiologic genu valgum	Infantile Blount's disease	Genu varum or valgum associated with metabolic bone disease	Genu valgum or varum associated with skeletal dysplasia

Table 20.3 An outline of the process of establishing the diagnosis of unilateral genu varum or valgum in the young child

Epiphyseal or growth plate abnormality				
History				
History of infection in the neonatal period	History of infection/trauma/radiation for tumor in the limb	No history of infection, trauma, or radiation	No history of infection, trauma, or radiation	No history of infection, trauma, or radiation
Onset of deformity in infancy	Onset of deformity after an interval following the infection/trauma/radiation	Deformity noted after child started walking	Shortening of the limb, loss of lateral rays of the foot, and foot deformity present from birth	Deformity often noted in older child
Rapid progression of deformity	Progression of deformity	Gradual progression of deformity	Gradual progression of deformity	Progression of deformity
Tendency for early recurrence following correction of deformity	Tendency for recurrence following correction of deformity	Tendency for recurrence following correction of deformity	No tendency for recurrence following correction	Tendency for recurrence present
Physical examination				
Genu varum or valgum	Genu varum or valgum	Genu varum	Genu valgum	Genu varum or valgum
Mediolateral instability may be present	Instability not present	Instability may develop when deformity becomes severe	No instability	No instability
Shortening present	Shortening present	Negligible shortening	Variable degree of shortening	No shortening
		Cover-up test positive		Palpable eccentrically situated swelling on medial or lateral side of the femoral or tibial epiphysis
Investigations				
Plain radiograph: Loss of a condyle, adjacent growth plate, and metaphysis	Plain radiograph: May show growth plate irregularity or a physeal bar	Plain radiograph: Metaphyseal beaking of proximal tibia Metaphyseal-diaphyseal angle greater than 11°	Plain radiograph: Hypoplasia of the lateral femoral condyle Fibular hypoplasia or aplasia	Plain radiograph: Bony outgrowth from the epiphysis

(continued)

Table 20.3 (continued)

Arthrogram: Will demonstrate if some unossified remnant of the epiphysis is present	Arthrogram not indicated	Arthrogram not routinely indicated in young child	Arthrogram not indicated	Arthrogram not indicated
MRI: Will demonstrate if some unossified remnant of the epiphysis or viable growth plate is present	MRI: Will demonstrate site and extent of physeal damage	MRI: Will demonstrate physeal changes	MRI not indicated	MRI : Will demonstrate the osteocartilaginous nature of the epiphyseal growth
Diagnosis				
Genu varum or valgum secondary to loss of a condyle of the femur or tibia following neonatal sepsis	Physeal damage secondary to infection /trauma/radiation	Unilateral Blount's disease	Genu valgum due to hypoplasia of the lateral femoral condyle in fibular deficiency	Dysplasia epiphysealis hemimelica (Trevor's disease)
Metaphyseal abnormality				
History				
No history of trauma/infection	No history of trauma/infection	No history of trauma/ infection	History of trauma present	History of trauma present
Deformity usually noted after becoming aware of bony swellings (family history of similar bony swellings may be present)	Deformity usually noted in early childhood	Deformity usually noted in early childhood	Deformity noted soon after completion of initial treatment of fracture of the femur or tibia	Valgus deformity of the tibia noted a couple of months after satisfactory treatment of initial proximal tibial fracture
Progressive deformity	Progressive deformity	History of some resolution of deformity over time may be present	History of some resolution of deformity may be present	History of some resolution of deformity over time may be present
Physical examination				
Multiple swellings on the limbs Deformities (varus or valgum) may be asymmetric	Multiple swellings on the limbs Deformities (varus or valgum) may be asymmetric	Varus deformity of the tibia (commonly); less frequently varus deformity of the distal femur and rarely valgus deformity of the distal femur	Varus or valgus deformity of the tibia or femur	Valgus deformity of the tibia

Investigations				
Plain radiograph: Multiple osteochondromata in the metaphyses of the long bones	Plain radiograph: Multiple lesions with calcification in the metaphyses of the long bones	Plain radiograph: Linear lytic defect in the metaphysis of the tibia or femur extending from the cortex surrounded by rim of sclerosis	Plain radiograph: Malunion of previous fracture	Plain radiograph: No evidence of previous fracture; valgus angulation at the metaphysis
Diagnosis				
Genu varum or valgum secondary to hereditary multiple osteochondromatosis	Genu varum or valgum secondary to enchondromatosis	Focal fibrocartilaginous defect in the tibia or femur	Malunion of the tibial or femoral fracture	Cozen phenomenon
Soft tissue pathology				
History				
History of paralytic illness		History of limited joint motion		
		History of injection into the thigh, trauma to the thigh, or infection (e.g., femoral osteomyelitis) may be present		
Physical examination				
Genu valgum with or without knee flexion deformity		Genu valgum with limitation of passive knee flexion		
Ober test positive		Ober test may be positive, or it may not be possible to perform the test due to knee stiffness		
Hip abduction, flexion, and external rotation deformity may be present		Quadriceps contracture present		
Paralysis of muscles around the hip and knee may be present		Patella may be dislocated		
Investigations				
Plain radiograph not indicated		Plain radiograph: To evaluate shape of the femoral condyles (not for establishing the diagnosis)		
Diagnosis				
Genu valgum due to contracture of the iliotibial band (typically seen following poliomyelitis)		Genu valgum due to contracture of the iliotibial band and vastus lateralis muscle (typically seen in acquired quadriceps contracture)		

20.2 Establishing the Diagnosis of the Cause of Flexion or Extension Deformity of the Knee

20.2.1 Introduction

Sagittal plane deformities are never physiologic in a child who is walking, and hence it is imperative that the underlying cause is diagnosed and appropriate treatment instituted.

20.2.2 Questions to Establish a Diagnosis

- **Are there signs of inflammation of the knee joint, and are movements of the knee joint painful?**
- **Is there evidence of muscle weakness, muscle spasticity, and muscle imbalance at the knee?**
- **Are the gastrocsoleus or hamstring muscles contracted?**
- **Is the deformity at the joint, epiphysis, physis, or metaphysis?**
- **Is the deformity progressing, resolving, or remaining static?**
- **Is the deformity associated with shortening?**

Are there signs of inflammation of the knee joint, and are movements of the knee joint painful?

One of the commonest causes of a flexion deformity of the knee is muscle spasm triggered by some painful pathology in the knee. Any form of synovitis or arthritis that affects the knee will result in spasm of the hamstring muscles with flexion of the knee.

Is there evidence of muscle weakness, muscle spasticity, and muscle imbalance at the knee?

Muscle imbalance due to weakness of the quadriceps with normally functioning hamstring muscles can result in a flexion deformity of the knee. Similarly spasticity of the hamstrings can cause a knee flexion deformity.

Are the gastrocsoleus or hamstring muscles contracted?

While it is obvious that a contracture of the hamstring muscles will produce a flexion deformity of the knee, the effect of contracture of the gastrocsoleus on the knee is less well understood. The gastrocsoleus normally produces a plantar flexor-knee extensor force couple, and consequently a contracted gastrocsoleus can result in hyperextension deformity of the knee.

Is the deformity at the joint, epiphysis, physis, or metaphysis?

It is imperative that the site of the deformity is accurately identified to plan appropriate treatment.

Is the deformity progressing, resolving, or remaining static?

Progression of the deformity will be seen if the underlying pathology is growth plate injury. A malunion of a fracture with flexion or extension deformity in the metaphyseal region of the femur or tibia is likely to resolve to a large extent. Deformities secondary to contractures tend to remain static.

Is the deformity associated with shortening?

Shortening associated with a flexion or extension deformity may be seen if the growth plate is damaged or if there is paralysis of muscles.

20.2.3 Physical Examination

Look

Note the deformity of the knee and observe if the knee is swollen.

Feel

Palpate the knee to determine if there is increased warmth over the joint and if the joint

is tender. Note if there is synovial effusion; the patella tap may be elicited if the joint is not acutely painful. Palpate the fold of synovium at the upper limit of the suprapatellar pouch, and note if it is thickened or tender. Palpate the quadriceps, hamstrings, and the gastrocsoleus muscles, and note if there are areas of induration or tenderness.

Move

Check the range of passive motion of the knee. Note if the passive range of motion of the knee alters with the position of the hip. In particular, note if the range of knee extension is reduced if the hip is flexed (suggestive of contracture of the hamstring muscles), and note if the range of knee flexion increases when the hip is flexed (suggestive of contracture of the rectus femoris).

20.2.4 Investigations to Confirm the Diagnosis

Inflammatory Markers

Full blood counts, erythrocyte sedimentation rate, C-reactive protein estimation, and tests for antinuclear antibodies should be done if the joint is swollen and inflamed. Tests to exclude a bleeding disorder like hemophilia should be done if the history suggests that spontaneous hemarthroses have developed on more than one occasion.

Plain Radiographs

Plain radiographs of the knee are mainly to confirm that there is no underlying bony abnormality such a malunion of a metaphyseal fracture or features of an anteriorly or posteriorly situated physeal bar.

20.2.5 Differential Diagnosis of Causes of Flexion or Extension Deformities of the Knee

Spasm of the Hamstrings

Any painful condition of the knee will cause the hamstring muscles to go into protective muscle spasm, and hence the knee is held flexed. If the underlying condition resolves, soon the deformity resolves. However, in chronic arthritic conditions, fixed deformities may develop on account of contracture of the hamstrings.

Spasticity of the Hamstrings

In cerebral palsy there is often spasticity of the hamstring muscles. This is not usually evident in the young child but tends to become manifest as the child grows older.

Contracture of the Hamstrings

The hamstring muscles may become contracted either as a consequence of primary muscle disease such as myopathies, intramuscular hemangiomas, and ischemic fibrosis or become secondarily contracted following long-standing muscle spasm or spasticity.

20.2.6 Establishing the Diagnosis

An outline of the process of establishing a diagnosis of the cause of flexion or extension deformities of the knee is shown in Table 20.4.

Table 20.4 Outline of establishing the cause of a flexion or extension deformity of the knee in a young child

Flexion deformity of the knee		
History		
Pain in the knee preceded the onset of the deformity	No pain preceding deformity	Pain may or may not have preceded onset of deformity
	History of developmental delay may be present	
Physical examination		
Fixed flexion deformity Gentle stretching may partially correct the deformity	Flexion deformity Gentle stretching corrects the deformity	Flexion deformity Gentle stretching does not partially correct the deformity
Flexion deformity gets accentuated when hip is flexed	Flexion deformity gets accentuated when hip is flexed	Flexion deformity gets accentuated when hip is flexed
Pain on attempting to extend the knee beyond the deformed position Pain may be present even on flexing the knee beyond the deformed flexed position	No pain on attempting to extend the knee beyond the deformed position	No pain on attempting to extend the knee beyond the deformed position unless the hamstring muscle is diseased
	Spasticity demonstrable	
Investigations		
Ultrasound: May confirm the presence of effusion	No investigation indicated to diagnose cause of flexion deformity	No investigation indicated to diagnose cause of flexion deformity
Plain radiograph to exclude bony lesion		
Diagnosis		
Protective muscle spasm of hamstrings due to inflammatory or traumatic pathology in the knee	Hamstring spasticity	Hamstring contracture
Extension or hyperextension (recurvatum) deformity of the knee		
History		
History of injections into the thigh or history of osteomyelitis of the femur	History of clubfoot or isolated equinus deformity of the ankle	History of injury to the distal femur or infection in the region of the knee
No progression of deformity	Negligible progression of deformity	Progression of deformity
No tendency for recurrence after correction	No tendency for recurrence after correction	Tendency for recurrence of deformity following correction
Physical examination		
Passive hyperextension of the knee may be present Limitation of active and passive flexion of the knee present	Passive hyperextension of the knee possible No limitation of passive flexion of the knee	Active and passive hyperextension of the knee possible Terminal flexion of the knee limited by the same degree as the degree of hyperextension
Total range (arc) of motion of the knee markedly reduced	Total range (arc) of motion of the knee greater than normal	Total range (arc) of motion of the knee normal
Patella may be high (patella alta)	Patella not high	Patella not high
No ankle deformity	Equinus of the ankle (either dynamic or true equinus contracture)	No ankle deformity
Unable to squat because of limitation of knee flexion	Able to squat but not normally because of equinus (if contracture of the gastrocsoleus is present)	Able to squat but not normally because of limitation of terminal knee flexion
Gastrocsoleus muscle normal	Spasticity or contracture of the gastrocsoleus present	Gastrocsoleus muscle normal

Table 20.4 (continued)

Investigations		
Plain radiographs: Evidence of old healed osteomyelitis of the distal femur may be present Patella alta may be present	Plain radiograph not indicated	Plain radiographs: Growth plate arrest anteriorly of the distal femoral physis or the proximal tibial physis may be evident in the lateral view
MRI not indicated	MRI not indicated	MRI: Extent and location of physeal damage can be identified more clearly on the MRI
Diagnosis		
Quadriceps contracture secondary to injection fibrosis, ischemic fibrosis, or postinfective fibrosis	Hyperextension of the knee (recurvatum) secondary to ankle equinus	Hyperextension of the knee secondary to anterior growth plate arrest of the distal femur or proximal tibia

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In-Toeing and Out-Toeing Gait

21

James Robb

21.1 Introduction

In-toeing and out-toeing, or inward and outward rotation of the foot with respect to the direction of walking, respectively, are common reasons for parental concerns and referral for a specialist opinion. A normal range of values can be considered to lie within two standard deviations on either side of the mean of the values for a given population (Jacquemier et al. 2008) (Fig. 21.1).

Values outside the normal range, however, do not necessarily imply pathology or that treatment is required. These normal variants are typically symmetric and painless and there is no joint stiffness.

21.2 Questions to Establish a Diagnosis

- **Was this gait pattern noted as soon as the child started walking or is it of recent onset?**
- **Is the in-toeing or out-toeing bilateral and symmetric?**
- **Does the child sit on the ground in the "W" position?**
- **What is the site of the problem?**
- **Is there an associated deformity of the hip, knee, or foot in the coronal plane?**

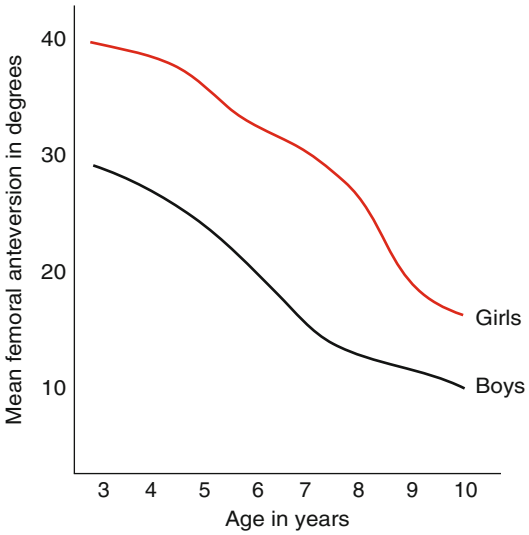

Fig. 21.1 Values for femoral anteversion measured by the maximum lateral trochanteric prominence test reduce as the child grows (Modified from Jacquemier et al. (2008))

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Was this gait pattern noted as soon as the child started to walk or is it of recent onset?

In-toeing or out-toeing of recent onset is likely to be pathologic, whereas most normal variants of torsion manifest as soon as the child starts to walk.

Is the in-toeing or out-toeing bilateral and symmetric?

Bilateral symmetric in-toeing or out-toeing is innocuous in the vast majority of instances, particularly if present from the time the child started walking (Staheli et al. 1985). On the other hand, unilateral in-toeing or out-toeing is likely to be pathologic.

Fig. 21.2 A child with excessive femoral anteversion sitting in the "W" posture

Does the child sit on the ground in the "W" position?

Children with increased femoral anteversion, one of the most common causes for in-toeing, tend to sit in the "W" position (Fig. 21.2).

What is the site of the problem?

The underlying problem that leads to in- or out-toeing may lie in the hip joint, femur, or tibia or in the hindfoot or forefoot (Table 21.1). It is important to identify the site of abnormality as treatment, if warranted, should be directed to the appropriate site of pathology.

Is there an associated deformity in the coronal plane?

Infantile coxa vara is often associated with retroversion of the femur which in turn causes an out-toeing gait. Tibia vara is usually associated with internal tibial torsion which results in an in-toeing gait. Hindfoot varus of clubfoot may be associated with internal tibial torsion and in-toeing.

Table 21.1 Site of pathology in children with in-toeing or out-toeing

Site of pathology	Abnormality	Effect on gait
Hip joint	Internal rotation contracture	In-toeing gait
	External rotation contracture	Out-toeing gait
	Spasticity of internal rotators of the hip	In-toeing gait
Femur	Femoral anteversion	In-toeing gait
	Femoral retroversion	Out-toeing gait
	Coxa vara with retroversion	Out-toeing Trendelenburg gait
	Malunion of femoral fracture with rotational malalignment	In- or out-toeing depending on the nature of malunion
Tibia	Internal tibial torsion	In-toeing gait
	External tibial torsion	Out-toeing gait
Hindfoot	Residual deformity of clubfoot with failure of rotation of the calcaneus	In-toeing gait
Forefoot	Residual forefoot adduction of clubfoot at midtarsal joint	In-toeing gait
	Metatarsus adductus	
	Metatarsus primus varus	

21.3 Physical Examination

21.3.1 Look

Foot Progression Angle

Have the child suitably undressed and observe the gait noting the foot progression angle (FPA) which describes the orientation of the child's foot with respect to the direction of progression (Fig. 21.3; Video 21.1). By skeletal maturity, the majority of children would have the adult pattern with an external FPA of about 15°.

Observe the orientation of the child's patellae with respect to the direction of progression to try to determine abnormal internal or external rotation of the femur in gait. Marking the outline of the patellae with a pen can help (Fig. 21.4).

Observe the gait to ensure that there is no Trendelenburg gait. Look for associated deformities of the tibia such as tibia vara and for features of residual deformities of clubfoot.

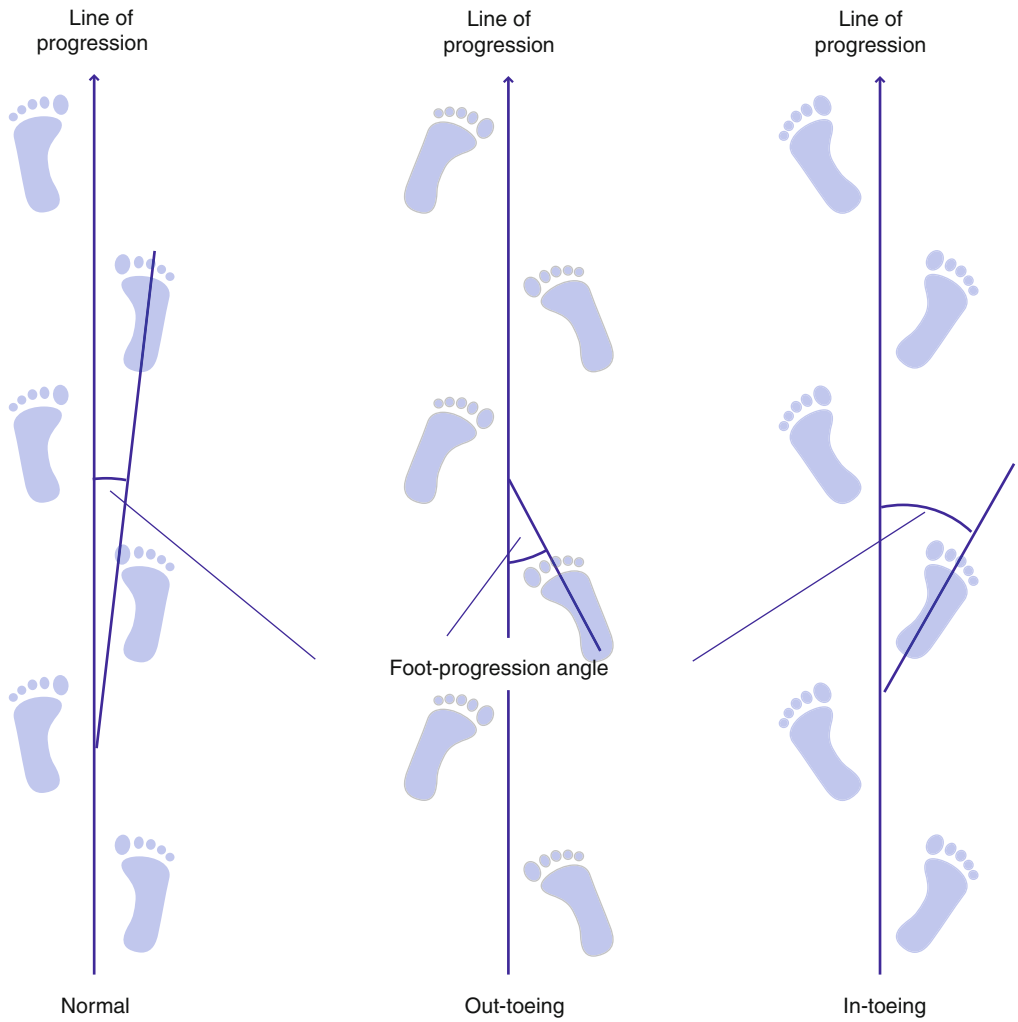

Fig. 21.3 The foot progression angle (*FPA*) relates to the plane of foot in relation to the line of progression

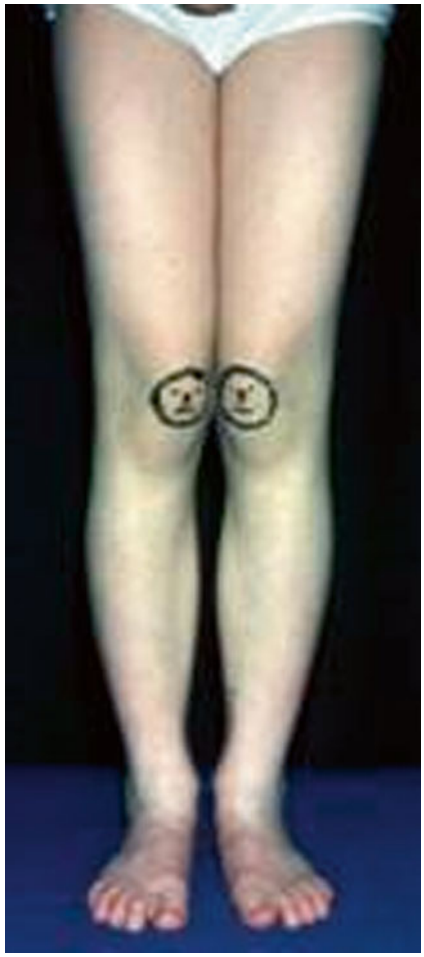

Fig. 21.4 The patellae are marked on this 12-year-old boy. The patellae face medially because of excessive femoral anteversion; the feet point forward because he has excessive external tibial torsion

21.3.2 Move

Hip Rotation

With the child prone, evaluate the arc of hip rotation by flexing the child's knees to 90° and using the tibiae as a pointer. Place both hips in maximal internal and then external rotation. The tibiae will cross during external rotation (Fig. 21.5).

Ensure that you do not tilt the pelvis at the end of range of hip movement. Other movement of the hips should also be carefully measured and they can be recorded on a grid (Rao and Joseph 2001) (Fig. 21.6).

The total arc of rotation is normally about 90° but can be more in children who have joint laxity, and both internal and external rotations are roughly equal. Internal rotation exceeds external rotation when femoral anteversion is present, and external rotation is greater when the femur is retroverted; the total arc of rotation remains normal in both these instances (Fig. 21.7a). When there is internal rotation or external rotation contracture, the arc of rotation is reduced in the opposite direction (Fig. 21.7b).

Clinical Estimation of Femoral Anteversion

An indirect, clinical estimation of femoral anteversion can be made at this stage by placing the child prone (Fig. 21.7). The limb not being examined is laid flat on the couch. Flex the knee of the side to be examined to 90° and use one hand to hold the tibia. Use the palm of the other hand to identify the lateral prominence of the greater trochanter on the lateral aspect of the ipsilateral proximal thigh. Rotate the tibia inwardly and outwardly until the greater trochanter is at its most prominent position. The anteversion value is the angle between the tibia and the vertical (Ruwe et al. 1992) (Fig. 21.8).

Foot-Thigh Angle

This is an indirect measurement of tibial torsion. The knee is flexed to 90° and the axis of the foot is compared to the axis of the thigh by visually superimposing the foot axis over the thigh (Fig. 21.5). The normal range is about 15–30° external. This test is sufficient for clinical and screening purposes (Staheli et al. 1985). Errors in measurement can be introduced by not aligning the coronal longitudinal axis of the calcaneus with that of the tibia and introducing rotation through the flexed knee.

Forefoot Adductus

The knee is flexed to 90° and the plantar surface of the foot inspected (Fig. 21.5). The lateral border is usually straight and a convex border suggests forefoot adductus. If there is adductus, the flexibility of the forefoot can be assessed.

Alternative Examination Techniques

If the child does not wish to lie prone, hip rotation can be evaluated with the child supine. The hips and knees are extended and the examiner can roll each thigh in turn into maximal internal

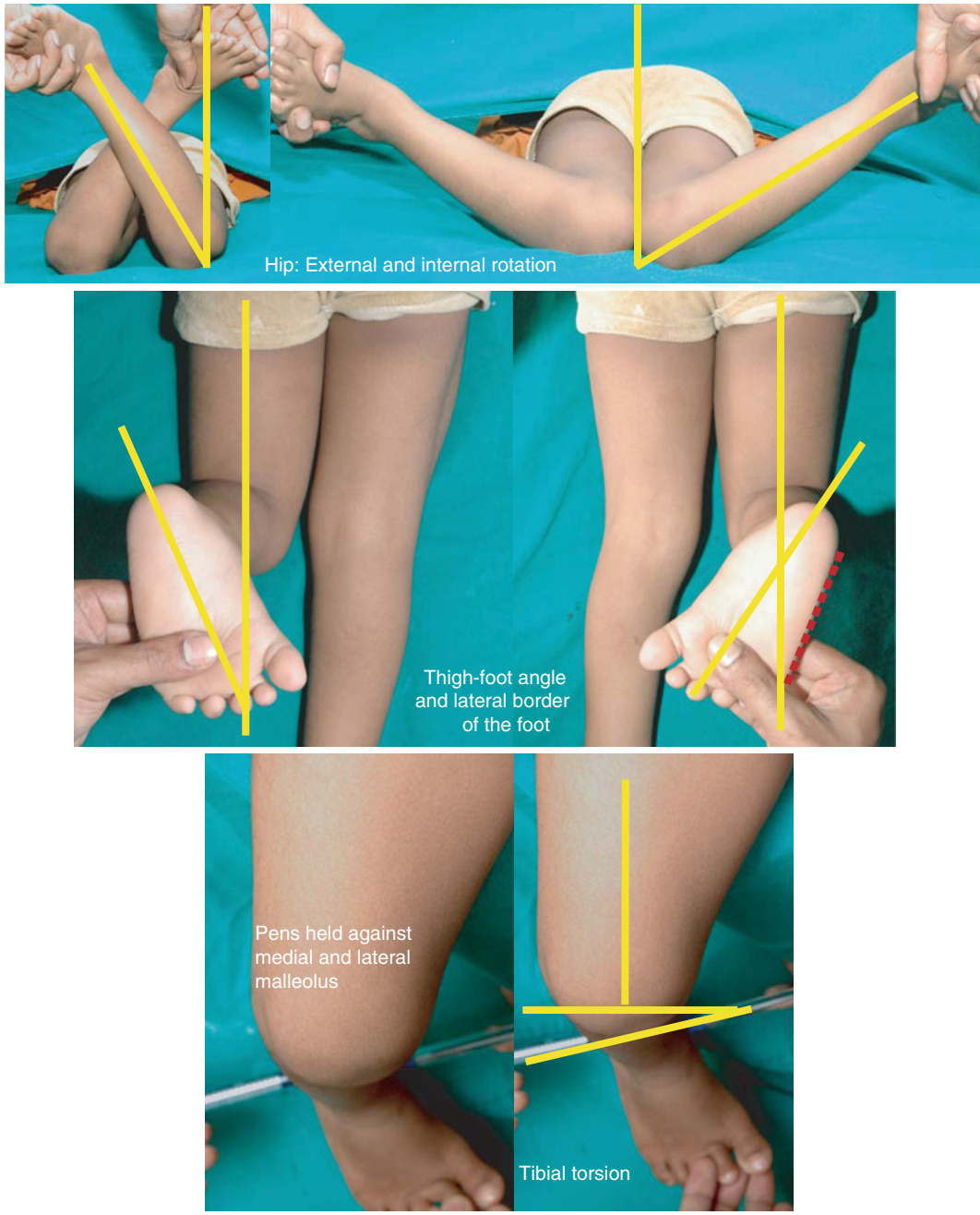

Fig. 21.5 Method of assessing the torsional profile clinically; the extent of external rotation with the hip in extension (*top left*) and the range of internal rotation are seen (*top right*). The thigh-foot angle is a measure of tibial torsion (*middle*); another method of estimating tibial torsion is to measure the transmalleolar axis (*bottom*). The lateral border of the foot is also noted (*red dotted line*)

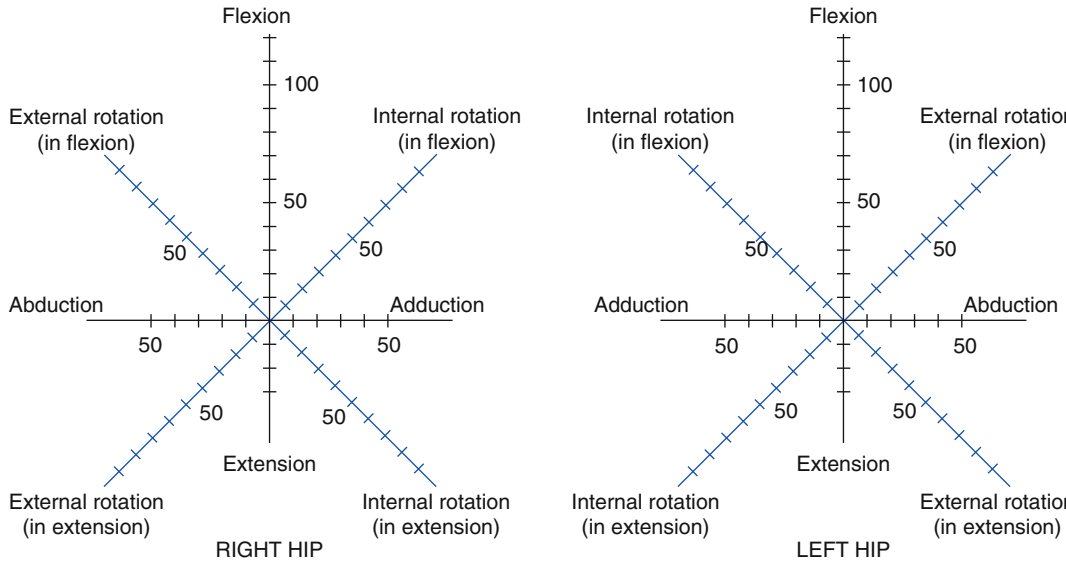

Fig. 21.6 The range of hip motion can be recorded on a grid

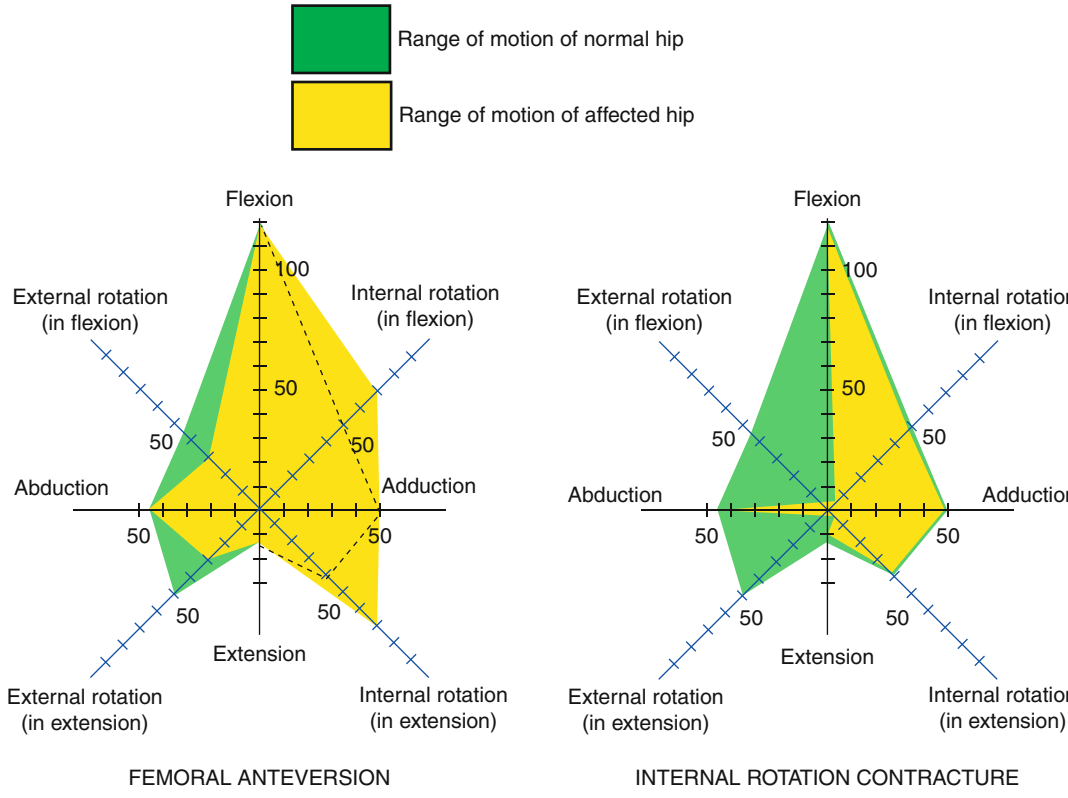

Fig. 21.7 The range of passive motion of the hip in a child with excess femoral anteversion is shown (**a**). There is a reduction in the range of external rotation at the expense of an increase in internal rotation. In a child with an internal rotation contracture resulting in a fixed internal rotation deformity of 5°, there is an absolute reduction in the total arc of motion (**b**)

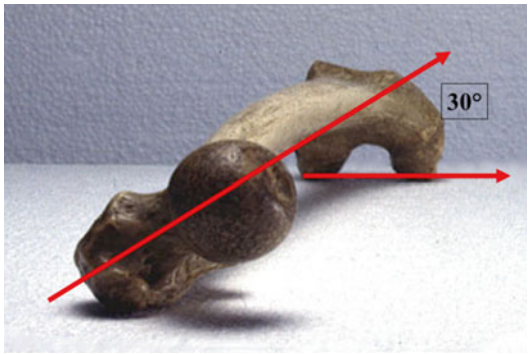

Fig. 21.8 Excessive anteversion in a cadaveric femur. When the trochanter is most prominent, the femoral neck will be horizontal and the transcondylar femoral axis will be 30° internally rotated. If the knee is then flexed, the tibia will also be 30° internally rotated with respect to the vertical

and external hip rotation. The child can also be positioned with their knees bent over the end of the couch and the examiner sits at the end of the couch and rotates the hips by holding the child's tibiae but taking care not to produce pelvic tilt. In the same position, an indirect estimation of tibial torsion can be obtained by comparing the transmalleolar axis at the ankle with the transcondylar femoral axis at the knee while the child lies or sits on the couch with their knees flexed over the end of the couch. Foot shape can also be evaluated in this position.

Do not omit to carry out a standard orthopedic examination of the child as well and exclude neuromuscular disorders such as cerebral palsy, spina bifida, or muscular dystrophy. If there are features of cerebral palsy or other upper motor neuron paralysis, look for evidence of spasticity of the internal rotators of the hip in addition to estimating the torsional profile. If a lower motor neuron paralysis is present, careful muscle power charting is mandatory.

21.4 Investigations to Confirm the Diagnosis

Clinical examination will suffice in the great majority of cases of torsional abnormalities of the femur and tibia, but where there is residual doubt or if torsions are not improving as anticipated, a CT scan or ultrasound scan may be indicated (Fig. 21.9). The only precaution needed while performing these scans is to ensure that the child does not move between acquisition of the proximal and distal scan of the bone. If there is an actual reduction in the arc of motion of the hip or evidence of a Trendelenburg limp, imaging of the pelvis and hip is essential. An anteroposterior radiograph of the foot is required if there is forefoot adductus.

21.5 Differential Diagnosis

21.5.1 In-Toeing

This is a normal feature during growth for many children but can be a source of parental concern. Parents may feel that their child trips frequently or may look awkward when running. Bear in mind that "excessive" tripping or falling may coincide with the child's developmental stage but may also be a symptom of progressive muscle weakness, for example, that seen in muscle dystrophies. In-toeing is more frequently encountered than out-toeing. There are three main causes for in-toeing (metatarsus adductus, internal tibial torsion, femoral anteversion, or internal femoral torsion) occurring at three different anatomical levels, foot, tibia, and femur, and these may appear in isolation or in combination.

Metatarsus Adductus

In metatarsus adductus the forefoot is adducted at the tarsometatarsal joints (Fig. 21.10a, b). It is usually apparent after birth and is flexible and bilateral in 50 % of patients. It may be associated with hip dysplasia in 10–15 % of children. This may be an example of a "packaging deformity" and secondary to intrauterine positioning (Fig. 21.11).

Metatarsus adductus usually resolves spontaneously and does not require any treatment. Occasionally the adducted forefoot is stiff and does not resolve spontaneously. Such feet may be associated with a plantar crease and a trapezoidal deformity of the medial cuneiform bone. A resistant deformity can be managed initially in serial above-knee casts to stabilize the hindfoot while abducting the forefoot, but surgery is rarely needed.

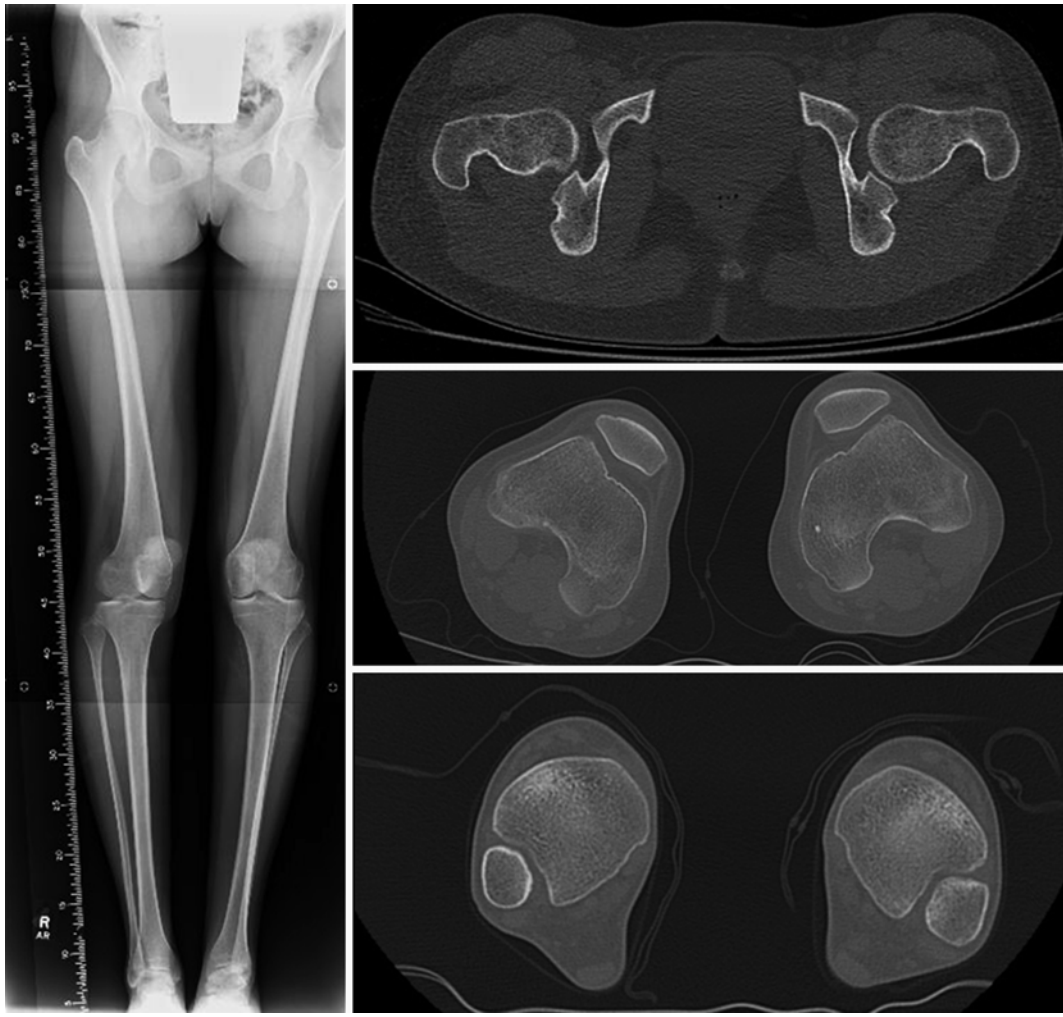

Fig. 21.9 Radiological evaluation of torsion with CT scans

Residual Forefoot Adduction in a Partially Treated Clubfoot

In-toeing may result from a partially treated clubfoot; a clue to this cause will be obtained from the child's history. The bony pathology is complex in clubfoot (Fig. 21.10c, d). It includes a subluxation of the talonavicular joint, largely responsible for the appearance of adductus that originates in the midfoot, and a loss of the normal divarication of the talus and calcaneus (Windisch et al. 2007). An inadequate reduction of the deformities or a relapse after treatment is the usual cause of residual forefoot adduction in clubfoot. In contrast, the deformity in metatarsus adductus is confined to the forefoot and there is no deformity of the mid- or hindfoot or ankle.

Internal Tibial Torsion

Up to about 30° of internal torsion may be present at birth. This is usually seen during the second year of life and is the most common cause of in-toeing. It affects both sexes equally and is bilateral in about two-thirds of affected infants (left side affected more than the right). Internal tibial torsion should normally resolve by about 10 years of age. Since the natural history is one of spontaneous resolution, no active treatment is

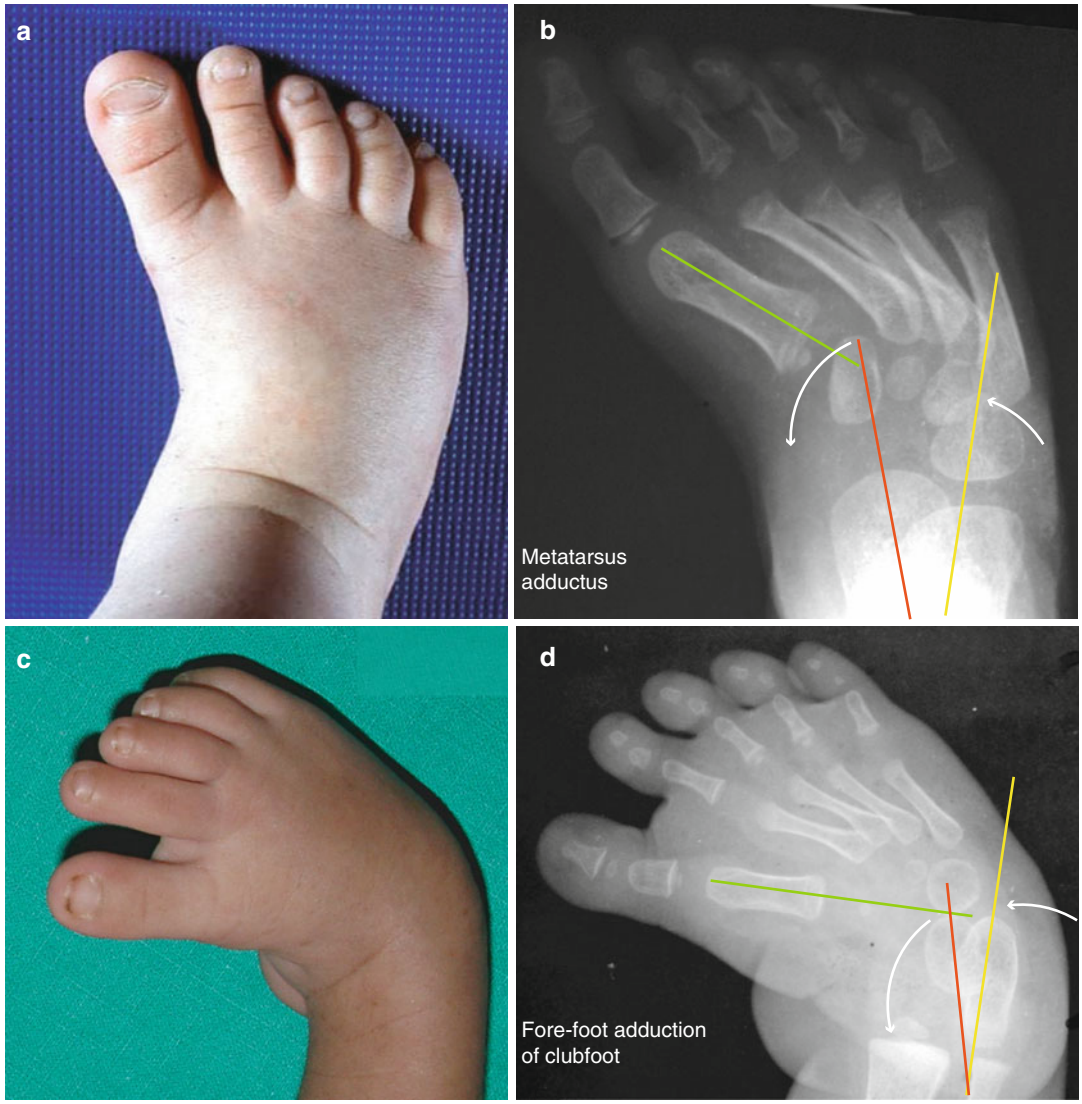

Fig. 21.10 Clinical appearance and radiographs of metatarsus adductus (**a**, **b**) and forefoot adduction of clubfoot (**c**, **d**). The long axes of the calcaneus (*yellow*), talus (*red*), and the first metatarsal (*green*) are marked. In metatarsus adductus, the long axis of the calcaneus bisects the cuboid, but in clubfoot it passes lateral to the cuboid. The deformity of metatarsus adductus is confined to the tarsometatarsal joint, while the forefoot adduction deformity of clubfoot is at the midtarsal joint (*white curved arrows*)

necessary. Braces, splints, and exercises are ineffective. Supramalleolar derotation of the tibia may be indicated if the torsion does not resolve and causes a functional problem.

Internal tibial torsion is often associated with idiopathic congenital clubfoot and in-toeing may persist after correction of the clubfoot deformity (Fig. 21.12).

Internal Femoral Torsion or Femoral Anteversion

Femoral anteversion is about 45° in the neonate and usually gradually decreases to about 10° and 15° in a skeletally mature male and female, respectively. Non-resolution of childhood anteversion to normal adult values is one cause of in-toeing. Females are more commonly affected

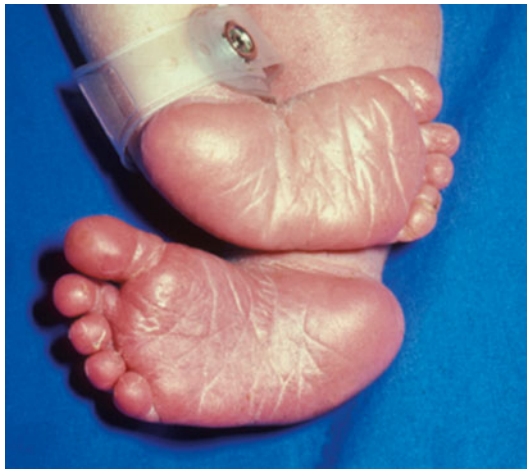

Fig. 21.11 Intrauterine molding of the lower limbs

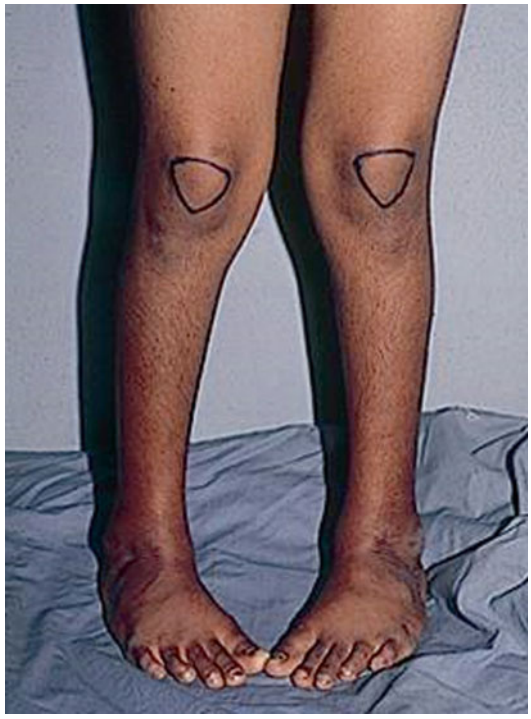

Fig. 21.12 In-toeing in a 10-year-old girl with treated idiopathic clubfeet. The in-toeing was on account of internal tibial torsion

(approximately 2:1), and the in-toeing is most apparent between 4 and 6 years of age and typically resolves by the age of 10. When standing, the patellae point medially, and the child sits in the "W" position because they lack sufficient external hip rotation, to sit cross-legged. The child appears awkward when running because of the greater arc of internal than external rotation. For some children higher anteversion values persist and these children will in-toe as adults. This rarely causes a functional problem. If the persisting internal torsion is severe and does cause a functional problem, it can be corrected by a femoral derotation osteotomy. Asymmetric hip rotation between sides is an indication for hip radiography.

Red Flags

- Unilateral in-toeing: examine the upper limbs and exclude hemiplegia.
- In-toeing and tripping that is not improving in a male toddler or one of school age: think of muscle disease, e.g., the dystrophies.
- Stiff, adducted forefoot that is not improving.
- In-toeing associated with pain, limp, or joint stiffness: search for other causes, e.g., bone or spinal tumor, infection, or inflammatory conditions of the joint or bone.

21.5.2 Out-Toeing

External tibial torsion may worsen with time and require surgical correction.

External femoral torsion (retroversion) is often associated with infantile coxa vara which needs treatment. Isolated external femoral torsion, however, does not usually cause symptoms severe enough to necessitate a derotation osteotomy but may be associated with an increased risk of developing a slipped upper femoral epiphysis (SUFE). Out-toeing may worsen with time and excessive tibial and femoral external rotation may also produce an externally rotated FPA and parents may feel that their child has a flat foot.

Red Flags

- Loss of internal femoral rotation and associated hip or knee pain: consider hip pathology such as Perthes' disease as it can occur occasionally in the preschool age. In the older child exclude a SUFE.

- Out-toeing associated with pain, limp, or joint stiffness: search for other causes, e.g., bone or spinal tumor, infection, or inflammatory conditions of the joint or bone.

21.5.3 Less Common Conditions

Miserable Malalignment Syndrome

This usually becomes noticeable in adolescence but the child should be followed up if the condition is suspected when younger. It consists of excessive internal femoral torsion and an external tibial torsion, but the FPA falls within a normal range (Figs. 21.3 and 21.13). The adolescent may complain of patellofemoral pain or patellar subluxation and occasionally patellar dislocation because the quadriceps tends to pull the patella laterally as the knee is rotated medially. Femoral and tibial derotation osteotomies are the only solution for persisting symptoms (Delgado et al. 1996).

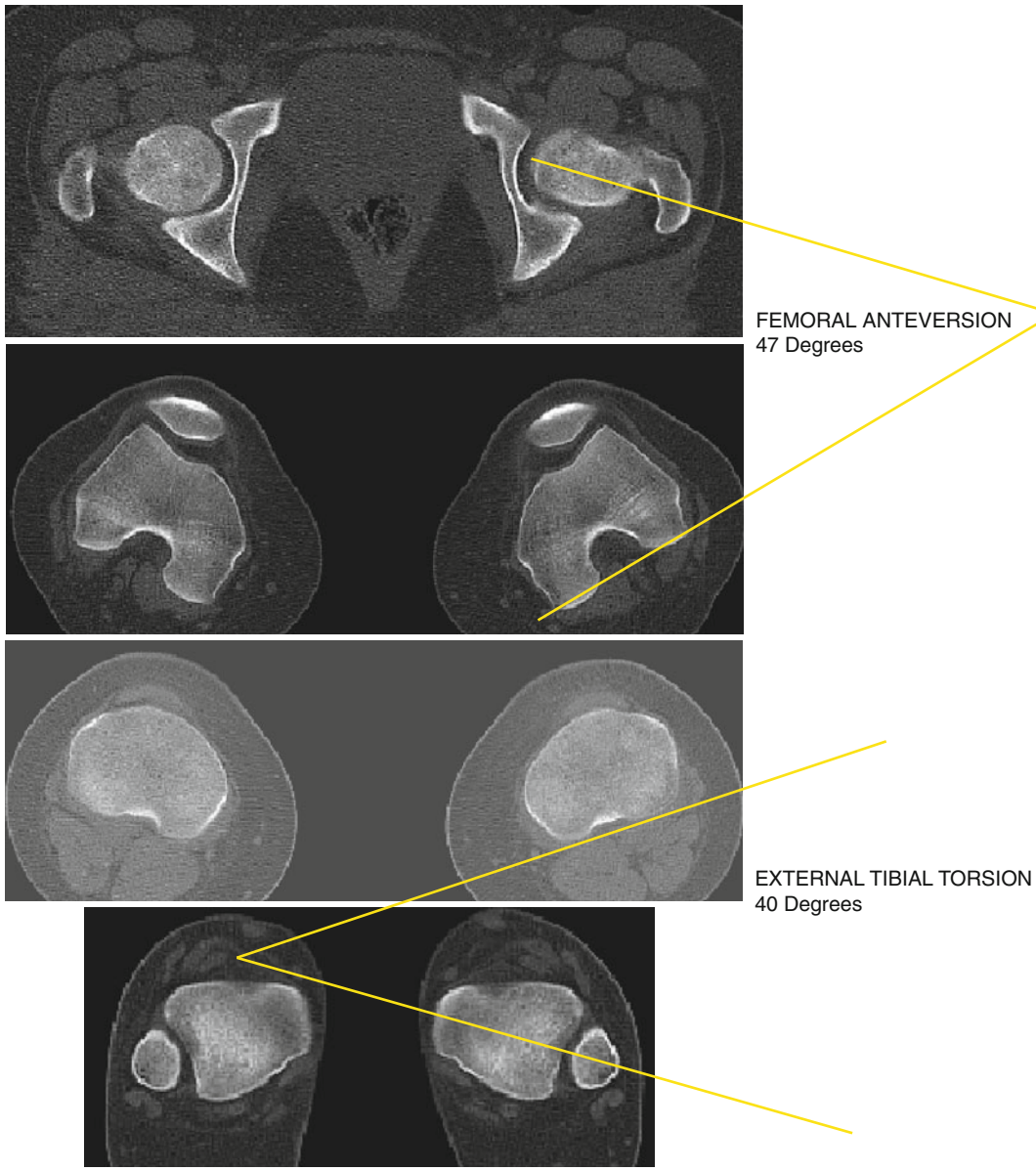

Fig. 21.13 Miserable malalignment

Skew Foot

These are rare but can give the appearance of forefoot adduction (Fig. 21.14). However, the midfoot is abducted and the heel in valgus in contrast to metatarsus adductus which only affects the forefoot (Berg 1986).

21.6 Establishing the Diagnosis

An outline of establishing the diagnosis of the cause of in-toeing is shown in Table 21.2.

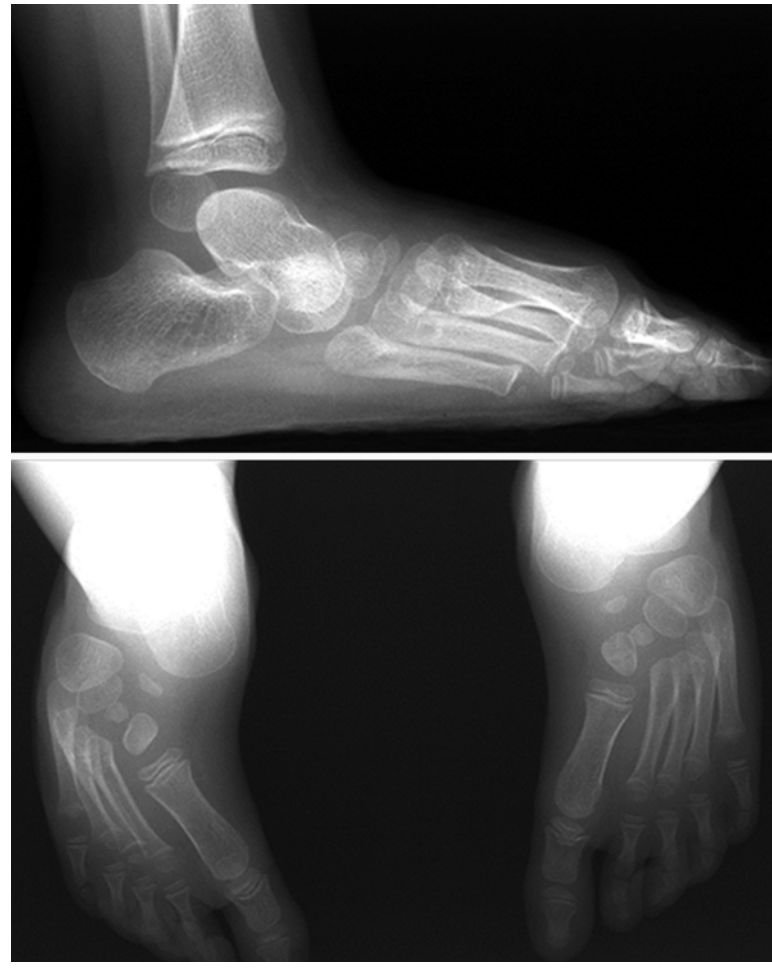

Fig. 21.14 Skew feet

Table 21.2 Establishing the diagnosis of in-toeing in toddlers and young children who have no pain or limp (apart from in-toeing)

History			
In-toeing noted since the child began to walk	In-toeing noted since the child began to walk	In-toeing noted since the child began to walk	In-toeing noted since the child began to walk
Sits in the "W" position	Sits normally	Sits normally	Sits normally
History of some spontaneous resolution may be present	History of some spontaneous resolution may be present	No history of spontaneous resolution	History of some spontaneous resolution may be present
No foot deformity at birth	History of treated congenital clubfoot may be present	History of treated congenital clubfoot present	History of clubfoot may or may not be present
Physical examination			
No limp apart from in-toeing	No limp apart from in-toeing	No limp apart from in-toeing	No limp apart from in-toeing
Internal rotation of the hip exceeds external rotation Other movements *not* affected	Hip movements normal (except if femoral anteversion coexists)	Hip movements normal	Hip movements normal
Clinical estimation of femoral anteversion shows that the angle is higher than normal	Normal anteversion angle (except if femoral anteversion coexists)	Normal anteversion angle	Normal anteversion angle
Cannot sit cross-legged	Able to sit cross-legged	Able to sit cross-legged	Able to sit cross-legged
Thigh-foot angle normal	Thigh-foot angle internal	Thigh-foot angle may be normal or internal (may be difficult to estimate accurately)	Thigh-foot angle may be difficult to estimate accurately because foot is deformed
Tibia not bowed	Genu varum and tibia vara may be present	Tibia not bowed	Tibia not bowed
Bi-malleolar axis normal	Bi-malleolar axis indicates internal tibial torsion	Bi-malleolar axis may indicate internal tibial torsion	Bi-malleolar axis normal
Lateral border of the foot straight	Lateral border of the foot straight	Lateral border of the foot straight (may be curved if forefoot adductus is also present)	Lateral border of the foot curved
Working diagnosis: Femoral anteversion	Working diagnosis: Internal tibial torsion	Working diagnosis: Malrotation of the calcaneus	Working diagnosis: Forefoot adduction due to residual deformity of clubfoot at midtarsal joint or metatarsus adductus
Investigations			
CT scan or ultrasound Increased femoral anteversion can be measured	CT scan or ultrasound Internal tibial torsion can be measured	Plain radiographs of the foot Reduced talocalcaneal angle in the anteroposterior view	Anteroposterior radiograph of the foot Increased talo-first metatarsal angle and calcaneocuboid malalignment in forefoot adduction of clubfoot Increased talo-first metatarsal angle and normal calcaneocuboid alignment in metatarsus adductus
Diagnosis			
Femoral anteversion	Internal tibial torsion	Calcaneal malrotation in residual clubfoot	Residual forefoot adduction of clubfoot *or* metatarsus adductus

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Toe Walking

22

James Robb

22.1 Introduction

Gait Maturation

Children begin walking independently on average at about 14 months of age. Infant gait is characterized by high stepping as the heel and toes are lifted and lowered to the ground simultaneously. A heel strike typically appears about 22 weeks after independent walking (Sutherland et al. 1980), and intermittent toe walking is a common pattern in healthy developing children less than 3 years old (Sutherland et al. 1980). Gait is mature by 7–8 years of age. Toe walking is more noticeable when walking barefoot than in shoes as the heel raise of the shoe can help compensate for any structural cause of toe walking.

The cause of toe walking may be central, an abnormality within the CNS, or peripheral, a structural abnormality in the affected limb or limbs. A screening examination to exclude a central cause is essential.

22.2 Questions to Establish a Diagnosis

- **Is there a history of prematurity or fetal distress perinatally?**
- **At what age did the child first walk independently?**
- **Has the child always walked on tiptoe?**
- **Is there a family history of toe walking?**
- **Is the toe walking of recent onset?**
- **Is the tiptoeing unilateral or bilateral?**
- **Is there an associated neurologic condition?**

Is there a history of prematurity or fetal distress perinatally?

Was there fetal distress perinatally and were Apgar scores abnormal at birth? Is there a history of other features of delayed motor milestones such as a delay in the acquisition of independent head control (about 3 months) or sitting (about 6 months)? If the answers to these questions are "yes" it is likely that the child has cerebral palsy. The first orthopedic manifestation of bilateral cerebral palsy in children born prematurely may be a delay in walking and equinus gait.

At what age did the child first walk independently?

Children acquire independent walking by about 14 months of age on average. Consider a delay beyond 18 months as an indication for further assessment for developmental delay. Ninety percent of children with spastic diplegia walk independently by the age of 4 years.

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Has the child always walked on tiptoe?

Toe walking right from when they start walking is common in typically developing children up to the age of three.

Is there a family history of toe walking?

A positive family history of toe walking may be present; one study found a positive family history in 71 % of children.

Is the toe walking of recent onset?

If so, this is likely to be pathologic. A history of recent onset of back pain, limb weakness, or impaired sphincter control must be elicited.

Is the tiptoeing unilateral or bilateral?

Unilateral toe walking suggests an underlying pathology. Is there normal function in the child's ipsilateral upper limb? If not, consider hemiplegia. Ninety percent of children with spastic hemiplegia walk independently by 18 months of age. One cause of unilateral toe walking is a short leg.

Is there an associated neuromuscular condition?

In the majority of instances, there is an underlying neurologic condition, and it is a useful dictum to assume that there is some neuromuscular disease in any toe walker unless proved otherwise.

22.3 Physical Examination

22.3.1 Look

Observe if the tiptoeing is unilateral or bilateral (Fig. 22.1a, b) and then observe the child standing and walking.

22.3.2 Standing

Observe if the child can stand with the feet flat on the ground. Note if in doing so, there is hyperex-

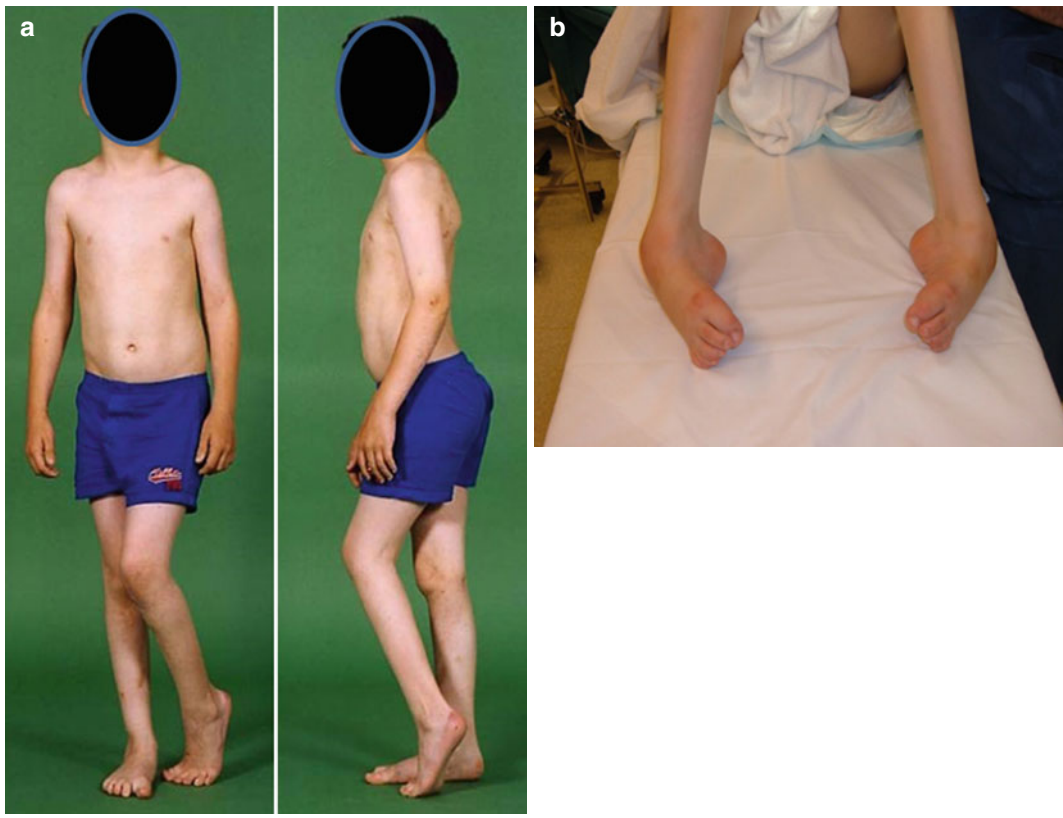

Fig. 22.1 Unilateral equinus in spastic hemiplegia (**a**) and bilateral equinus in spastic diplegia (**b**)

tension of the knee or excessive hindfoot valgus with apparent flattening of the medial arch of the foot. If so, exclude a structural cause for a flatfoot by having the child stand on tiptoe or perform Jack's test (1953) (dorsiflexion of the great toe in the weight-bearing foot will cause an elevation of the medial foot arch in a nonstructural flatfoot).

22.3.3 Walking

Note if consistently initial contact is with the toes and if heel contact is made at any part of the stance phase. Note if there is hyperextension of the knee in stance phase of the gait cycle and hindfoot valgus in the swing phase. See if the child can walk on their heels.

22.3.4 Move

Check if the range of passive ankle dorsiflexion is normal or reduced. If there is a fixed equinus deformity, distinguish between a contracture of the gastrocnemius and the soleus by performing Silferskiöld's test (Fig. 22.2a, b).

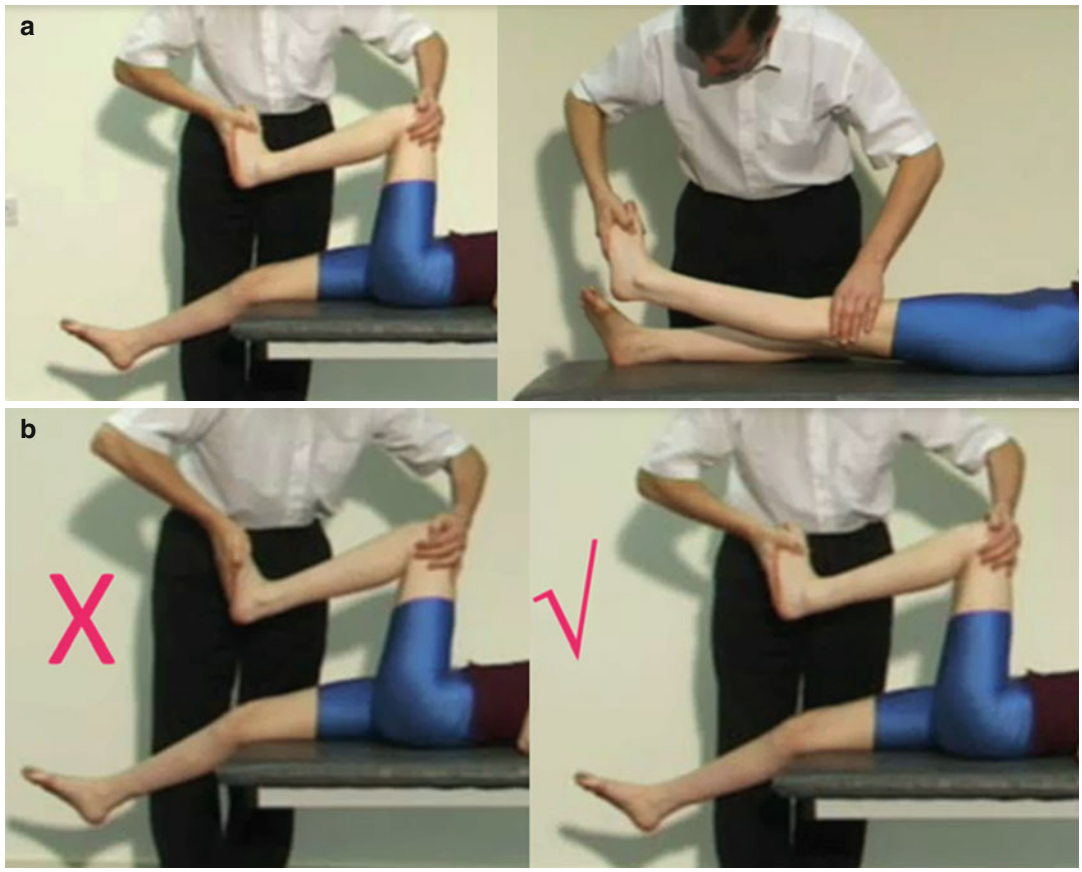

Fig. 22.2 Silferskiöld's test to evaluate gastrocnemius and soleus length individually. In this example (**a**) soleus length is within a normal range, but gastrocnemius is short because the ankle plantar flexes as the knee is extended since gastrocnemius crosses the knee and ankle joints. (**b**) Shows a pitfall in examination. Ensure that the midline of the calcaneus in the coronal plane is aligned with the midline of the calf in the same plane by inverting the subtalar joint into the neutral position (*right side*). This avoids eversion of the foot (*left side*) and "escape hindfoot valgus" which gives the examiner an impression of more ankle dorsiflexion and soleus length than might otherwise be the case

22.3.5 Measure

Measure the limb lengths in children with unilateral toe walking.

22.3.6 Special Tests

Neurologic Examination
Perform a neurologic examination that includes the evaluation of muscle tone and deep tendon reflexes to exclude a UMN or an LMN lesion.

Evaluate muscle strength in all major muscle groups of the upper and lower extremity especially if the child appears weak or floppy.

22.4 Differential Diagnosis

22.4.1 Idiopathic Toe Walking (ITW)

ITW may be defined as occurring in children older than 3 years still walking on their toes without signs of neurologic, orthopedic, or psychiatric diseases (Kallen et al. 1986). It is a diagnosis of exclusion and estimated to occur in 7–24 % of the childhood population (Fox et al. 2006). There may be a positive family history and boys are more commonly affected than girls (Sala et al. 1999). It is important to be aware that the diagnosis of ITW is one of exclusion of all the conditions listed below.

22.4.2 Toe Walking Associated with Dyspraxia or Autism

This is bilateral and has been described as a "ballerina pattern." Children have normal calf muscle length on Silferskiöld's test and normal calf muscle tone and can get their heels on the ground in physiologic alignment without back-kneeing.

22.4.3 Bilateral Cerebral Palsy

There may be a history of prematurity. There will be increased tone in gastrocnemius/soleus which may also be anatomically short on Silferskiöld's test. Calcaneal tendon reflexes will be exaggerated and there may be clonus at the ankles. There may also be a contracture or increased tone in the hip or knee flexors with or without shortening of the gastrocnemius/soleus; the equinus may be a compensation for a more proximal lower limb joint contracture or short limb.

22.4.4 Muscular Dystrophy (MD)

There may be a positive family history in Duchenne's and Becker's MD which only affects boys. In DMD the boy may have a delay in walking as well as learning difficulties and the tiptoeing occurs because of calf muscle weakness. Some boys achieve independent walking and then lose the ability. Calf muscle length will be normal in the early stages, but as the condition progresses boys develop structural shortening of the posterior calf muscles and often calf muscle pseudohypertrophy (Fig. 22.3). Calf muscle tone is normal and calcaneal reflexes may be normal or diminished as a result of the muscle weakness. Gower's sign is when a patient has to use their hands to "walk" up their body from a squatting position because of muscle weakness in the lower limbs. This may be seen in any disorder associated with bilateral lower limb weakness. Creatine kinase (CK) levels are markedly elevated in DMD and to a lesser extent in BMD which runs a more benign course. There is a dystrophin deficiency in DMD and abnormal DMD expression in BMD.

22.4.5 Myopathy

Children with a myopathy are floppy at birth and have a delay in motor development (Fig. 22.4). Posterior calf muscle length may be long, normal, or short as a consequence of immobility, and children are at risk of developing foot deformities and contractures. The tiptoe walking results from calf muscle weakness or foot deformity. Gower's sign may be positive and CK levels are normal.

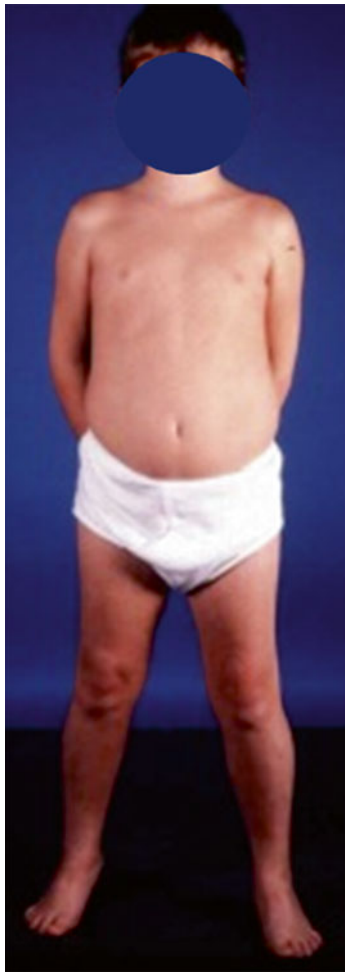

Fig. 22.3 Calf pseudohypertrophy and equinus in DMD

22.4.6 Short Gastrocnemius or Soleus

Here the gastrocnemius and/or the soleus may be anatomically short on Silferskiöld's test, in the absence of any UMN signs. It is usually bilateral (Fig. 22.5).

22.4.7 Unilateral Tiptoeing

This may result as a compensation for a short limb (Fig. 22.6), unilateral contracture of a hip, and knee, ankle, or foot abnormality, e.g., cavus. It is important to ensure that the child has normal upper limb function and not a hemiplegia.

22.4.8 Other Causes

Poliomyelitis
The resulting tiptoe gait results from asymmetric bilateral lower limb weakness or as a compensation for unilateral weakness.

Spinal Dysraphism
Tiptoeing results from bilateral muscle weakness or as a unilateral compensation if the cord lesion is asymmetric.

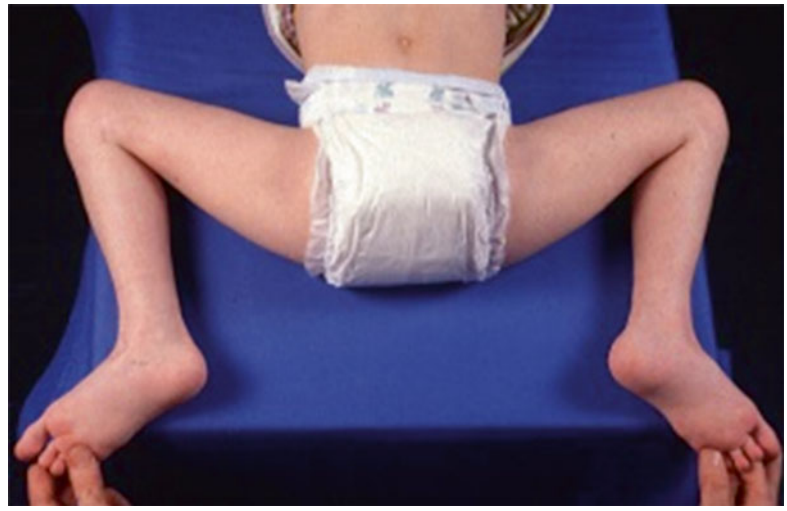

Fig. 22.4 Floppy child with central core disease

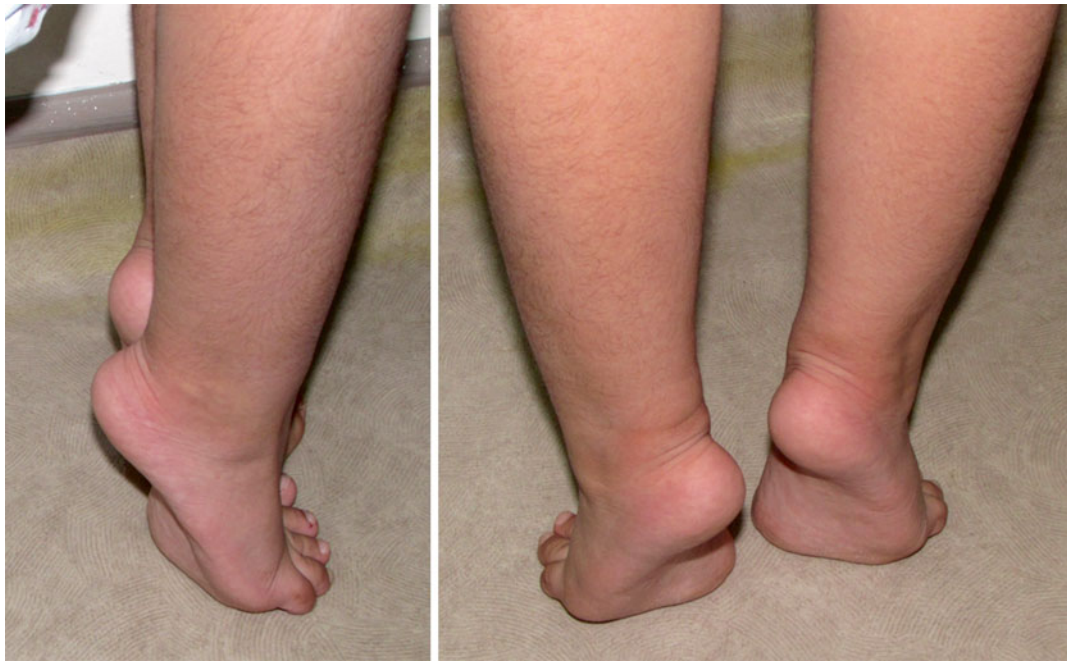

Fig. 22.5 Bilateral equinus in a child with short gastrocsoleus. Lengthening of the Achilles tendons had to be performed to get the feet plantigrade

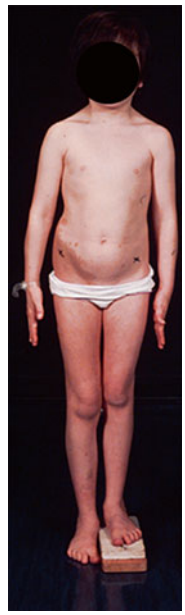

Fig. 22.6 Limb length discrepancy due to a right lower limb hypertrophy. The child is standing on a wooden block to equalize leg length

Spinal Cord Tumor

A spinal cord tumor is a differential diagnosis in children who develop a tiptoe gait after having previously walked normally. Loss or instability of sphincter function is an important feature in their history.

22.5 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Table 22.1.

Table 22.1 Establishing the diagnosis of bilateral symmetric toe walking

History				
History of prematurity and/or global developmental delay	History of being a floppy infant may be present	Nothing contributory in history	Speech delay	Nothing contributory in history
No family history	Family history may be present	No family history	No family history	Family history may be present
Delayed walking	Walking may be delayed if infant was floppy	Walking not delayed	Walking not delayed	Walking not delayed
Physical examination				
Muscle tone increased	Muscle tone decreased	Muscle tone normal	Muscle tone normal	Muscle tone normal
Gastrocsoleus may be of normal length or the gastrocnemius may be short	Gastrocnemius and soleus short	Gastrocnemius and soleus short	Gastrocsoleus of normal length	Gastrocsoleus of normal length
Spasticity of muscles	Proximal muscle weakness present	No proximal muscle weakness	No proximal muscle weakness	No proximal muscle weakness
			Features of autism or dyspraxia	
Working diagnosis: Cerebral palsy	Working diagnosis: Muscle disease	Working diagnosis: Short gastrocsoleus	Working diagnosis: Toe walking associated with autism/dyspraxia	Working diagnosis: Idiopathic toe walker
Investigations				
Estimation of muscle enzymes not indicated	Muscle enzymes CK and dystrophin elevated Abnormalities in electromyography	Normal muscle enzymes	Normal muscle enzymes	Normal muscle enzymes
Diagnosis				
Cerebral palsy	Muscular dystrophy/ myopathy	Short gastrocsoleus	Toe walking associated with autism	Idiopathic toe walker

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The Flatfoot

23

James Robb

23.1 Introduction

There is no universally accepted definition of flatfoot (Mosca 2010; Dare and Dodwell 2014), but it can be considered as a foot form where there is a larger than normal plantar contact area with the ground because the medial longitudinal arch is depressed. Hindfoot valgus and abduction of the forefoot are also integral deformities of a flatfoot, apart from loss of the medial longitudinal arch. The term "planovalgus" is thus a more descriptive term for flatfoot.

Flatfeet may be physiologic or pathologic, resulting from failure of the normal ligaments and muscles that support the arch.

Natural Evolution of Foot Form in Children

Infants and toddlers have a prominent fat pad on the medial side of the foot which can obscure the bony contour of the medial arch, thus giving the appearance of a flatfoot. Toddlers also have a wide base of support when beginning to walk which also gives the appearance of a flatfoot. The medial arch appears between the ages of 2 and 3 years, and the child may still appear to have flatfeet because of physiologic joint laxity or because of physiologic knock-knees between 3 and 4 years of age. There is spontaneous development of the medial longitudinal arch of the foot in the first decade (Mosca 2010).

When faced with a child with flatfoot, the following questions need to be answered:

23.2 Questions to Establish a Diagnosis

- **What is the age of the child?**
- **Is the flatfoot flexible or rigid?**
- **Is the flatfoot painful or not?**
- **Is the flatfoot unilateral or bilateral?**
- **Is there any weakness or spasticity in the muscles of the calf and leg?**
- **Are the ankle plantar flexors short?**
- **Is there generalized hypermobility of joints?**
- **Is the child obese?**
- **Does the child have genu valgum or external tibial torsion?**

What is the age of the child?

From the foregoing remarks, it is clear that in infants and toddlers, there may be a risk of overdiagnosing flatfoot.

Is the flatfoot flexible or rigid?

This is perhaps the most important question to be answered. In the vast majority of instances, a flexible flatfoot is asymptomatic and warrants no treatment. On the other hand, rigid flatfeet often require treatment.

Is the flatfoot painful or not?

Pain is far more common in rigid flatfeet than in flexible flatfeet and the presence of pain often dictates the need for intervention.

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_23

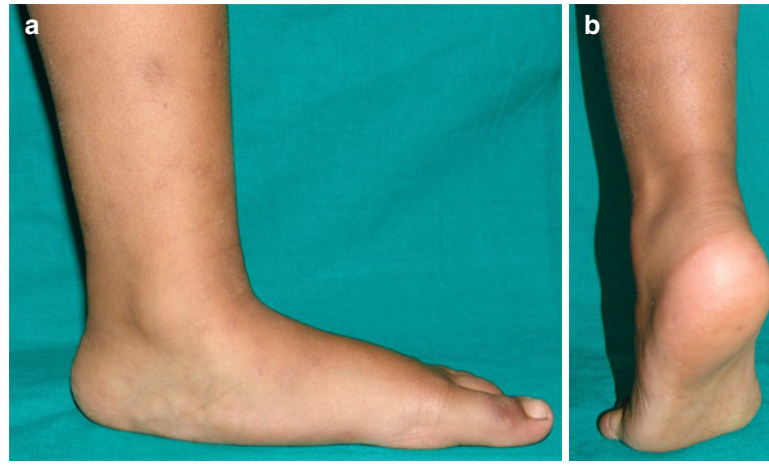

Fig. 23.1 Apparent flattening of the foot (**a**). The medial longitudinal arch appears when the patient stands on tiptoe (**b**)

Is the flatfoot unilateral or bilateral?
Unilateral flatfoot suggests pathology.

Is there any weakness or spasticity in the muscles of the calf and leg?
Weakness of muscles that support the arch of the foot (especially the tibialis posterior) can result in flatfoot. Similarly spasticity of the gastrocsoleus and the peronei can cause flatfoot.

Is the gastrocnemius or soleus or both short?
Shortening of either or both of these two plantar flexors may be seen in some children with flatfeet.

Is there generalized hypermobility of joints?
Generalized hypermobility of joints is associated with flatfeet as the lax ligaments of the foot yield on weight bearing.

Is the child obese?
The arch is more liable to collapse in obese children.

Does the child have genu valgum or external tibial torsion?
Flatfeet appear to be more evident in children with genu valgum and external tibial torsion.

23.3 Physical Examination

23.3.1 Look

Observe the gait and check if the child's gait pattern is appropriate for their age. Examine the child's shoes if they have them and, if not, observe the pattern of plantar calluses on the child's feet. Look for asymmetry. Check lower limb alignment in the coronal, sagittal, and transverse planes; specifically look for knock-knees and excessive external tibial torsion.

Ask the child to stand on tiptoe. In children with flexible flatfoot, the arch will appear and the calcaneus will go into varus when the child stands on tiptoe (Fig. 23.1).

23.3.2 Feel

Palpate the hindfoot and forefoot and ensure that there is no point of tenderness.

23.3.3 Move

Assess motion at the ankle, subtalar, and midtarsal joints to exclude a fixed deformity. Note if passive movement of the subtalar and midtarsal joints causes pain.

23.3.4 Special Tests

Jack's Test
In the younger child the great toe extension (Jack's) test is useful. The great toe is passively dorsiflexed; in flexible flatfoot the medial arch

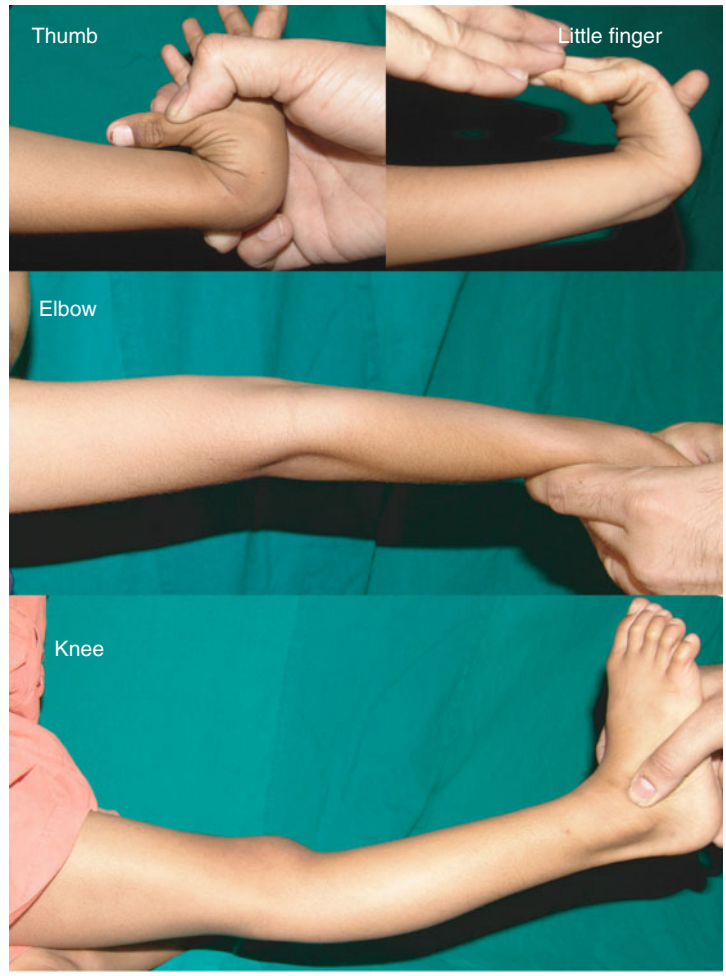

Fig. 23.2 Beighton score for hypermobile joints. Score 1 for each thumb if it can be made to touch the volar aspect of the forearm. Score 1 for each little finger if it can be made to hyperextend till it is parallel to the dorsum of the forearm. Score 1 for each elbow that hyperextends beyond neural. Score 1 for each knee that hyperextends beyond neutral. Add 1 score if the child can place both hands on the ground while standing with knees extended. A total of 4 or more out of 9 is regarded as generalised hypermobility

should rise because of the windlass action of the plantar fascia (see also Sect. 22.3).

Silferskiöld's Test

The test is to evaluate the length of the gastrocnemius and the soleus if there is limitation of ankle dorsiflexion (see Sect. 22.3, Fig. 22.2a, b).

Testing for Generalized Ligament Laxity

The Beighton score (Beighton et al. 1973) is often used to evaluate joint laxity and consists of nine points. A score greater than four suggests joint laxity. The test comprises one point for each elbow that hyperextends, one point for each thumb that can touch the anterior aspect of the forearm, one point for each little finger that can be hyperextended beyond 90°, one point for each knee that hyperextends, and one point for the ability to touch the ground with the palms of both hands while maintaining knee extension (Fig. 23.2).

23.3.5 Investigations

Radiography

Radiographs of typical flexible flatfeet are not necessary but may be indicated where there is unexplained foot pain and a flexible flatfoot. In this circumstance a radiograph should be taken weight bearing, and the lateral radiograph will typically show plantar flexion of the calcaneus and talus. Measurements of angles in young children's feet are not particularly reliable

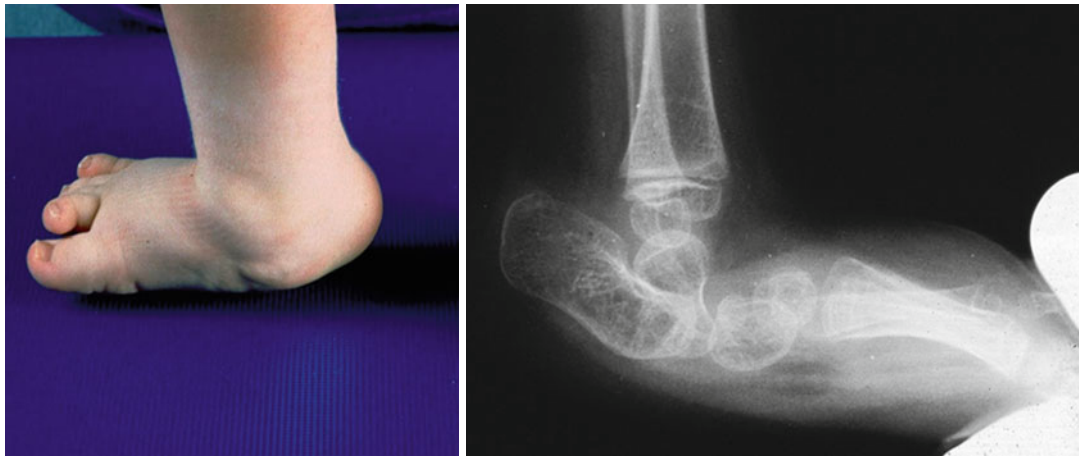

Fig. 23.3 Congenital vertical talus

because ossification is incomplete. Oblique views are needed if the foot is a rigid flatfoot, and a calcaneonavicular coalition in the older child is best seen from this view (see Chap. 31). An AP radiograph of the ankle will show true ankle varus or valgus which is helpful in determining causes of pes valgus secondary to an ankle deformity.

23.4 Differential Diagnosis

23.4.1 Physiologic Flatfoot

Physiologic Flatfoot with Normal Calf Muscle Length

The children have normal feet and normal calf muscle lengths with flexible, painless flatfeet.

Physiologic Flatfoot with Short Gastrocsoleus

The children have normal feet but have a short soleus or gastrocnemius or both. In this type of flatfoot, the calcaneus will have a tendency to go into escape valgus in stance to offset the short calf muscles, but there will be physiologic varus of the calcaneus when the child stands on tiptoe.

23.4.2 Pathologic Flatfoot

Congenital Vertical Talus

In reality the foot is not flat; the arch is reversed. Clinically the forefoot is dorsiflexed and abducted, whereas the hindfoot is in fixed equinus and valgus (Fig. 23.3). There is an irreducible dorsal dislocation of the navicular on the talus, which is vertical, and a subluxation of the talocalcaneal joint. These features will show on a radiograph but the navicular does not ossify before about the age of 4 years which may make interpretation more difficult in the younger child (Thometz et al. 2010). These bony deformities are accompanied by soft tissue abnormalities; peroneus longus and tibialis posterior then act as dorsiflexors, and there is fixed equinus due to a short heel cord and tight ankle capsule.

Congenital vertical talus should be distinguished from paralytic flatfoot, flatfoot in diplegic CP, and an overcorrected (rocker bottom) in clubfoot.

Congenital Oblique Talus

The foot is flat and a lateral radiograph of the foot will show that the talus is severely plantar flexed as in a vertical talus. The deformity is, however,

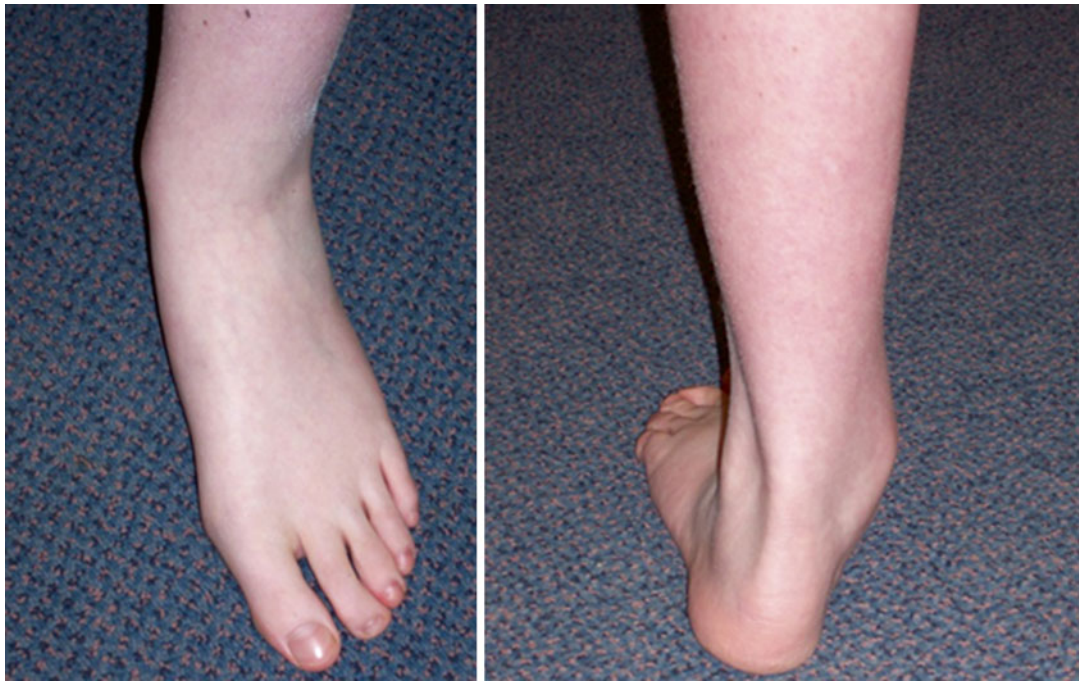

Fig. 23.4 Pes valgus in spastic diplegia

less rigid than a vertical talus, and on plantar flexing the foot, the normal tarsal relationships are restored (see Fig. 6.3).

Paralytic Flatfoot

The commonest cause is spastic diplegia where shortening of gastrocnemius, soleus, or both or increased tone in the triceps surae results in escape valgus of the heel when the child stands (Fig. 23.4). If uncorrected, structural changes occur, and the child typically develops a short lateral column of the foot eventually. Spasticity of the peronei can aggravate the deformity.

Lower motor neuron lesions, for example, spina bifida (Swaroop and Dias 2011), can also result in ankle and foot instability and a resultant flatfoot (Fig. 23.5). Foot instability and apparent or true flattening may also be seen in myopathies. True ankle valgus is a feature of spina bifida (Fig. 23.6) and should be distinguished from foot instability causing pes valgus, although both features may be present.

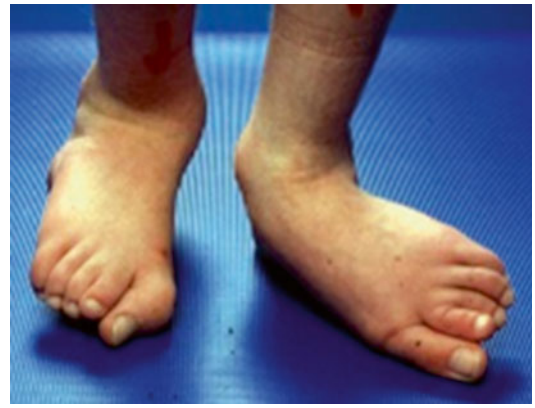

Fig. 23.5 Foot instability in spina bifida

Isolated paralysis of the tibialis posterior can occur in polio, and this results in a paralytic flatfoot.

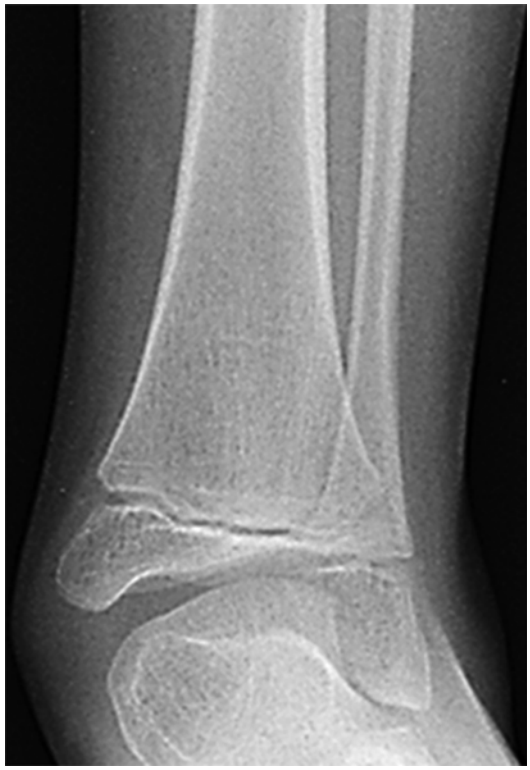

Fig. 23.6 Ankle joint valgus in spina bifida

Juvenile Idiopathic Arthritis

A common site for JIA is at the subtalar joint which becomes stiff and a rigid hindfoot and flatfoot can ensue.

Trauma and Sepsis

Damage to joints in the foot from the sequelae of trauma or sepsis can result in a rigid flatfoot.

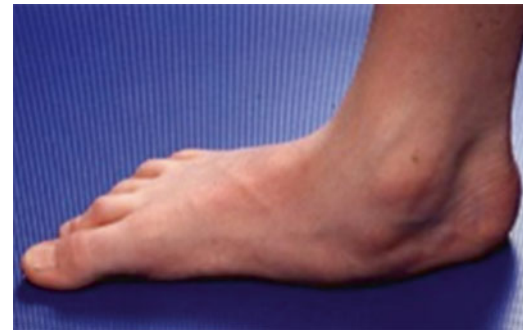

Fig. 23.7 Post laceration of tibialis posterior tendon producing a reversal of the medial longitudinal arch

Tarsal Coalition

This is not generally seen in the preschool child but is one cause of a pathologic flatfoot in children of school age. See Chaps. 31 and 38.

23.4.3 Less Common Causes of Flatfoot

Laceration of Tibialis Posterior

This results in loss of one of the dynamic stabilizers of the medial arch (Fig. 23.7).

Overcorrected Clubfoot

Defunctioning of the tibialis posterior may contribute to flatfoot following radical surgery for clubfoot.

23.5 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Table 23.1.

Table 23.1 Establishing the diagnosis of the cause of flatfoot

History				
Nothing contributory	Nothing contributory	History of pain in hindfoot/history of previous trauma or sepsis/or no history of pain	Developmental delay	History of sharp trauma or paralytic episode
Physical examination				
Ligament laxity often present	Ligament laxity may be present	Ligament laxity not a feature	Ligament laxity not a feature	Ligament laxity not a feature
On tiptoeing: Arch restored Heel goes into varus	On tiptoeing: Arch restored Heel goes into varus	On tiptoeing: Arch not restored Heel does not go into varus	On tiptoeing: Arch not restored Heel does not go into varus	On tiptoeing: Arch may be restored Heel does not go into varus
Jack's test: Arch restored	Jack's test: Arch restored	Jack's test: Arch not restored	Jack's test: Arch not restored	Jack's test: Arch restored
Length of gastrocsoleus normal	Gastrocsoleus short	Length of gastrocsoleus normal or short	Gastrocnemius short	Length of gastrocsoleus normal
Tibialis posterior function normal	Tibialis posterior function normal	Tibialis posterior function normal	Tibialis posterior function difficult to determine	Tibialis posterior paralyzed
Muscle tone normal	Muscle tone normal	Muscle tone normal	Muscle tone increased	Muscle tone may be decreased
Movement of subtalar joint exaggerated	Movement of subtalar joint exaggerated	Movement of subtalar joint reduced	Movement of subtalar joint reduced	Movement of subtalar joint reduced
			Peronei may be spastic	
			Foot pronated with a break in the midtarsal joint	
Diagnosis				
Physiologic flexible flatfoot	Physiologic flexible flatfoot with tight gastrocsoleus	Rigid flatfoot as in tarsal coalition, JIA, sequelae of trauma, and infection involving the subtalar joint	Spastic flatfoot	Flatfoot due to tibialis posterior dysfunction (LMN paralysis or tendon injury)

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The Child with a Painless Limp

24

James Robb

24.1 Introduction

24.1.1 Gait Maturation

A limp in a child is a deviation from the gait pattern expected at the particular stage in the child's development. Children start walking around 12–14 months of age and do not acquire an adult walking pattern until after the age of 4 years (Sutherland et al. 1980). Toddlers have a broad base of support, a faster cadence (steps per minute), and a slower velocity than an adult and do not have reciprocal arm movements. Their legs are externally rotated, knees remain flexed in stance, and heel strike does not usually occur until around 15–18 months of age when reciprocal arm movements begin to appear as well. Running and the ability to change direction smoothly occur after the age of 2 years, and by the age of three, the child's base of support is approaching that of an adult, and most will have reciprocal arm movements. It is important that these immature patterns of gait are not misinterpreted as being abnormal.

Electronic supplementary material The online version of this chapter (doi:10.1007/978-81-322-2392-4_24) contains supplementary material, which is available to authorized users.

24.1.2 Prerequisites for Normal Gait

To walk normally the following prerequisites must be fulfilled:

1. The skeleton must be strong enough to support the weight of the body.
2. Both lower limbs must be of equal length.
3. The muscles of the lower limbs must be sufficiently strong to provide both stance phase stability and adequate mobility in the swing phase.
4. The joints of the lower limb must be supple enough to permit the requisite movement needed for normal forward progression.
5. The joints of the lower limb should not be deformed.
6. The joints of the lower limb should be stable.

It follows that abnormalities of the skeleton (fragility, unequal length of the limbs), the muscles (weakness, spasticity, contracture), or the joints (stiffness, deformity, and instability) can all result in a limp (Fischer and Beattie 1999; Bleck and Robb 2010). More than one of these abnormalities may contribute to a limp, and it is important to identify all potential causes (Table 24.1).

Pain arising from the bones, muscles, or joints can also cause a limp. The causes of a painful limp are listed in Chap. 31.

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_24

Table 24.1 Causes for limp in different situations

Condition	Factors contributing to limp
DDH	Shortening + joint instability
Tom Smith's arthritis (destruction of the femoral head following neonatal septic arthritis)	Shortening + joint instability
Coxa vara	Shortening + deformity
Cerebral palsy with crouch gait	Muscle spasticity + muscle contracture + muscle weakness + joint deformity
Post-polio residual paralysis	Shortening + deformity + muscle weakness + joint deformity

24.2 Questions to Establish a Diagnosis

- **Is the limp unilateral or bilateral?**
- **When was the limp first noted?**
- **Is there a suggestion of progressive worsening of the limp?**
- **Is the tone of the muscles of the lower limbs normal or altered?**

Is the limp unilateral or bilateral?

Bilateral symmetrical limp narrows down the possible diagnoses to a small number of conditions, most of which are neurological conditions such as diplegia in cerebral palsy, hereditary spastic paraplegia, hereditary motor-sensory neuropathy (HMSN), spina bifida, traumatic brain injury, and muscular dystrophy. Bilateral symmetrical limp may also be seen in bilateral DDH, bilateral coxa vara, and other symmetric congenital or developmental anomalies.

When was the limp first observed?

A limp associated with congenital anomalies will be noted as soon as the child starts to walk. Limp in a child with a delay in walking is characteristically seen in cerebral palsy.

Is there a suggestion of progressive worsening or spontaneous resolution of the limp?

Progressive deterioration of the gait pattern is seen in progressive paralytic disorders such as muscular dystrophy. Deterioration may also occur with progressive growth disturbance following aphyseal injury. Resolution of the limp may occur with remodeling of a malunited fracture.

Is the tone of the muscles of the lower limbs normal or altered?

Examination of muscle tone is useful in separating conditions that cause a child to limp.

24.3 Physical Examination

24.3.1 Look: Observe the Gait

Have the child undressed sufficiently to be able to view their spine and lower limbs and observe their gait in the sagittal and coronal planes.

General Observations

(a) Is their gait expected for their age? If not, consider developmental delay.
(b) Which side seems abnormal?
(c) Is arm movement reciprocal and symmetrical? Then consider each limb separately.

Sagittal Plane

(a) Which part of the foot makes contact with the ground first?
(b) Does the whole of the foot make contact with the ground in stance?
(c) Is the timing of heel rise normal?
(d) Does the knee extend fully in stance?
(e) Is there excessive knee flexion in stance?
(f) Is there adequate foot clearance in swing?
(g) What is the position of the foot in terminal swing?

Coronal and Transverse Planes

(a) What is the foot progression angle relative to the plane of progression?
(b) Is there circumduction?
(c) Is there excessive trunk sway to one side?

Look for specific gait patterns listed in Table 24.2.

24.3.2 Feel

Palpate any joint that appears deformed. Palpate the bones and joints to ensure that there is no site of tenderness.

Table 24.2 Characteristic features of some common types of limp

Type of limp	Characteristic features	Conditions where it occurs
Short-limb gait	In mild degrees of discrepancy, compensatory mechanisms may help in "equalizing" limb lengths by effectively "shortening" the longer limb by flexing the knee or by effectively "lengthening" the shorter limb by plantar flexing the ankle and standing on the toes (see Table 24.3)	Shortening of one lower limb due to any cause
Trendelenburg limp	During the stance phase, the opposite hemi-pelvis dips Compensatory mechanism: Leaning over the affected hip (the trunk and shoulder shift toward the affected side) Bilateral Trendelenburg limp will result in a waddling gait	Loss of the fulcrum of the abductor lever (2 Ds, i.e., *D*estroyed femoral head or *D*islocated femoral head) Abductor lever arm ineffective (2 Bs, i.e., *B*ending of neck, coxa vara, or *B*reaking of the neck, femoral neck fracture) Abductor force inadequate (1 P, i.e., *P*aralysis of the gluteus medius and minimus)
Stiff-knee gait (Video 24.1)	Decreased sagittal plane motion during the swing phase Difficulty in getting clearance of the foot during the swing phase Compensatory mechanisms: Circumduction of stiff limb OR vaulting of opposite limb	Quadriceps contracture Extra-articular or intra-articular ankylosis of the knee in extension Cerebral palsy with co-spasticity of the rectus femoris and the hamstrings
Hand-to-thigh gait (Video 24.2)	During the swing phase of gait, the child uses the hand to stabilize the knee by pushing the thigh backward	Quadriceps paralysis (polio)
High-stepping gait (Video 24.3)	Excessive hip and knee flexion in the swing phase of gait to facilitate foot clearance The gait sequence is a toe-heel sequence rather than the normal heel-toe sequence	Paralysis of the ankle dorsiflexors
Equinus gait (Videos 24.4 and 24.5)	The toe is in contact with the ground throughout the stance phase. At no time does the heel rest on the ground	Equinus contracture or spasticity of the gastrocsoleus muscle Habitual toe walking (to begin with there may be no contracture of the gastrocsoleus muscle)

24.3.3 Move

Move the hips, knees, and ankles through their ranges of motion and confirm the joints are not stiff or painful.

24.3.4 Measure

Measure the limb lengths, including the lengths of the thigh and leg segments.

24.3.5 Additional Tests

Ask the child to run as this may accentuate the limp.

Ask the child to get up off the floor; this test is useful to detect proximal muscle weakness.

Ask the child to pick up an object off the floor – this may highlight spinal pathology if the child flexes at the knees rather than at the spine.

If the child is old enough to cooperate, ask the child to walk on tiptoe, walk on the heels, and stand on one leg. These tests will give a rough idea of muscle strength and can be done without having to lie the child down on the examination couch which can be stressful for young children.

Perform a standard orthopedic examination of the child's limbs and spine.

Perform a neurological examination including muscle strength, tendon reflexes, comparison of thigh and calf muscle bulks, and sensations. Pay

Table 24.3 Compensatory gait mechanisms to cope with limb length inequality

	Compensatory mechanisms	
	The longer limb	The shorter limb
To facilitate swing phase clearance of the longer limb	Circumduction of the limb	Vaulting (premature plantar flexion)
To equalize limb length in double support of the stance phase	Flexing the knee	Plantar flexion of the ankle and standing on tiptoe

specific attention to the tone of the muscles of the lower limb.

Perform the Trendelenburg test on each side.

24.4 Investigations to Confirm the Diagnosis

Radiography

Plain radiographs are usually sufficient to identify structural causes for a limp such as limb length discrepancy and developmental dysplasia of the hip.

Ultrasound

Ultrasound examination of the hip is useful if transient synovitis is suspected (Deanehan et al. 2014).

Serum creatine kinase estimation and genetic analysis should be considered if muscular dystrophy is suspected; additional tests are seldom indicated in a child with a painless limp.

24.5 Differential Diagnosis

24.5.1 Limb Length Inequality

While severe degrees of limb length inequality can be easily appreciated on observing the gait of the child, the more subtle degrees of discrepancies in limb length may be masked by compensatory mechanisms in gait (Table 24.3). These compensatory mechanisms may be noted either in the long or the short limb.

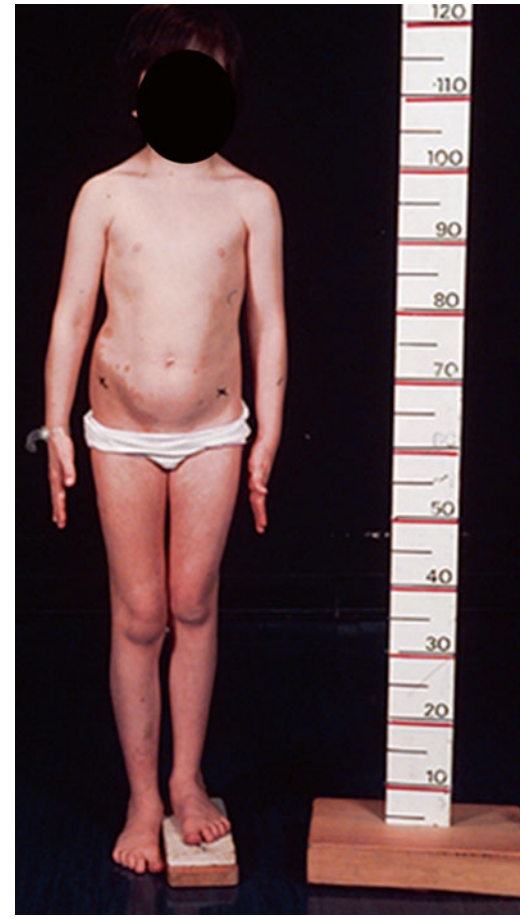

Fig. 24.1 Limb length discrepancy. There is shortening in both the thigh and the leg

Causes of limb length inequality that has been present from birth include congenital short femur, congenital coxa vara, fibular and tibial hemimelia, congenital posteromedial bowing of the tibia, and hemihypertrophy. Limb length inequality develops later if it is secondary to physeal damage following infection or trauma.

The history is relevant to exclude a trauma or infection. Physical examination should include a comparison of the lengths of the thigh and leg segments using blocks to evaluate the overall limb length discrepancy (Fig. 24.1). Long-leg films or a CT scanogram for leg length will confirm the diagnosis.

True limb length inequality must be distinguished from apparent discrepancy. An apparent limb length discrepancy results from pelvic

Table 24.4 Muscle tone and the pattern of involvement in children with a limp due to muscle weakness

Increased tone (upper motor neuron lesion)		Decreased tone (lower motor neuron lesion)		Normal or decreased tone
Bilateral involvement (symmetric/asymmetric)	Unilateral involvement	Bilateral involvement (symmetric/asymmetric)	Unilateral involvement	Bilateral involvement (symmetric)
Diplegic cerebral palsy (symmetric) Hereditary spastic paraplegia (symmetric) Traumatic brain injury (symmetric or asymmetric)	Hemiplegic cerebral palsy Traumatic brain injury (hemiplegic)	Polio (asymmetric) Spina bifida (often symmetric) Spinal tumor (often symmetric) Hereditary motor-sensory neuropathy (symmetric)	Polio Spinal tumor Spinal dysraphism	Muscular dystrophy

obliquity which develops if there is a fixed adduction or abduction deformity of the hip or if there is structural lumbar scoliosis. A fixed abduction deformity will result in pelvic obliquity with apparent lengthening of the limb on the side of the hip pathology, while a fixed adduction deformity will result in pelvic obliquity with apparent shortening of the limb on the side of hip pathology. In a child with lumbar scoliosis, apparent lengthening of the limb will be present on the side of the convexity of the curve. Visual observation of gait and orthopedic examination of the child's back and hips should help to distinguish between infrapelvic and suprapelvic causes for pelvic obliquity.

24.5.2 Muscle Weakness

Diagnosis of the underlying condition is often straightforward in many instances when the typical features are evident, but when the weakness is mild, careful examination of the muscle tone and noting the pattern of involvement may help in making a correct diagnosis (Table 24.4).

Upper Motor Neuron Lesion

The commonest cause is hemiplegic or diplegic cerebral palsy. A combination of spasticity, weakness, incoordination, co-spasticity, and deformities may all contribute to the limp.

Lower Motor Neuron Lesion

Polio and spina bifida result in weakness that can contribute to a limp. Muscle paralysis in polio may affect one or both lower limbs; bilateral involvement is typically asymmetric. Paralytic deformities of joints and limb length inequality, if present, will contribute to the limp.

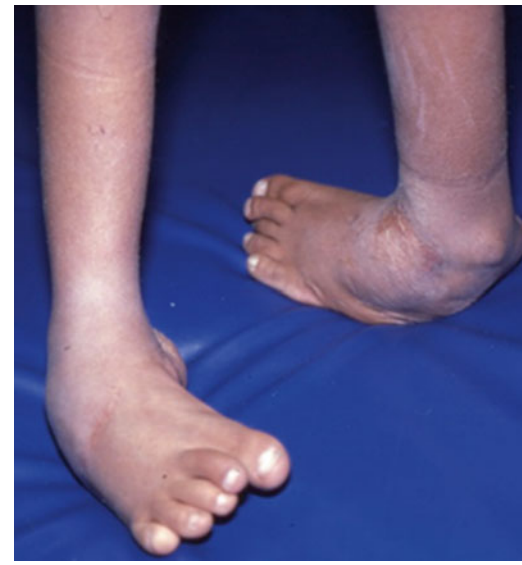

Fig. 24.2 Charcot-Marie-Tooth disease. There is marked wasting of the calf musculature, loss of peroneal muscle strength, and a resulting varus posture of the foot on standing

Motor and Sensory Neuropathy

The first manifestation of hereditary motor-sensory neuropathy (HMSN), which is often familial, may be a limp (Fig. 24.2). For example, in Charcot-Marie-Tooth disease, the child may first present with a cavus foot and on examination

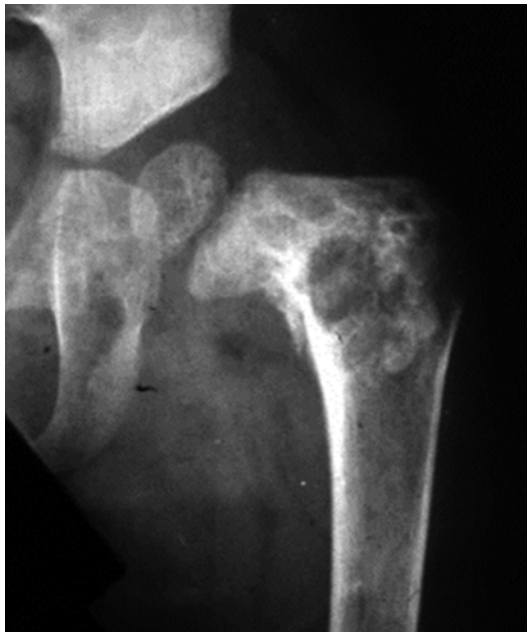

Fig. 24.3 Acquired coxa vara in fibrous dysplasia. There is an undisplaced fracture of the femoral neck

will have peroneal muscle weakness and loss of vibration sense at the ankles.

Muscle Disease
Alterations in gait and limping are seen in muscle disease. It is important to take a family history as, in general terms, dystrophies and myopathies are usually autosomal dominant or sex linked and atrophies are generally autosomal recessive. The commonest muscular dystrophy is Duchenne's, and boys typically walk at a normal stage in development and then begin to lose walking ability (see Chap. 22, Fig. 22.3). In early stages the boy may begin to walk in equinus and appear to limp. Serum creatine kinase is elevated many times higher than normal, and the diagnosis can be confirmed on genetic testing.

24.5.3 Joint Stiffness

Though stiffness of the hip, knee, ankle, or foot will alter the gait, limitation of hip motion may not always produce an obvious limp. On the other hand, knee stiffness always results in a demonstrable limp. A stiff knee produces a functional, rather than a structural, leg length discrepancy; the typical compensations are circumduction of the limb with the stiff knee or vaulting on the normal side. A stiff-knee gait is seen if there is congenital or acquired quadriceps contracture, co-spasticity of the rectus femoris and the hamstrings in cerebral palsy, and intra-articular stiffness in juvenile idiopathic arthritis (JIA) or following septic arthritis and extra-articular or intra-articular ankylosis of the knee.

24.5.4 Joint Deformity

Deformities of the hip, knee, ankle, and foot will all result in a limp.

Coxa Vara
The child may present with a Trendelenburg gait pattern resulting from an abnormal abductor lever arm. The deformity of the femoral neck may be congenital or secondary to disease in the femoral neck (Fig. 24.3). If the involvement is unilateral, there will be some degree of shortening as well.

Muscle Contractures
Muscle contracture is the commonest cause of a joint deformity. Trauma, infection, ischemia, intramuscular injections, intramuscular bleeds, and intramuscular hemangiomas can all result in fibrosis of the muscles which in turn can cause joint deformities. For example, contractures of muscles of the calf following compartment syndrome of the superficial posterior compartment will result in an equinus deformity and an equinus gait.

24.5.5 Joint Instability

Hip

Delayed Presentation of DDH
This is the commonest cause of a painless limp in a toddler. There may be a positive family history, and risk factors of breech delivery and female gender are relevant. The child with unilateral dysplasia will typically walk with a short leg or

Trendelenburg positive pattern and on physical examination will have a positive Galeazzi sign and restricted hip abduction. Bilateral DDH is less obvious to diagnose clinically; the child walks with a bilateral Trendelenburg lurch (waddling gait) and exaggerated lumbar lordosis. High-riding greater trochanters and some foreshortening of the thigh segment may be clinical clues. Pelvic radiography will confirm the diagnosis (Fig. 24.4).

Hip Instability in Down Syndrome

Children with Down syndrome have joint laxity and can develop a silent subluxation of a hip.

Hip Instability in Congenital Insensitivity to Pain

The painless Charcot hip joint becomes disorganized and then grossly unstable often with multidirectional instability (Fig. 24.5).

Knee

A varus thrust may be seen in infantile Blount's disease (Video 24.6). Knee instability is occasionally seen in conditions with gross ligament

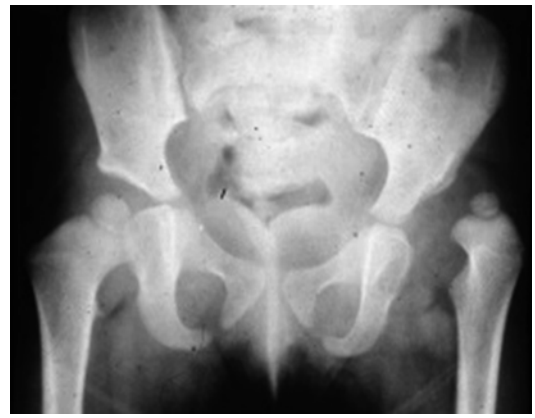

Fig. 24.4 Delayed presentation of DDH

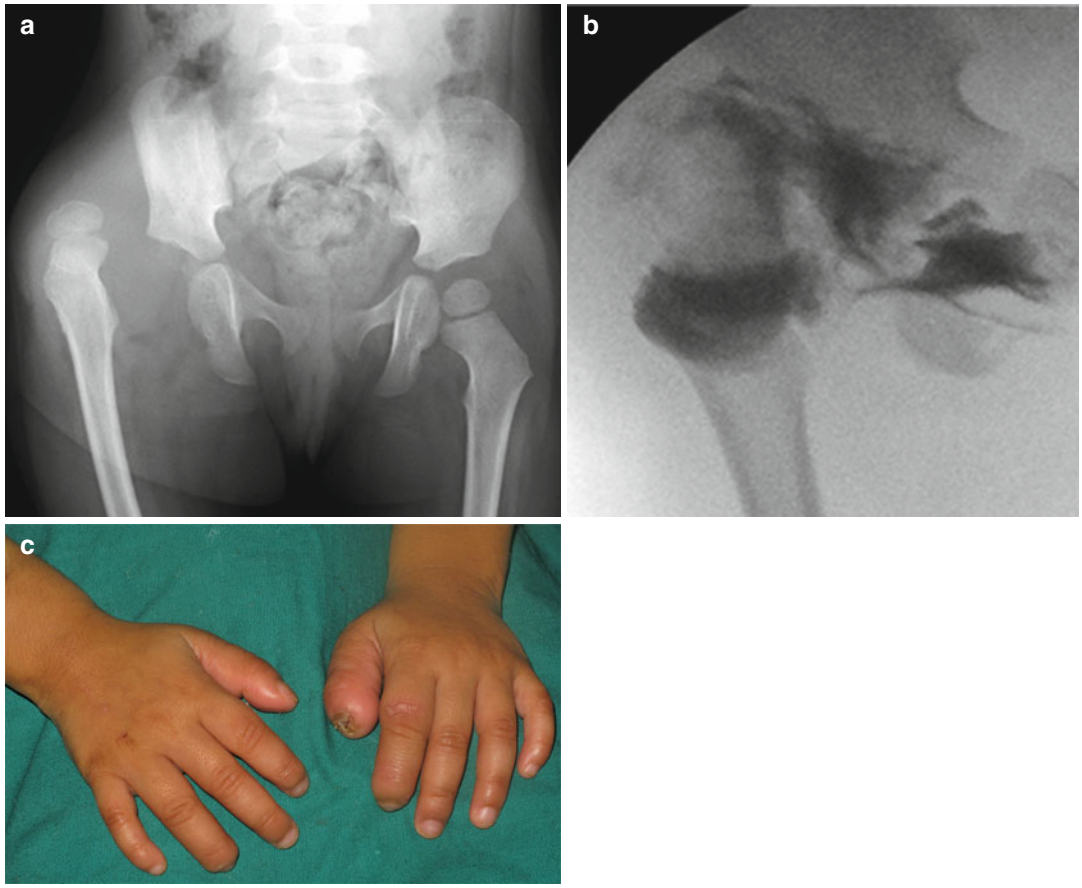

Fig. 24.5 Hip dislocation in a child with congenital insensitivity to pain (**a**); the arthrogram shows an extremely distended joint capsule (**b**). Trophic changes on the fingertips are seen (**c**)

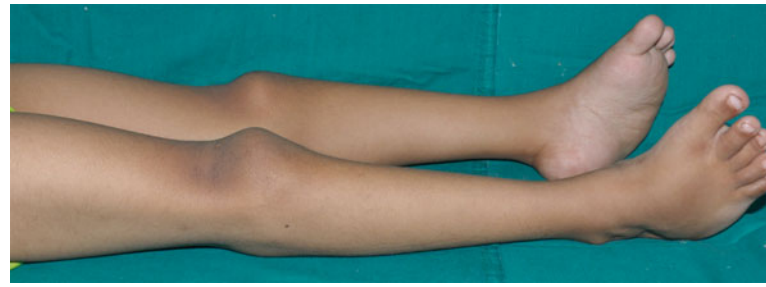

Fig. 24.6 Knee subluxation in a boy with Larsen syndrome

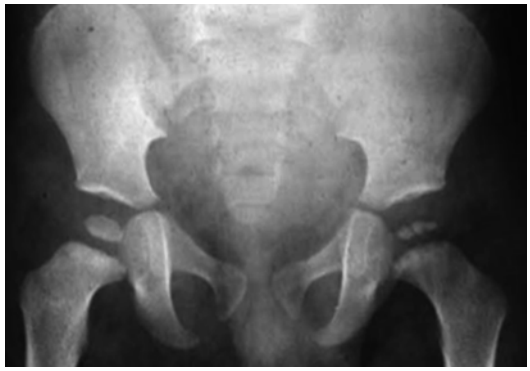

Fig. 24.7 Perthes' disease of the left hip

laxity such as Larsen syndrome (Fig. 24.6, Video 24.7). The knee will become unstable once a Charcot joint develops in congenital insensitivity to pain. A painless limp develops in each of these situations.

24.5.6 Less Common Conditions

Perthes' Disease (Fig. 24.7) **and Transient Synovitis** (Fig. 24.8)
Though these conditions usually present with pain, they may occasionally present as a painless limp in the very early stages.

Malignancies
Leukemia is the commonest malignancy of childhood and should be excluded if the child has an unexplained limp, malaise, and anemia. Symptoms from a spinal cord tumor are often insidious but may first present as a neuropathy of the limbs rather than spinal pain.

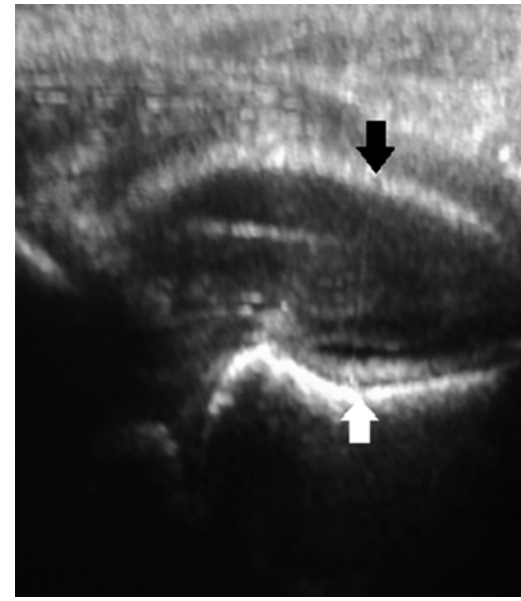

Fig. 24.8 Ultrasound of the hip showing a large effusion. The child was well and had a transient synovitis. The *white arrow* indicates the anterior aspect of the metaphysis, and the *black arrow* the anterior margin of the distended hip capsule

Juvenile Idiopathic Arthritis
Involvement of joints in the lower limb may produce, in the early stages, a painless effusion and/or stiffness and a limp. A young child may complain of fatigability rather than pain in the early stages.

Discoid Lateral Meniscus
In gait there may be loss of full knee extension, "locking" or giving way of the knee. On examination there may be fullness on the lateral side of the joint, and a "clunk" may be demonstrable

during flexion and extension. There may be widening on the lateral side of the joint on plain radiography, and the diagnosis can be confirmed on MRI (see Chap. 37).

24.5.7 Rare Conditions

Aplasia of Muscles

Congenital aplasia of muscles of the lower limb manifests as a painless limp; aplasia of several muscles of the lower limb has been reported in the literature (Varghese and Joseph 2007; Peterson and Currarino 1981; Tibrewal et al. 2014; Flynn et al. 2007; Uzel et al. 2007).

24.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis in children with different gait patterns is shown in Tables 24.5, 24.6, and 24.7.

Key Points

- A healthy child of less than 4 years of age presenting with a painless limp should be assumed to have DDH, unless proven otherwise.
- Consider malignancy in a child with unexplained limp.

Table 24.5 Establishing the diagnosis of the cause of equinus gait

History				
History of clubfoot deformity at birth which was treated	Apparently normal at birth	May give history suggestive of difficult labor/delayed cry after birth/ prematurity/twin birth	Apparently normal at birth	Normal at birth
Limp ever since the child started to walk	Limp ever since the child started to walk	Delayed walking Limp ever since the child started to walk	Walked normally to begin with. Limp appeared later	Walked normally to begin with. Limp appeared after febrile paralytic episode
No history of progression	No history of progression	No history of progression	History of progression present	No progression of limp
Physical examination				
Unilateral/bilateral involvement	Bilateral symmetric involvement	Unilateral/bilateral involvement	Bilateral symmetric involvement	Unilateral/bilateral asymmetric involvement
Normal muscle tone	Normal muscle tone	Increased muscle tone	Decreased muscle tone	Decreased muscle tone
Normal muscle strength (no weakness of ankle dorsiflexors)	Normal muscle strength (no weakness of ankle dorsiflexors)	Selective motor control may not be present to test active dorsiflexor power	Weak dorsiflexor power	Weak or paralyzed ankle dorsiflexors
Fixed equinus contracture involving the gastrocnemius and the soleus (Silferskiöld test negative)	May not be a contracture of the gastrocsoleus initially (may develop later)	Spasticity of the gastrocsoleus If contracture is present, usually the gastrocnemius alone is contracted (Silferskiöld test positive)	Fixed equinus contracture involving the gastrocnemius and the soleus (Silferskiöld test negative)	Fixed equinus contracture involving the gastrocnemius and the soleus (Silferskiöld test negative)
Working diagnosis: Residual or recurrent equinus of clubfoot	Working diagnosis: Habitual toe walker	Working diagnosis: Cerebral palsy (hemiplegic or diplegic)	Working diagnosis: Muscular dystrophy	Working diagnosis: Post-polio residual paralysis
Investigations				
None indicated	None indicated	Neurological evaluation	Serum creatine kinase estimation Neurological evaluation Genetic analysis	None indicated
Diagnosis				
Residual or recurrent equinus of clubfoot	Habitual toe walker	Cerebral palsy	Muscular dystrophy	Post-polio residual paralysis

Table 24.6 Establishing the diagnosis of stiff-knee gait

History			
May have history of limitation of knee flexion since birth OR limitation of knee flexion may have developed later	May have history of total loss of movement at the knee since birth OR total loss of movement at the knee may have developed after infection or injury	No history of loss of motion of the knee	No history of loss of motion of the knee
Involvement may be unilateral or bilateral	Unilateral involvement	Unilateral or bilateral involvement	Unilateral involvement
		History of developmental delay often present	History of dislocation of the patella or a "click" in the knee and locking
Physical examination			
Knee fixed in extension	Knee fixed in extension or flexion	Knee not fixed in extension	Knee not fixed in extension
Very limited passive motion in the knee	Absolutely no passive or active flexion of the knee	Full range of passive motion	Some limitation of passive motion when child is conscious due to apprehension
Muscle tone normal	Muscle tone normal	Muscle tone increased Demonstrable spasticity of the rectus femoris and hamstrings	Muscle tone normal
Patella may be high	Patella may be fixed to the femur	Patella may be elevated if the knee is in flexion	Patella may mal-track
No pain on attempted movement of the knee	No pain on attempted movement of the knee	No pain on movement of the knee	Movement of the knee may cause pain Apprehension sign may be positive
Working diagnosis: Quadriceps contracture (congenital or acquired)	Working diagnosis: Ankylosis of the knee (congenital or acquired)	Working diagnosis: Co-spasticity of the rectus femoris and hamstrings in cerebral palsy	Working diagnosis: Stiff-knee gait due to apprehension (protective mechanism to avoid pain on flexing the knee)
Investigations			
Plain radiograph: To exclude underlying bony abnormality and to assess the shape of the femoral condyle Patella alta may be present	Plain radiograph: Will show intra-articular or extra-articular bone trabeculae crossing the joint	Plain radiograph: If the knee is in flexion, patella alta may be present	Plain radiograph: Skyline view may show lateral subluxation (patellar instability)/lateral joint space widened (discoid meniscus)
Diagnosis			
Quadriceps contracture (congenital or acquired)	Ankylosis of the knee (congenital or acquired)	Co-spasticity of the rectus femoris and hamstrings in cerebral palsy	Stiff-knee gait due to apprehension

Table 24.7 Establishing the diagnosis of the cause of unilateral Trendelenburg gait

History			
History of breech delivery or family history of hip disease may be present	History of febrile episode with decreased hip movement (may or may not have been treated by surgical drainage of hip)	No history of illness in infancy	History of swelling in the lumbosacral region at birth
Limp noted as soon as child started walking	Limp noted as soon as child started walking	Limp may be noted as soon as child started walking/limp noted later	Limp noted as soon as child started walking
Physical examination			
Shortening of the limb (supra-trochanteric)	Shortening of the limb (supra-trochanteric)	Shortening of the limb (supra-trochanteric)	Shortening of the limb (supra-trochanteric)
Loss of resistance in the femoral triangle	Loss of resistance in the femoral triangle	No loss of resistance in the femoral triangle	Loss of resistance in the femoral triangle
Femoral head palpable in gluteal region	Femoral head not palpable in the gluteal region	Femoral head palpable in femoral triangle	Femoral head palpable in the gluteal region
Limitation of hip abduction	Exaggeration of hip movements	Limited hip abduction and internal rotation and increased adduction and external rotation	Limitation of hip abduction
Normal power of hip abductors	Normal power of hip abductors	Normal power of hip abductors	Hip abductors paralyzed, hip flexors and knee extensors functioning at least at MRC Grade III
Working diagnosis: DDH	Working diagnosis: Destruction of the femoral head following septic arthritis in infancy	Working diagnosis: Coxa vara	Working diagnosis: Paralytic hip dislocation
Investigations			
Plain radiograph of the pelvis	Plain radiograph of the pelvis	Plain radiograph of the pelvis	Plain radiograph of the pelvis and spine
Diagnosis			
Developmental dysplasia of the hip	Destruction of the femoral head following septic arthritis in infancy	Coxa vara	Paralytic hip dislocation in spina bifida

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The Delayed Walker

25

James Robb

25.1 Introduction

Global developmental delay is a delay in two or more developmental domains that include cognition, speech and language, motor skills, personal skills, social skills, play, and activities of daily living. A specific developmental delay, e.g., motor, is when one area of development is affected (Silove et al. 2013).

When assessing a child with a delay in walking, it is necessary to be aware of the average ages at which the common motor milestones are achieved. Children walk on average between 12 and 14 months of age, and other relevant motor milestones are as follows:

Age	Milestone
6 months	Sits
9 months	Crawls
10 months	Stands
12–14 months	Walks

25.2 Questions to Establish a Diagnosis

- **Is there a history of consanguinity?**
- **Is there a family history of motor delay?**
- **Is there a history of maternal substance abuse?**
- **Was the child born prematurely?**
- **Was there a problem during labor?**
- **Was the child floppy at birth?**
- **Is there a history of neonatal sepsis?**
- **Were joint contractures evident at birth?**
- **Was a spinal deformity evident at birth?**
- **Did the child crawl normally?**
- **Is the child a boy with developmental delay and possibly learning difficulties?**
- **Does the child have stiff limbs?**
- **Is there a history of cranial or spinal trauma?**
- **Does the child have normal vision?**
- **Are the child's stature and body proportions normal?**
- **Does the child have the features of rickets?**

Is there a history of consanguinity?

It is important to rule out consanguinity as a potential cause as this can be associated with metabolic and genetic causes of developmental delay.

Is there a family history of motor delay?

In general terms, dystrophies and myopathies are autosomal dominant or sex linked, and atrophies are autosomal recessive (Bleck and Robb 2010). Infants and children with a genetically determined motor disorder tend to present with weakness and delayed motor skills, whereas those with an acquired motor disorder tend to present with progressing weakness and loss of function.

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Is there a history of maternal substance abuse?

The fetal alcohol syndrome may result in learning difficulties and motor delay.

Was the child born prematurely?

Cerebral palsy (CP), the commonest cause of childhood disability, is associated with prematurity. Very premature infants are at risk of having total body involvement CP, whereas those born later, e.g., at 32 weeks are at risk of having diplegic CP.

Was there a problem during labor?

Obstetric complications during labor can cause an ischemic insult to the fetal brain resulting in CP.

Was the child floppy at birth?

This is an important clinical question as floppiness is associated with several conditions, some more common than others. Ischemic encephalopathy is associated with a floppy infant. Floppiness is associated with genetic and muscle disorders. Spinal muscular atrophy is the most common autosomal recessive disease to cause infantile death and disability. Muscle disorders associated with floppiness include congenital muscular dystrophy, congenital myopathies, and congenital myotonic dystrophy.

Is there a history of neonatal sepsis?

Severe neonatal sepsis can result in an infant having global developmental delay, and survivors of severe meningitis may have lost limbs as a result of the vasculitis associated with the condition. Poliomyelitis in an infant can cause a delay in walking if the anterior horn cells of muscles required for ambulation are affected.

Were joint contractures evident at birth?

Arthrogryposis, a descriptive and not a diagnostic term, may cause a delay in walking.

Was a spinal deformity evident at birth?

Spinal dysraphism that affects muscles needed for ambulation may cause a delay in walking and may also be associated with contractures of the feet, knees, and hips, depending on the affected neurological level. A hairy patch, sacral dimple, meningocele, and an open lesion are all associated with spina bifida.

Did the child crawl normally?

About 12 % of children who have a delay in walking do not have a normal crawling pattern, or only do so transiently, and are mobile either by shuffling on their bottoms, by rolling, or by commando crawling (Minns 2010).

Is the child a boy with developmental delay and possibly learning difficulties?

If so, Duchenne's muscular dystrophy must be considered.

Does the child have stiff limbs?

Consider an upper motor neuron lesion or arthrogryposis.

Is there a history of cranial or spinal trauma?

Traumatic brain or spinal injury can result in a delay in walking.

Does the child have normal vision?

Visual impairment may cause a delay in walking.

Are the child's stature and body proportions normal?

Achondroplasia and spondyloepiphyseal dysplasia are associated with developmental motor delay.

Does the child have the features of rickets?

Rickets is associated with a proximal myopathy which can cause a delay in standing and walking.

25.3 Physical Examination

A careful neurological examination is most important; any further investigation should be planned on the basis of the findings as shown in Table 25.1.

25.4 Differential Diagnosis of the Cause of Delayed Walking

A delay in walking can be considered as follows: delay but the child is otherwise normal and delay and the child has underlying pathology (Noritz et al. 2013).

25.4.1 Delay in an Otherwise Normal Child

There are normal variants in the way mobility develops in the young child (Minns 2010); the majority first crawl and then walk. About 10 % of children bottom shuffle at about 12 months do not crawl and walk independently at about 19 months. Some children with Down's syn-

drome are shufflers which can also be a manifestation of acquired brain damage. Rollers are children who are mostly mobile by rolling and crawl briefly before they walk at about 16 months. This accounts for about 1 % of the population. Crawlers again account for about 1 % of the population and creep in a commando crawl fashion at about 12 months before walking at 20 months.

25.4.2 Delay in a Child with a Disorder of the CNS, Peripheral Nerves, or Muscle

Cerebral Palsy
This is the commonest chronic disability in children. The majority of children with CP have a delay in achieving independent walking. About 90 % of hemiplegics will walk by 18 months, 90 % of diplegics will walk by 48 months, and only 10 % of quadriplegics will walk by 48 months (Bleck 1987).

Muscular Dystrophy
Some boys with Duchenne's muscular dystrophy never achieve independent walking, and others may achieve it but then start to lose the ability to walk (see Chap. 22). Many have associated learning difficulties. Congenital muscular dystrophies are mostly autosomal recessive, and affected infants are floppy at birth and may also have joint contractures.

Spinal Muscular Atrophy (SMA)
SMA is the commonest cause of a floppy baby, and the incidence is about 1 in 15,000–20,000. There is a progressive loss of anterior horn cells, and it is the most common autosomal recessive disease to cause infantile death and disability. Clinically it can be divided into three types: acute infantile, chronic infantile, and chronic proximal. The first is evident between birth and 6 months of age, the weakness is progressive, and the mean survival is 6 months, and 95 % of affected infants are dead by 18 months. The chronic infantile type appears at about 6 months of age and is usually obvious by 2 years of age. It is slowly progressive, and children may survive to their third decade. The child is floppy and may develop head control and sit, but does not walk. The chronic proximal type is usually diagnosed in later childhood. The child will walk with exaggerated lumbar lordosis with a Trendelenburg gait. The hip abductors and extensors are weak. This type may be confused with DMD, but the CK level is normal.

Myopathy
Infants are floppy and have motor delay. Central core disease is an autosomal dominant condition, and severe cases do not achieve walking and die in early life.

Congenital Myotonic Dystrophy
Infants are floppy and may also have arthrogryposis, and there is a global developmental delay.

Spinal Dysraphism
Functioning quadriceps (L2, 3, 4) are needed for walking in the longer term, assuming that other factors such as motivation and learning difficulties are not a problem for ambulation. Smaller children with a motor level of L2/3 may begin to walk after a delay, but as the child grows, the energy costs are usually so great that they lose the ability eventually. Diastematomyelia may be associated with a loss of function in muscles required for ambulation (Fig. 25.1). In syringomyelia, children also have sensory dissociation with impaired pain and temperature sensation but intact touch at the level of the lesion. There is also wasting of the muscles distal to the lesion.

Paraplegia in an Infant Due to Traumatic Spinal Cord Injury or Anterior Spinal Artery Occlusion
Paraplegia due to these causes would prevent the child from walking.

Visual Impairment
This may be an isolated problem or associated with global developmental delay, for example, in severe cerebral palsy.

25.4.3 Other Causes

Arthrogryposis
Arthrogryposis is a descriptive term, and children with multiple congenital contractures (MCC)

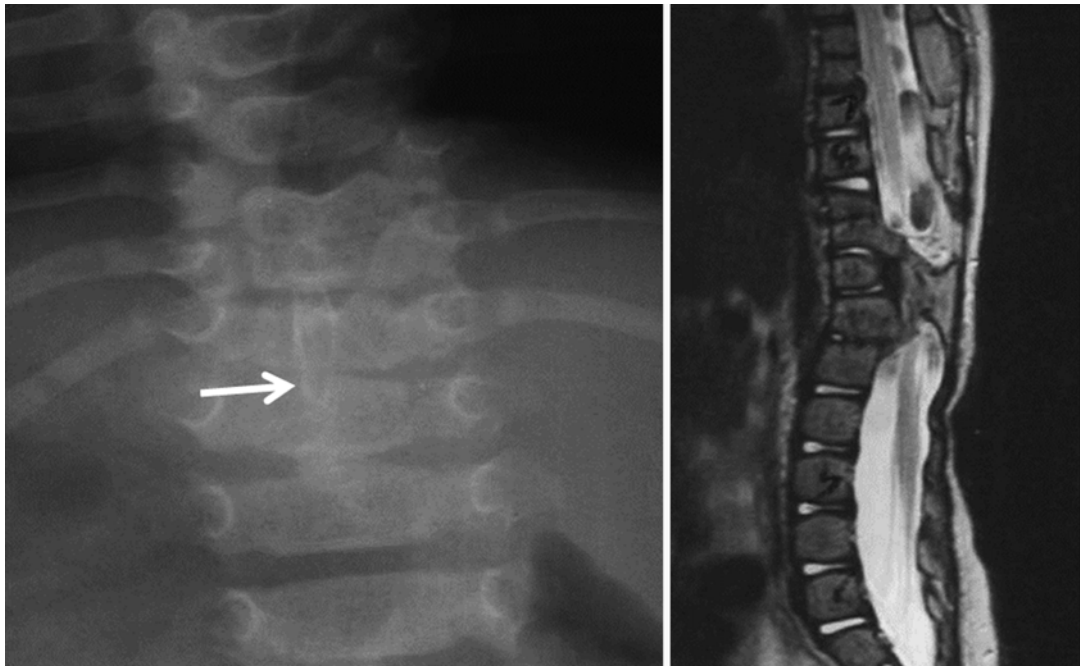

Fig. 25.1 Spina bifida and diastematomyelia (*arrow*)

may have a delay in walking as a result. MCCs are associated with neurological and muscle and connective tissue disorders, and Hall (2008) has pointed out that there are over 150 diagnoses that may be associated with it.

Genetic and Metabolic Causes

Gross motor developmental delay of about 6 months occurs in achondroplasia, and children with spondyloepiphyseal dysplasia congenita may also have a delay in walking which is usually achieved by 24 months. Proximal myopathy is seen in severe rickets and can cause a delay in standing and walking (Fig. 25.2).

Infection

Poliomyelitis affecting anterior horn cells innervating muscles required for efficient ambulation will cause a delay in walking.

Severe meningitis may leave an infant with global developmental delay and possibly limb loss.

Fetal Alcohol Syndrome

This can result in motor delay and learning difficulties.

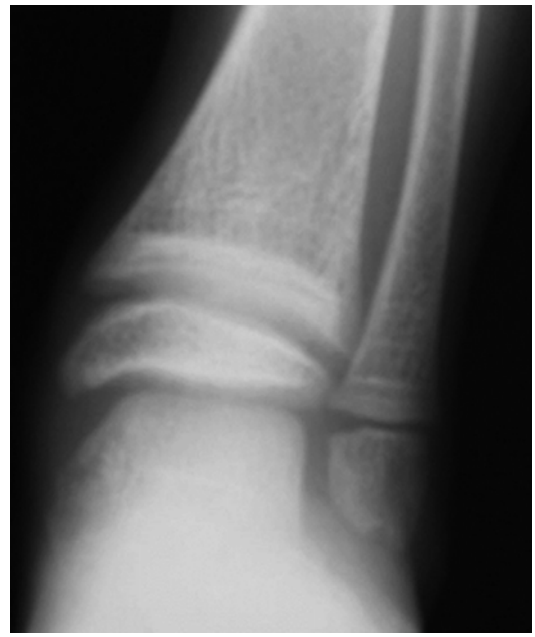

Fig. 25.2 Rickets. There is thickening and widening of the physis and cupping of the metaphysis

25.5 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Tables 25.1 and 25.2.

Table 25.1 Establishing the diagnosis of the cause of delayed walking

History					
No perinatal problems	Prematurity/perinatal problems	No perinatal problems	Akinesia (diminished fetal movements) during pregnancy	No perinatal problems	No perinatal problems
Not a floppy infant	Floppy infant occasionally	Not a floppy infant	Not a floppy infant	Not a floppy infant	Floppy infant
No global developmental delay (only delayed walking)	Global developmental delay	No global developmental delay (only delayed walking)	Global developmental delay often present	Global developmental delay may or may not be present	Global developmental delay may be present
No learning difficulties	Learning difficulties	No learning difficulties	No learning disabilities	No learning disabilities	Learning disability may or may not be present
		Acute febrile flaccid paralytic episode in infancy		Lesion in the lumbar or lumbosacral region at birth (hairy patch, dimple, swelling, or open lesion)	
				History of surgery to the back (and brain occasionally) may be present	
	No progression of symptoms	No progression of symptoms	No progression of symptoms	No progression of symptoms	Progression of symptoms
Physical examination					
Normal muscle power	Reduced muscle power	Reduced muscle power	Reduced muscle power	Reduced muscle power	Reduced muscle power
Normal muscle tone	Increased muscle tone	Decreased muscle tone	Muscle tone difficult to assess on account of contractures and joint stiffness	Decreased muscle tone	Decreased muscle tone
No spasticity	Spasticity	No spasticity	No spasticity	No spasticity	No spasticity
Normal deep tendon reflexes	Exaggerated deep tendon reflexes	Absent or diminished deep tendon reflexes	Absent or diminished deep tendon reflexes	Absent or diminished deep tendon reflexes	Absent or diminished deep tendon reflexes
Normal cognitive function	Cognitive function often impaired	Normal cognitive function	Normal cognitive function	Normal cognitive function often	Cognitive function may be impaired
Sensation normal	Sensation normal	Sensation normal	Sensation normal	Sensation impaired	Sensation normal

(continued)

Table 25.1 (continued)

			Multiple joint contractures with symmetrical deformities of the upper and lower limbs	Bowel and bladder incontinence may be present	
				Spinal deformity may be present from birth	
Working diagnosis:	Working diagnosis:	Working diagnosis:	Working diagnosis:	Working diagnosis:	Working diagnosis:
Physiologic delay	Cerebral palsy	Polio (anterior horn cell destruction)	Anterior horn cell depletion in utero (arthrogryposis)	Spinal dysraphism	Myopathy/muscular dystrophy/spinal muscular atrophy
Investigations					
–	–	–	–	Radiograph of the spine will show failure of fusion of the posterior elements of the vertebral column in the lumbar or lumbosacral region	Creatine kinase Dystrophin EMG Muscle biopsy Edrophonium test Choice as shown in Table 25.2
Diagnosis					
Physiologic delayed walker	Cerebral palsy	Poliomyelitis	Arthrogryposis	Spinal dysraphism	See Table 25.2

Table 25.2 Establishing the diagnosis of the cause of delayed walking – the floppy infant

Physical examination				
Normal muscle tone	Decreased muscle tone	Decreased muscle tone	Normal muscle tone	Normal muscle tone
Normal sensation	Normal sensation	Normal sensation	Normal sensation	Normal sensation
Muscle weakness progressive	Muscle weakness may be profound and progressive	Muscle weakness	Muscle weakness	Muscle weakness after any exertion with improvement on resting
Pseudo-hypertrophy of calf muscles may develop	No calf hypertrophy	No calf hypertrophy	No calf hypertrophy	No calf hypertrophy
Deep tendon reflexes diminished commensurate with weakness	Deep tendon reflexes weak	Deep tendon reflexes weak	Deep tendon reflexes absent or weak	Deep tendon reflexes may be normal
May have learning difficulties	No learning difficulty	No learning difficulty	Learning difficulty may be present	–
–	–	Joint contractures and foot deformities frequent	–	–
–	–	Scoliosis frequent	–	–
–	–	–	Muscle contraction persists after voluntary contraction has ended	–
Working diagnosis: Muscular dystrophy	Working diagnosis: Spinal muscular atrophy	Working diagnosis: Myopathy	Working diagnosis: Myotonia	Working diagnosis: Myasthenia
Investigations				
Creatine kinase (CK) markedly elevated	CK level normal	CK level normal	CK level normal	CK level normal
Dystrophin: Deficient in Duchenne's Abnormal expression in Becker Normal in limb girdle type Normal in congenital muscular dystrophy	Dystrophin level normal	Dystrophin level normal	Dystrophin level normal	Dystrophin level normal
–	–	Muscle biopsy diagnostic	–	–
–	–	–	Electromyography (EMG) has a characteristic pattern	Edrophonium test
Diagnosis				
Muscular dystrophy	Spinal muscular atrophy	Myopathy	Myotonia	Myasthenia

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Part IV

The Child in the School-Going Age

26 Deformities and Limitation of Movements of the Shoulder Girdle

Benjamin Joseph

26.1 Introduction

The shoulder has some peculiar characteristics that need to be appreciated in order to make an accurate diagnosis of the cause of a deformity or limitation of movement of the joint. Firstly, the shoulder has such large arcs of motion in different planes that often mild degrees of limitation of motion may go undetected. This is because extreme ranges of motion are seldom required for activities of daily living. Secondly, since what appears as movement of the "shoulder" is in reality the sum total of glenohumeral and scapulothoracic motion, a fixed deformity of the glenohumeral joint may result in a compensatory posture of the scapula. In fact, this abnormal position of the scapula is what draws attention to the underlying abnormality (Fig. 26.1).

Abnormalities in the muscles of the shoulder girdle account for a large proportion of causes of restricted motion of the shoulder. Muscle weakness, muscle contracture, co-contraction of antagonistic muscles, and muscle spasticity can all lead to limitation of shoulder motion. For example, abduction of the shoulder can be limited by weakness of the deltoid, spasticity or contracture of the pectoralis major, or co-contraction of the pectoralis major or latissimus dorsi when the deltoid is attempting to bring about abduction (Gu et al. 2000; Chuang et al. 1998a, b) (Fig. 26.2). Abnormalities in scapular morphology, position, and mobility can also limit shoulder motion (Baulot et al. 1998; Domzalski et al. 2007; Samartzis et al. 2007; Cho et al. 2000; Williams 2003) (Fig. 26.3).

26.2 Questions to Establish a Diagnosis

Questions to establish the etiology:
- **Is there a history of developmental delay?**
- **Is there a history suggestive of obstetric brachial plexus palsy?**
- **Is there a history of injections into the deltoid muscle?**
- **Are the spine, neck, and hairline normal?**

Questions to establish the pathology:
- **Is the contour of the shoulder normal?**
- **Is the position, orientation, and size of the scapula normal?**
- **Is there muscle weakness or spasticity?**
- **Is there evidence of co-contraction of the antagonistic muscles during active movement?**
- **Is there evidence of muscle contracture with a positive scapular elevation sign?**

Is there a history of developmental delay, obstructed labor, or history of injections into the deltoid muscle?

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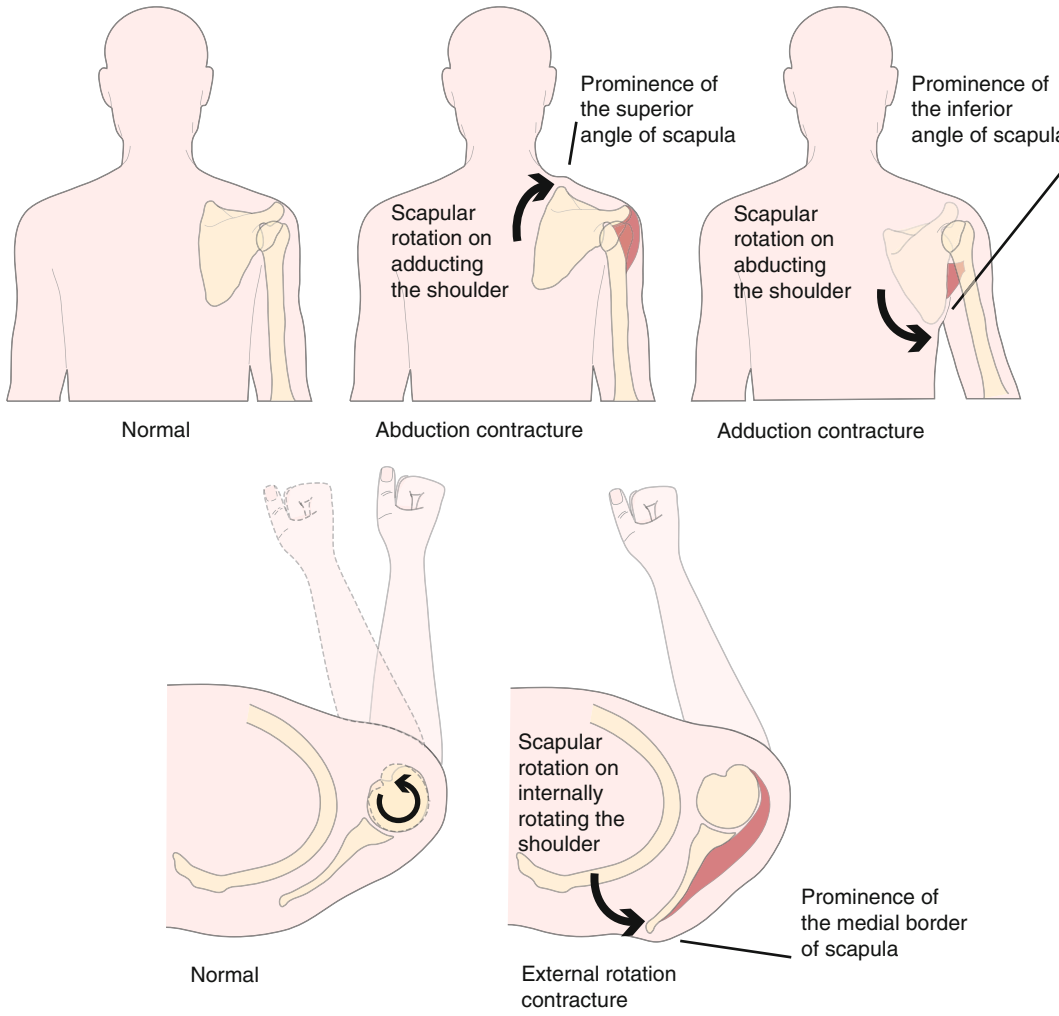

Fig. 26.1 Diagram showing how different contractures of the glenohumeral joint result in elevation of the scapula. The superior angle of the scapula, the inferior angle or the medial border may be elevated depending on the contracture that is causing the scapular elevation sign

Developmental delay raises the possibility of cerebral palsy, while a history of obstructed labor or shoulder dystocia may be forthcoming in a child with obstetric palsy. A history of repeated injections into the deltoid may be present in a child with a deltoid contracture.

Are the spine, neck, and hairline normal?

A low hairline and a webbed neck are characteristic of Klippel-Feil syndrome which is often associated with congenital elevation of the scapula or Sprengel's shoulder. Scoliosis and spina bifida are also commonly present with Sprengel's deformity (Cavendish 1972), and hence, these anomalies need to be excluded.

Is the contour of the shoulder normal?

The normal contour of the shoulder is lost when there is wasting of the deltoid; the shoulder is flattened and the acromion is prominent.

Is the position, orientation, and size of the scapula normal?

The scapula is small, elevated, and malrotated in Sprengel's shoulder and in children with obstetric brachial plexus palsy (Cavendish 1972; Nath and Paizi 2007). The abnormal rotation of the scapula contributes to the limitation of shoulder abduction in Sprengel's shoulder. On the other hand, the abnormal rotation of the scapula in obstetric

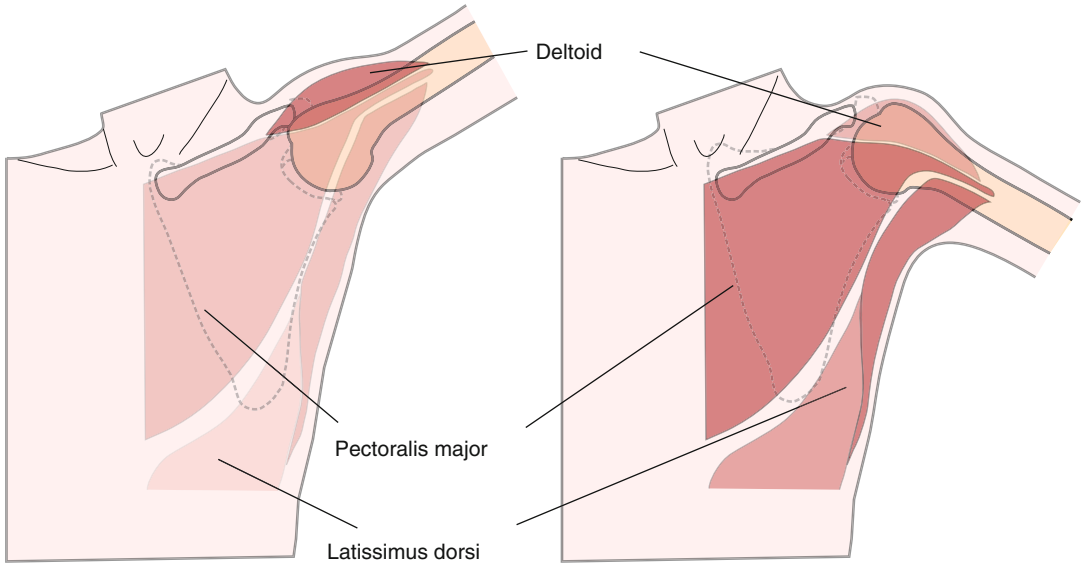

Fig. 26.2 Diagram showing the different muscular causes for limitation of shoulder motion

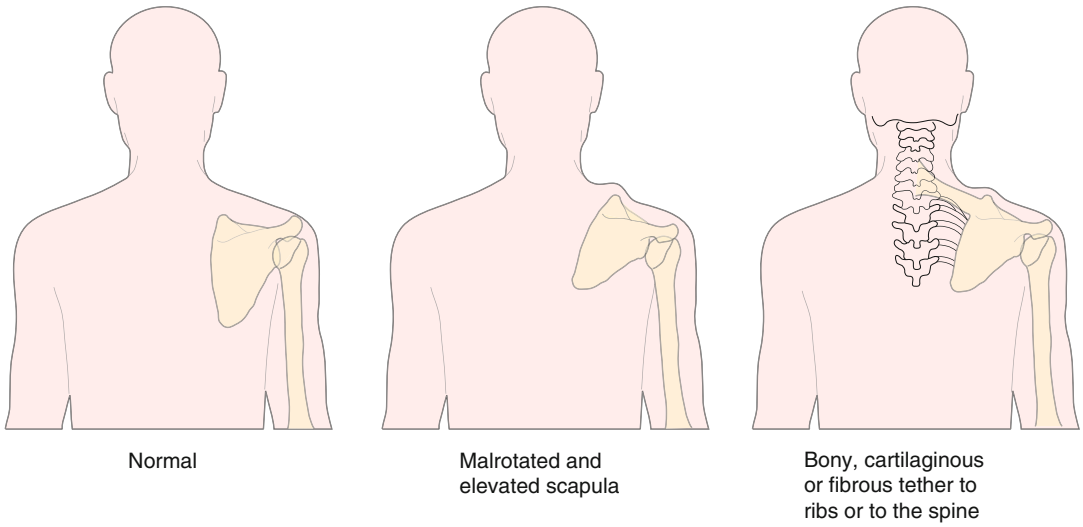

Fig. 26.3 Diagram showing the abnormalities of the scapula that can result in limitation of movement of the shoulder

palsy is due to contracture of muscles. Clearly, strategies to improve the range of motion in these two conditions are very different.

Is there muscle weakness or spasticity?

Muscle weakness or spasticity indicates an underlying neurological abnormality. While weakness itself can account for limitation of

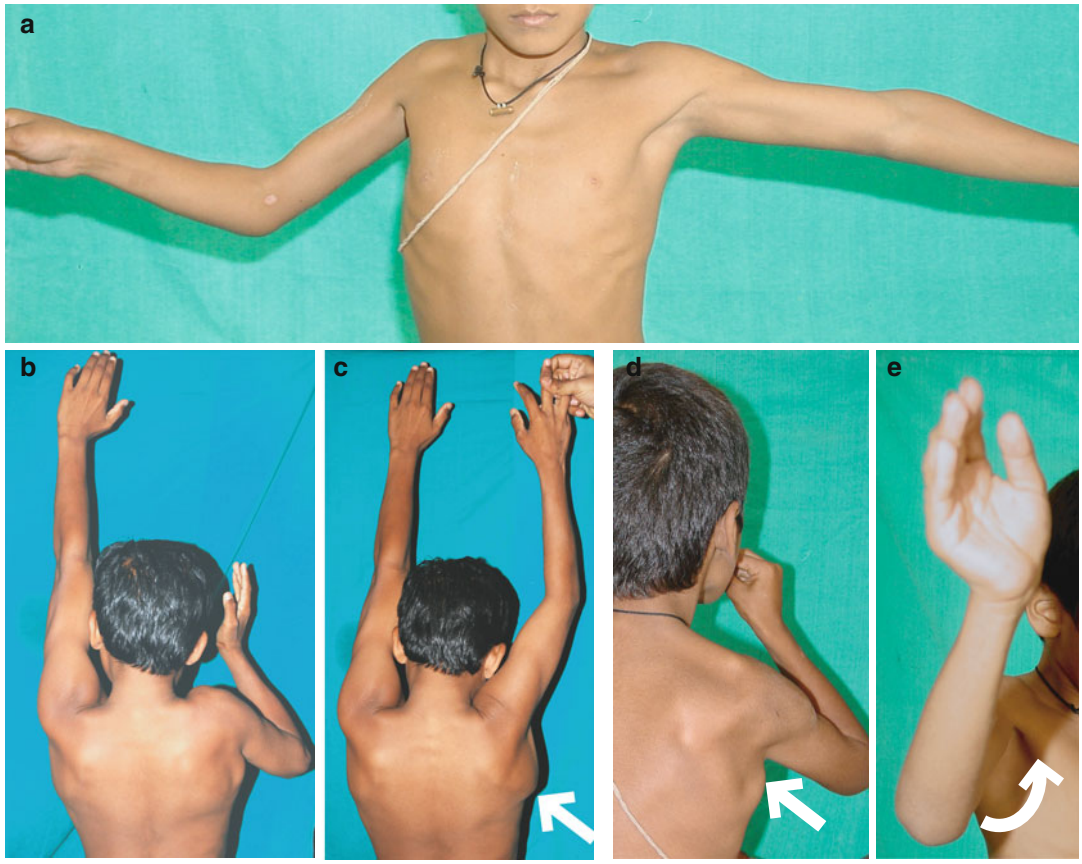

Fig. 26.4 While testing active movements of the shoulder, note if the range of motion is similar to the normal side (**a**); note if the range of active motion is less than the range of passive motion (**b**, **c**); note if the scapular elevation sign is positive (**c**, **d** – *straight arrows*), and see if there is evidence of co-contraction of the antagonistic muscles (**e** – *curved arrow*)

active motion, muscle imbalance that ensues leads to contractures which in turn restrict movement further. It is important that a clear distinction is made between limitation of movement due to weakness and limitation of movement due to a contracture as the treatment options differ.

Is there evidence of co-contraction of the antagonistic muscles during active movement?

Co-contraction is a phenomenon where antagonistic muscles contract simultaneously and this prevents normal movement. This has been attributed to cross innervation as spontaneous recovery of a brachial plexus injury occurs (Chuang et al. 1998a). One of the most common patterns of co-contraction involves the shoulder abductors and the adductors; the pectoralis major and the latissimus dorsi contract involuntarily when the child attempts to abduct the shoulder actively (Fig. 26.4e). This results in a diminished range of active abduction of the shoulder.

Is there evidence of muscle contracture with a positive scapular elevation sign?

The scapular elevation sign refers to elevation or prominence of the inferior angle, the superior angle, or the medial border of the scapula when the shoulder is actively moved in the presence of a contracture of a muscle that takes origin on the rib cage or the scapula and gets inserted on the humerus. This should not be confused with proximal displacement of the scapula. The scapula elevation sign is commonly seen in children with obstetric brachial plexus palsy (Table 26.1).

Table 26.1 Muscle contractures seen in obstetric brachial plexus palsy that cause a scapular elevation sign

Muscle that is contracted	Movement that produces scapular elevation
Subscapularis	External rotation
Teres major, latissimus dorsi	External rotation and abduction
Pectoralis major	Abduction
Infraspinatus, teres minor	Internal rotation
Supraspinatus	Adduction

26.3 Physical Examination

An accurate diagnosis can often be made by careful clinical examination alone.

26.3.1 Look

It is important that the child is observed from the front, the back, and above, having the child appropriately undressed and seated on a stool may facilitate this. Look at the contour of the shoulder, the position, size, and orientation of the scapula. Look for wasting in the supraspinous and infraspinous fossae; note wasting of the deltoid. Note if the affected limb is shorter than the normal side.

26.3.2 Feel

Systematically palpate the entire proximal humerus and the scapula including the medial and lateral borders, the spine of the scapula, the acromion process, and the coracoid to look for any palpable osteochondromas or abnormal length and inclination of the acromion and coracoid processes. Palpate the clavicle and ensure that it is normally developed.

26.3.3 Move

Ask the child to perform active movements of both shoulders simultaneously. Sitting facing the child and performing the movements yourself often encourages the child to do the same.

Ask the child to spread and raise both arms as high as possible (abduction), touch the back of the head with both hands (functional range of external rotation and abduction), touch the hands on opposite shoulders, take the hands to the mouth (functional range of internal rotation), and take the hands behind the back with the elbows flexed to 90° (maximum internal rotation). Finally ask the child to keep the elbows flexed to 90° at the side of the body and spread hands as widely away from the body as possible (maximum external rotation). These tests can provide a very good idea of the range of active movements of the shoulder.

While these active movements are being done, observe if the range of motion is normal (Fig. 26.4a); note if the medial border or the superior or inferior angle of the scapula stand out prominently (scapular elevation – Fig. 26.4c, d). If the scapular elevation is positive during any of these movements, it implies that there is a fixed deformity of the shoulder. Also note if antagonist muscles are acting when a particular movement is attempted; normally there is reciprocal inhibition of antagonistic muscles when any active movement is attempted. To note this, observe if the anterior and posterior axillary folds stand out prominently when abduction is attempted. Finally, observe the maximum range of motion achieved actively and then attempt to passively move the shoulder further (Fig. 26.4b, c). If further passive movement is possible, it implies that there is a lag in active movement due to weakness of the muscle or co-contraction of the antagonistic muscle.

Measure the passive ranges of movement of the shoulder; first fix the scapula and measure the ranges of motion of the glenohumeral joint. Then test the mobility of the scapula as the arm is put through each of the movements.

Test the muscle strength of the individual muscles acting on the shoulder. Test the muscle tone and note if any muscle is spastic.

Examine the mobility of the cervical and thoracic spine.

26.4 Investigations to Confirm the Diagnosis

Plain Radiographs
An anteroposterior radiograph of the chest will help to confirm the position, size, and shape of the scapula. Anomalies of the spine can also be identified on the radiograph. An omo-vertebral bone, if present, may be clearly delineated.

CT Scan and MRI
CT scans with 3-D reconstruction have been used to study the structural changes in Sprengel's shoulder (Cho et al. 2000) but are not needed to make the primary diagnosis. MR imaging of the entire spine is indicated if there are congenital vertebral anomalies in order to exclude intraspinal anomalies that may be present.

26.5 Differential Diagnosis

26.5.1 Obstetric Brachial Plexus Palsy

Children who have suffered obstetric brachial plexus injury develop a group of typical bony changes in the shoulder termed as SHEAR by Nath et al. (Nath and Paizi 2007) and include scapular hypoplasia, elevation, and rotation deformity (Terzis et al. 2003). It is important to be aware that precisely the same three abnormalities of the scapula are seen in Sprengel's anomaly (Samartzis et al. 2007). The affected arm, forearm, and hand are shorter than the normal side, and the degree of shortening appears to be related to the severity of neurological damage (Uysal et al. 2007).

Apart from these bony abnormalities, muscle abnormalities will invariably be present to lesser or greater degrees. These include paresis, muscle contracture, co-contraction of antagonistic muscles, and deformities. Muscle contractures are common, and the scapular elevation sign will be elicited when the contracted muscle is stretched. Often a fixed internal rotation deformity is associated with SHEAR in obstetric palsy (Nath et al. 2009).

26.5.2 Sprengel's Shoulder (Congenital "Elevation" of the Scapula)

The term "congenital elevation of the scapula" is a misnomer as it is actually a failure of descent. Sprengel's shoulder is the commonest anomaly of the scapula. While it may be seen in isolation, it is frequently associated with vertebral and rib anomalies including failure of vertebral formation (hemivertebrae) or segmentation (block vertebrae), scoliosis, spina bifida, and diastematomyelia which may be distant, as in the lumbar region (Williams 2003; Cho et al. 2000). It is important that these vertebral anomalies are diagnosed as they may also need to be addressed. In 25–50 % of patients with a Sprengel's shoulder a fibrous, cartilaginous or bony connection between the posterior elements of the cervical vertebrae and the scapula is present. The consequent restriction of scapulothoracic movement may be total if the omo-vertebral bar is bony, while moderate restriction will be noted if the connection is fibrous.

26.5.3 Injection Fibrosis of the Deltoid Muscle

Fibrosis and contracture of muscles following intramuscular injection have been noted in certain races, and the contracture of the deltoid is well recognized (Chatterjee and Gupta 1983; Shanmugasundaram 1980; Chen et al. 1988). The scapular elevation sign will be present when the shoulder is adducted.

26.5.4 Cerebral Palsy

In hemiplegic cerebral palsy, the shoulder tends to be internally rotated and the internal rotators and adductors are spastic. Consequently, abduction and external rotation movements are limited.

26.5.5 Mechanical Impingement and Growth Disturbance of the Proximal Humerus

If there is an osteochondroma of the proximal humerus or on the scapula near the glenoid, mechanical impingement may limit motion. If there is asymmetric growth arrest of the proximal humeral physis, a deformity of the proximal humerus will develop and get progressively worse. Consequently, limitation of shoulder motion will ensue. Humerus varus is one such example, but the limitation of motion may only become apparent later in childhood. Internal rotation contracture following obstetric brachial plexus palsy can lead onto a posterior dislocation of the shoulder with loss of external rotation.

26.5.6 Rare Conditions

Congenital Deltoid Bands
Rare cases of fibrotic bands in the deltoid have been described (Bhagat et al. 2008). The clinical features are very similar to deltoid contracture following intramuscular injection.

Congenital Short Costocoracoid Ligament
An autosomal dominant condition has been reported in a family with altered contour of the shoulder and limitation of shoulder movements (Bamforth et al. 1989). The underlying anomaly in the affected individuals was a congenitally short costocoracoid ligament. Rotations of the shoulder and retraction of the scapula are limited in this condition.

26.6 Establishing the Diagnosis

Outlines for establishing the causes of deformities of the shoulder with limitation of motion are shown in Tables 26.2 and 26.3.

Table 26.2 An outline of the process of establishing the diagnosis of deformities and diminished movement of the shoulder in the school-going child

History				
A positive family history is seldom present	No positive family history	No positive family history	A positive family history may occasionally be present	No positive family history
	History of difficult labor may be present	History of injections into the arm may be present		Delay in milestones may be present
	History of flaccid paralysis of the upper limb soon after birth present			
Physical examination				
Short neck with webbing, scoliosis may be present	Neck and spine normal	Neck and spine normal	Neck and spine normal	Neck and spine normal
SHEAR (scapular hypoplasia, elevation, and malrotation) present	SHEAR (scapular hypoplasia, elevation, and malrotation) present	Scapula malrotation alone present	No SHEAR	No SHEAR

(continued)

Table 26.2 (continued)

Active and passive ranges of motion are the same	Active motion more restricted than passive motion in some planes	Active and passive ranges of motion are the same	Active and passive ranges of motion are the same	Active motion more restricted than passive motion
Scapula remains elevated, but scapular elevation sign does not manifest with different movements of the shoulder	Scapular elevation sign may be noted during active adduction, abduction, external, or internal rotation	Scapular elevation sign present only on adduction of the shoulder	Scapular elevation sign usually not present	Scapular elevation sign not present
Co-contraction of antagonistic muscles not seen	Co-contraction of antagonistic muscles frequently seen	Co-contraction of antagonistic muscles not seen	Co-contraction of antagonistic muscles not seen	Co-contraction of antagonistic muscles not seen
Muscle weakness not present	Muscle weakness present	Muscle weakness not present	Muscle weakness not present	Muscle weakness and spasticity present
No abnormal bony mass on proximal humerus	No abnormal bony mass on proximal humerus or the scapula	No abnormal bony mass on proximal humerus or the scapula	Palpable bony prominence on proximal humerus or the scapula	No abnormal bony mass on proximal humerus or the scapula
Scapulothoracic movements markedly restricted	Scapulothoracic movements unaffected	Scapulothoracic movements unaffected	Scapulothoracic movements unaffected	Scapulothoracic movements unaffected
Investigations				
Plain radiograph of the trunk to show both scapulae, the rib cage, and the spine Confirms abnormalities of scapular size and position Demonstrates presence of omo-vertebral bone, if present Demonstrates associated congenital abnormalities of the spine	Plain radiograph not routinely indicated	Plain radiograph not routinely indicated	Plain radiograph of the shoulder to confirm the cause of mechanical impingement (e.g., osteochondroma)	Plain radiograph not routinely indicated
Diagnosis				
Sprengel's shoulder	Obstetric brachial plexus palsy with muscle contracture	Deltoid fibrosis (congenital – if no history of injections into the muscle; injection fibrosis if there is a definite history of injections)	Osteochondroma of the humerus or scapula	Cerebral palsy

Table 26.3 Outline of establishing the pathologic process responsible for reduction in the range of motion of the shoulder based on clinical signs

Clinical sign	Muscle paralysis	Muscle spasticity	Muscle contracture	Muscle co-spasticity	Bony deformity or prominence	Scapular malrotation	Scapular tether
Reduced active range of motion	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Reduced passive range of motion	No	Yes (if spasticity severe)/No	Yes	No	Yes	Yes	Yes
Range of passive motion > range of active motion	Yes	Yes (if spasticity is mild)/No	No (both are equal)	Yes	No	No	No
Reduced muscle power	Yes	Yes/No	No	Yes/No	No	No	No
Increased muscle tone	No	Yes	No	No	No	No	No
Scapular position at rest	Normal	Normal	Normal/abnormal (if deltoid is contracted)	Normal	Normal/abnormal (depends on the site of bony prominence)	Abnormal	Abnormal
Scapular elevation sign on active movement	No	No	Yes	No	No	No	No
Scapulothoracic movement	Normal	Normal	Normal	Normal	Normal/affected if the lesion is on the scapula	Normal (if there is no associated scapular tether)	Reduced

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Deformities and Limitation of Movement of the Elbow

27

Benjamin Joseph

27.1 Introduction

Deformities of the elbow in the sagittal plane (flexion and extension deformities) will result in reduction in the range of motion of the elbow but coronal plane deformities will not restrict motion (Fig. 27.1). Unlike what is seen in the shoulder, minor degrees of reduction of both flexion and extension will attract the attention of the parents even though they may not cause significant functional disability.

Cubitus varus and valgus develop secondary to bony abnormalities, while flexion and extension deformities are more often due to soft tissue contractures.

Apart from deformities of the elbow that are evident at birth and persist into later childhood (see Chap. 8), there are developmental and acquired deformities that initially manifest in the young child; these are discussed in this chapter.

27.2 Establishing the Diagnosis of the Cause of Cubitus Varus and Valgus

27.2.1 Questions to Establish a Diagnosis of Cubitus Varus and Valgus

- **Are there bilateral symmetric deformities?**
- **Is there a history of trauma preceding the onset of the deformity?**
- **Is the deformity progressive?**
- **Are the radius and ulna of normal length and alignment?**
- **Are the stature and body proportions normal?**

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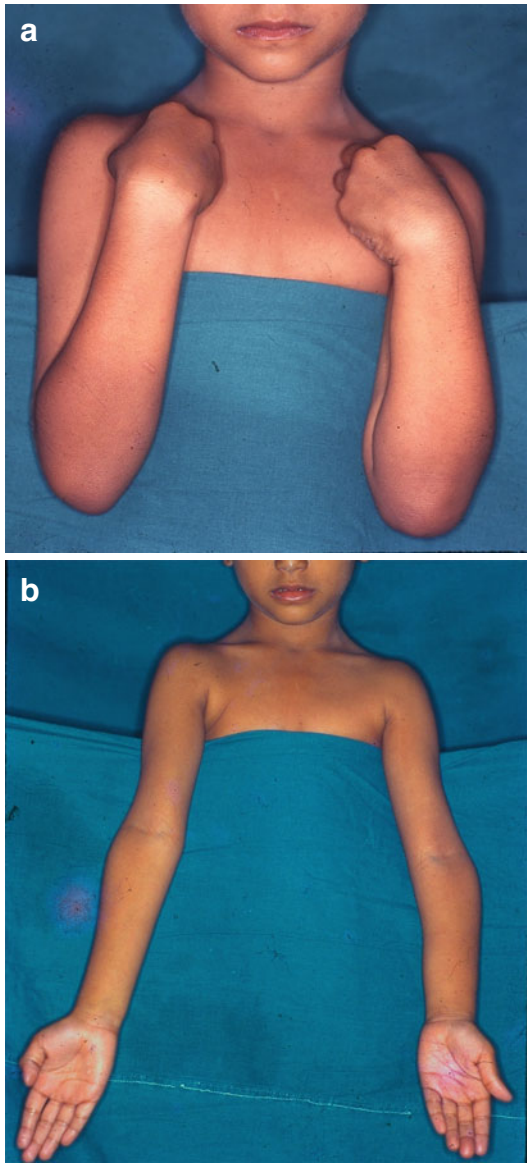

Fig. 27.1 (a, b) Full range of motion of the elbow in a child with cubitus varus

Are there bilateral symmetric deformities?

Unilateral cubitus varus or valgus is likely to be on account of trauma or localized growth abnormality. Bilateral symmetric deformities occur in skeletal dysplasia and in children with chromosomal abnormalities.

Is there a history of trauma preceding the onset of the deformity?

The commonest cause for unilateral cubitus varus in childhood is malunion of a supracondylar fracture of the humerus. The deformity that develops due to the malunion is aptly described as a gunstock deformity (Fig. 27.2). Cubitus valgus more commonly follows a fracture of the lateral condyle. Though these injuries may occur in the preschool age, they are more frequently seen in children who are a few years older.

Is the deformity progressive?

Progression of deformity suggests that the growth plate is affected.

Are the radius and ulna of normal length and alignment?

A short ulna as in hereditary multiple osteochondromatosis may be associated with cubitus varus (Fig. 27.3). A chronically dislocated radial head may be associated with cubitus valgus.

Are the stature and body proportions normal?

Cubitus varus and cubitus valgus are seen in some skeletal dysplasias where altered body proportions and short stature may be present. Sex chromosomal anomalies associated with short stature tend to have cubitus valgus (Baughman et al. 1974).

27.2.2 Physical Examination

Look

Note the stature of the child and if there is proportionate or disproportionate dwarfism.

Examine the limb in full extension as cubitus varus and valgus may be masked if the elbow is flexed.

Feel

Palpate the distal end of the humerus, proximal ulna, and radius. Note if they are of normal lengths, if their relative positions are maintained, and if there are abnormal prominences close to the end of any of these bones.

Move

Check the active and passive movements of the elbow and forearm.

Measure

Measure the carrying angle with a goniometer and compare the values against available norma-

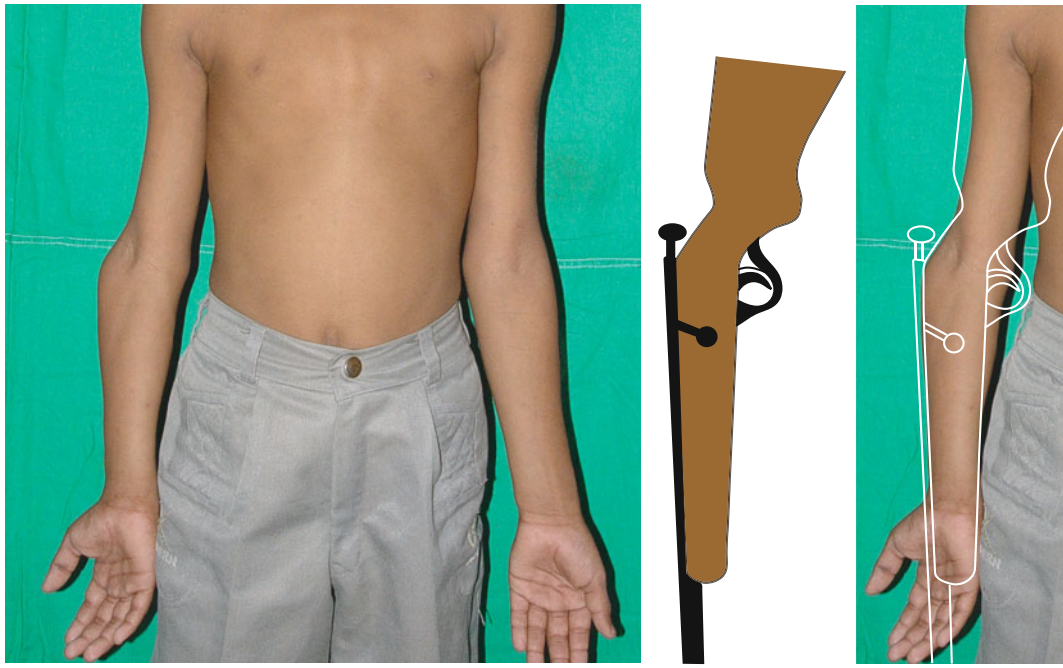

Fig. 27.2 Cubitus varus of the right elbow due to malunion of a supracondylar fracture of the humerus. The loss of the carrying angle as compared to the normal side and the typical gunstock deformity is seen

tive data for the age and gender of the child (Balasubramanian et al. 2006; Golden et al. 2007; Yilmaz et al. 2005). The normal mean carrying angle measured clinically in boys aged 5 years is 8.6° ± 4.2° and 10.0° ± 3.0° in girls of the same age (Balasubramanian et al. 2006).

Measure the lengths of the humerus, radius, and ulna.

27.2.3 Investigations to Confirm the Diagnosis

Plain Radiographs

Plain radiographs of the affected limb should enable a definitive diagnosis and the severity of the deformity can be estimated by measuring the Baumann's angle provided the elbow extends fully (Acton and McNally 2001) (Fig. 27.4).

MRI Scan

If growth plate damage is suspected, MR imaging may be undertaken to delineate the growth plate clearly (Kim et al. 2002).

An outline of the approach to establishing the cause of cubitus varus or valgus in the school-going child is shown in Table 27.1.

27.2.4 Differential Diagnosis

Complication of Fracture of Distal Humerus

Cubitus varus and valgus deformities following fractures of the distal humerus are frequently seen in older children; cubitus varus is most frequently seen after malunion of a supracondylar fracture, while cubitus valgus is seen following a lateral condylar fracture. Though this is the common pattern, in rare instances the reverse pattern may be encountered.

Hereditary Multiple Osteochondromatosis

Abnormal growth patterns are seen in bones in hereditary multiple osteochondromatosis. The location of the osteochondroma and its proximity to the growth plate determine the pattern of growth abnormality (Cho and Jung 2014; Matsubara et al. 2006).

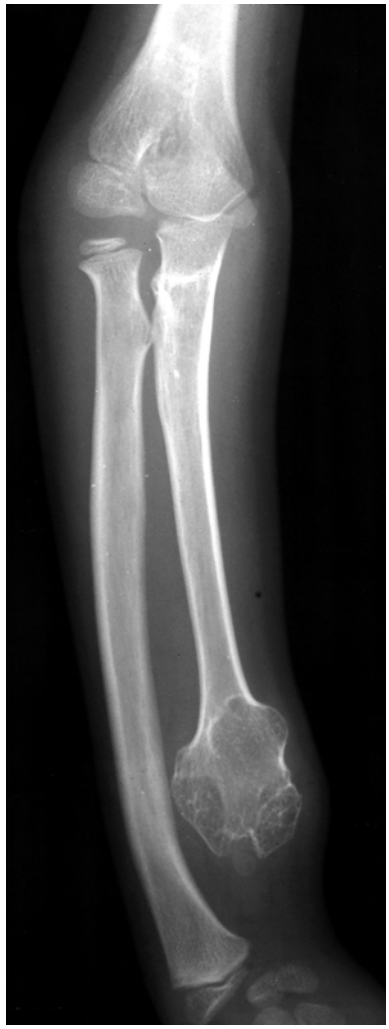

Fig. 27.3 Radiograph of the forearm of a child with hereditary multiple osteochondromatosis and cubitus varus

27.3 Establishing the Cause of Limitation of Elbow Motion and Flexion or Extension Deformity of the Elbow

Limitation of active motion of the elbow is mainly due to weakness of the flexors or extensors, while the loss of passive motion is due to fixed deformities. Fixed flexion or extension deformities that are not evident at birth and first noted in the young child are almost always due to soft tissue contractures frequently involving the flexor or extensor muscles.

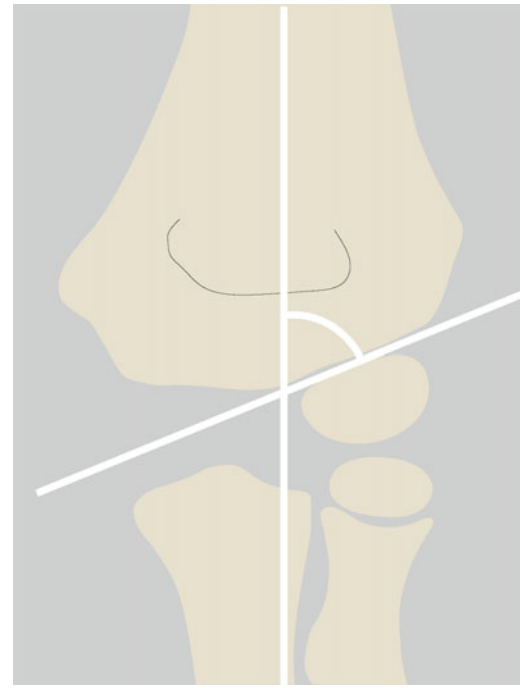

Fig. 27.4 Technique of measuring the Baumann's angle from an anteroposterior radiograph

27.3.1 Questions to Establish a Diagnosis of Flexion or Extension Deformity of the Elbow

- **Is there a history of injections into the arm?**
- **Are there features of obstetric brachial plexus palsy or cerebral palsy?**
- **Are there features of a skeletal dysplasia?**

Is there a history of injections into the arm?

A history of injections into the triceps in infancy may be present.

Are there features of obstetric brachial plexus palsy or cerebral palsy?

A history of a difficult birth or delayed milestones may point to a neurological insult resulting in muscle contracture.

Are there features of a skeletal dysplasia?

If there are features such as dwarfism and symmetric deformities of the upper and

Table 27.1 An outline of the process of establishing the diagnosis of the cause of cubitus varus or valgus

History				
History of trauma present	History of trauma present	No history of trauma	No history of trauma	No history of trauma
No history of progression of deformity	History of progression of deformity present	History of progression of deformity present	No history of progression of deformity	No history of progression of deformity
Physical examination				
Unilateral deformity	Unilateral deformity	Unilateral or bilateral	Bilateral symmetric deformities	Bilateral symmetric deformities
Stature normal	Stature normal	Short stature may be present	Short stature and altered body proportions may be present	Short stature may be present (with cubitus valgus)
Humerus and forearm of normal length	Humerus or forearm may be shorter than normal side	Forearm often short Ulna usually shorter than the radius	Forearm may be short	Humerus and forearm of normal length
				Abnormalities in genitalia and secondary sexual characteristics
Investigations				
Plain radiograph of forearm and elbow Evidence of malunion of fracture of distal humerus (common)/ evidence of old fracture of the lateral condyle/dislocation of the radial head	Plain radiograph of forearm and elbow Growth plate irregularity may be evident	Plain radiograph of forearm and elbow Presence of osteochondroma on the radius or ulna Short ulna Radius may be bowed	Plain radiograph of elbow and skeletal survey for features of skeletal dysplasia Symmetric growth abnormalities and changes in epiphyses/metaphyses/or the spine	Plain radiograph not indicated
–	MRI: May demonstrate asymmetric growth plate damage	–	–	–
–	–	–	–	Chromosomal studies: Abnormalities in the sex chromosome number
Diagnosis				
Cubitus varus or valgus due to malunion of fracture of distal humerus or radial head dislocation	Cubitus varus or valgus due to growth plate injury to distal humeral growth plate	Cubitus varus due to hereditary multiple osteochondromatosis	Cubitus varus or valgus in skeletal dysplasia	Cubitus valgus or varus in sex chromosomal abnormalities

lower limbs, the child may have a skeletal dysplasia.

Is there a history of infection at or in the vicinity of the elbow?

Fibrosis of the flexor or extensor muscles of the arm can ensue after osteomyelitis of the humerus or bony ankylosis of the elbow can follow septic arthritis of the elbow.

27.3.2 Physical Examination

Look

Note if there is wasting of the muscles of the shoulder, arm, and forearm. Note if the affected limb is hypoplastic. Note if there are other symmetric abnormalities suggestive of a skeletal dysplasia.

Ask the child to raise the arm above the shoulder and note if the elbow remains extended (Fig. 27.5) or if it drops into flexion.

Move and Measure

Measure the ranges of active and passive movements of the elbow and note if they are the same. Measure and record the muscle power of the flexors and extensors of the elbow.

27.3.3 Investigations to Confirm the Diagnosis

Plain Radiographs

Plain radiographs of the affected limb and a complete skeletal survey are indicated if a skeletal dysplasia is suspected.

27.3.4 Differential Diagnosis

Obstetric Brachial Plexus Palsy

A flexion deformity of the elbow may develop in children with weak elbow extensors, while an extension deformity may develop if the flexors are weaker than the extensors. Demonstrable weakness of the antagonistic muscle is usually present in these children; the weakness however may not be profound (Fig. 27.5).

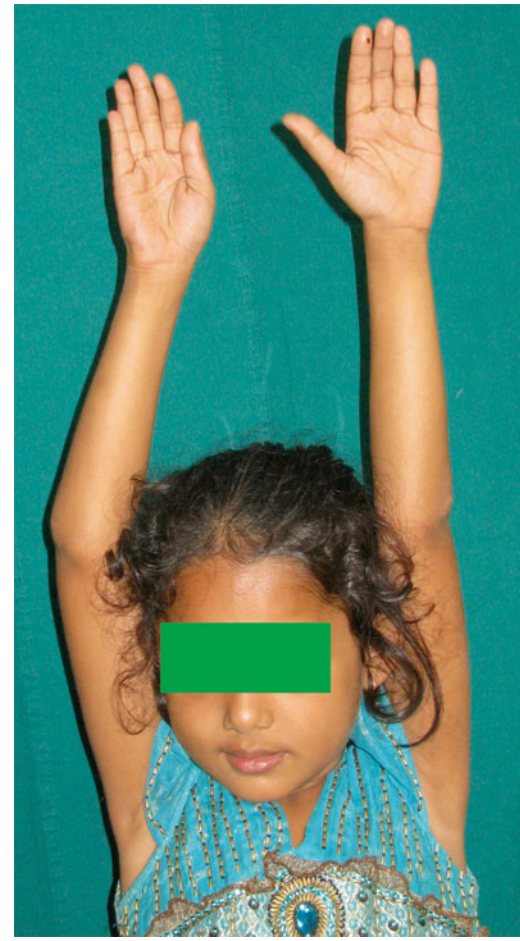

Fig. 27.5 Flexion deformity of the right elbow in a child with obstetric brachial plexus palsy. Note that when she raises her arms above the shoulder, the elbow remains extended up to the point of the fixed flexion deformity. This demonstrates that the power of the triceps is at least Grade III on the MRC scale. Note that the limb is hypoplastic; this is a feature of paralytic conditions affecting growing children

Cerebral Palsy

Flexion deformity of the elbow may be seen in children with cerebral palsy. This is usually in association with pronation of the forearm and flexion of the wrist due to spasticity of the flexors and pronators (Landi et al. 2003; Manske et al. 2001).

Arthrogryposis

The elbow may be in flexion or extension in arthrogryposis. When the elbow is extended often, there are associated weakness of the biceps and contracture of the triceps. Both these issues will need to be addressed to restore function.

Injection Fibrosis of the Triceps Muscle

Fibrosis and contracture of the triceps muscle have been described following injections into the muscle (Babhulkar 1985).

Ankylosis of the Elbow

The elbow joint is particularly prone to develop extra-articular stiffness following trauma or infection. Rarely bony ankylosis may develop as a complication of joint sepsis. The position of fusion determines the extent of functional disability. An elbow ankylosed in some degree of flexion is more functional than one fused in full extension (Fig. 27.6).

Fig. 27.6 Ankylosis of the elbow following septic arthritis complicating osteomyelitis of the humerus. The functional limitations of an elbow fused in flexion are shown

Fixed Flexion Deformity of the Elbow Associated with a Dysplasia or Syndrome

Flexion deformity of the elbow may be seen in several syndromes and skeletal dysplasias including chondrodysplasia punctata, mucopolysaccharidoses, and Schmid metaphyseal dysplasia, Larsen syndrome (Fig. 27.7), and distal humeral dysplasia (Joseph 2000; Joseph and Varghese 2003). In some of these situations, the radial head may be dislocated and in a few the distal end of the humerus may be dysplastic and the elbow dislocated (Hermanns et al. 2008; Joseph and Varghese 2003).

An outline of establishing the cause of limitation of motion and flexion or extension deformities of the elbow in a young child is shown in Table 27.2. The table does not set out the process of identifying the etiology but attempts to show how to identify the pathology since the treatment hinges on recognizing the pathology and correcting it.

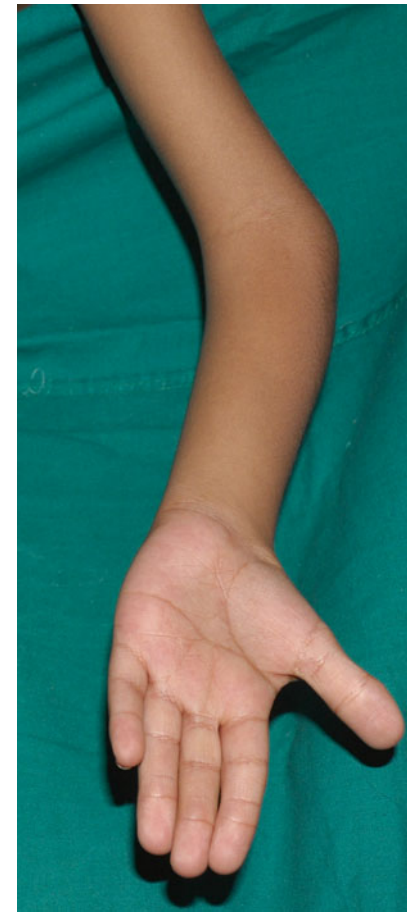

Fig. 27.7 Flexion deformity of the elbow in a child with Larsen syndrome

Table 27.2 An outline of the process of establishing the diagnosis of the pathologic cause of flexion or extension deformity of the elbow

Physical examination					
Muscle weakness		Muscle contracture		Muscle weakness + contracture of antagonist	
Power of triceps decreased	Power of biceps decreased	Power of biceps normal	Power of triceps normal	Power of triceps decreased	Power of biceps decreased
No fixed deformity	No fixed deformity	Fixed flexion deformity	Fixed extension deformity	Fixed flexion deformity	Fixed extension deformity
Range of active elbow extension appears normal if power tested with child sitting or standing (as gravity assists extension of elbow) Range of active extension decreased when child attempts to raise arm above the head if power < Grade III	Range of active elbow flexion decreased if power < Grade III	Range of active extension reduced Power of extension normal within the possible range (up to the position of fixed deformity)	Range of active flexion reduced Power of flexion normal within the possible range (up to the position of extension deformity)	Range of active extension reduced Power of extension reduced within the possible range (up to the position of fixed deformity)	Range of active flexion reduced Power of flexion reduced within the possible range (up to the position of extension deformity)
Range of passive extension full	Passive flexion full range	Range of passive extension reduced to the same extent as reduction of active extension	Range of active flexion reduced to the same extent as reduction of active flexion	Range of passive extension reduced to the same extent as reduction of active extension	Range of passive flexion reduced to the same extent as reduction of active flexion
Diagnosis					
Isolated triceps weakness	Isolated biceps weakness	Contracture of elbow flexors (usually involves biceps and brachialis)	Contracture of the triceps	Contracture of the elbow flexors with weakness of the triceps	Contracture of triceps with weakness of the elbow flexors
Treatment implication: If weakness is severe enough to warrant treatment, a tendon or muscle transfer needs to be considered		Treatment implication: If the fixed deformity is severe enough to warrant treatment, the contracture must be released		Treatment implication: If treatment is warranted, contracture release must be combined with a tendon transfer to augment the weak antagonist	

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Deformities and Limitation of Motion of the Forearm

28

Benjamin Joseph

28.1 Introduction

Pronation and supination are complex movements of the forearm that enable positioning the hand appropriately to execute functions related to the activities of daily living. Recent studies suggest that pronation and supination are not brought about by mere movement of the radius around a static ulna but entails movement of the radius, ulna, and the interosseous membrane (Weinberg et al. 2000; Nakamura et al. 1999). The integrity and normal structure of the proximal and distal radioulnar joints, the radius and ulna, the interosseous membrane, and the muscles that pronate and supinate the forearm are all needed for normal forearm motion. It follows that abnormalities in any of these structures can compromise pronation and supination (Ogino and Hikino 1987; Dal Monte et al. 1987; Yasutomi et al. 2002; Tynan et al. 2000; Price et al. 1990; Sibinski et al. 2007; Kreulen et al. 2007) (Fig. 28.1).

The normal range of pronation and supination of the forearm is so wide (80–90° each of pronation and supination) that mild reduction of motion may go unnoticed. It also needs to be emphasized that even when there is profound reduction of one or both of these movements, the parents may only notice that the child performs some activities of daily living in an awkward fashion; they may not attribute this to loss of pronation or supination. The activities that obviously require pronation and supination are typically those that involve screwing and unscrewing motions but very many other self-care activities require a good range of pronation, supination, or both. For example, limitation of supination of the forearm can compromise simple activities such as combing one's hair (Video 28.1), getting food to the mouth (Video 28.2), wiping the forehead (Video 28.3), or touching the shoulder (Fig. 28.2). Awkwardness in any such activities of daily living is what attracts the parents' attention to the problem. Consequently, only when the child is old enough to do some of these activities will the parents notice that something is amiss. In addition, some compensations occur at the shoulder for restricted forearm motion. This explains why a condition like congenital radioulnar synostosis is seldom diagnosed in early infancy.

The extent of disability from limitation of pronation or supination will depend on which of the movements is compromised, the severity of the restriction of motion and the position in which the forearm is fixed (Fig. 28.3). It will also depend on how much compensation for the limitation of motion of the forearm occurs at the shoulder and wrist (Ogino and Hikino 1987).

Electronic supplementary material The online version of this chapter (doi:10.1007/978-81-322-2392-4_28) contains supplementary material, which is available to authorized users.

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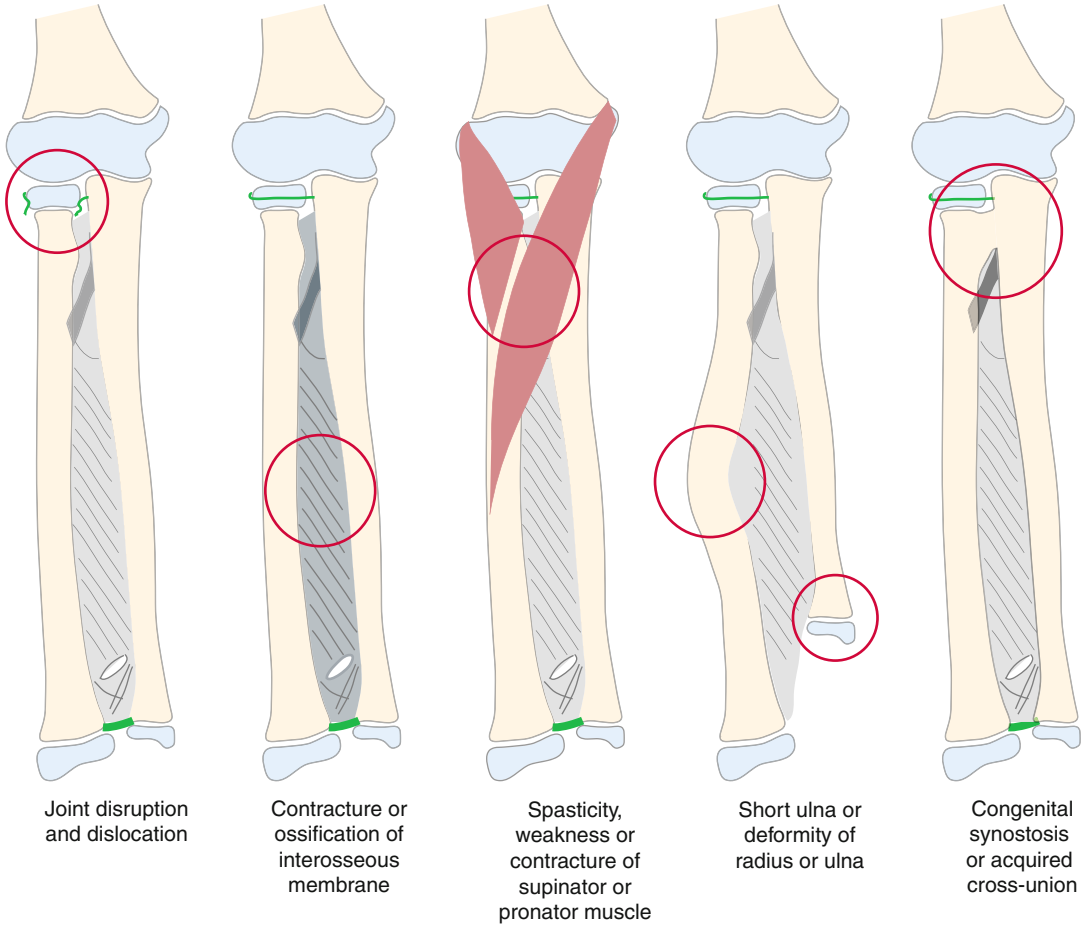

Fig. 28.1 Factors that affect the motion of the forearm

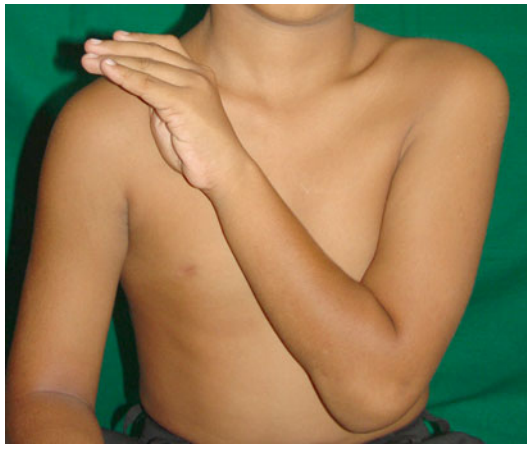

Fig. 28.2 A child with congenital radioulnar synostosis is attempting to touch his shoulder. The fixed pronation of the forearm prevents the palm from touching the shoulder

The more common causes of limitation of pronation and supination include congenital, developmental, paralytic, and traumatic causes.

28.2 Questions to Establish a Diagnosis

- **Is the limitation of motion unilateral or bilateral?**
- **Is there a history of trauma?**
- **Was the child a large baby at birth and was there a history of a difficult or obstructed labor?**
- **Is there some developmental delay?**

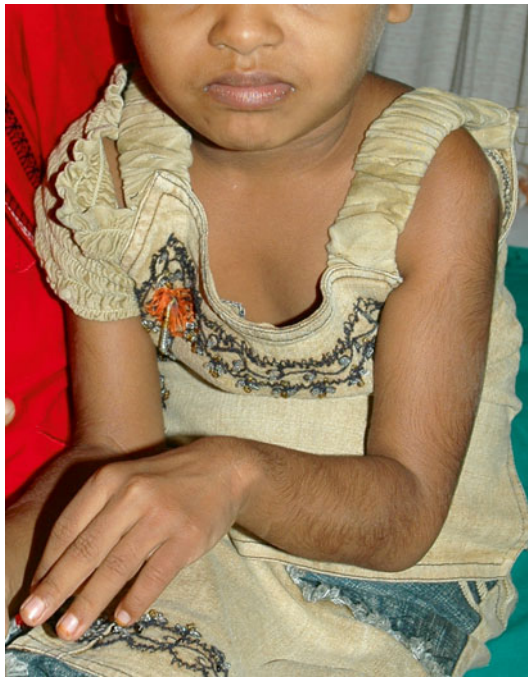

Fig. 28.3 The forearm of this child with congenital radioulnar synostosis is fixed in 75° of pronation

- **Is there facial dysmorphism and features of any recognized syndrome?**
- **Is there a family history of limitation of motion of the forearm?**

Is the limitation of motion unilateral or bilateral?

Bilateral limitation of motion of the forearm is likely to be on account of a congenital anomaly such as congenital radioulnar synostosis or congenital radial head dislocation, both of which frequently affect both sides (Dal Monte et al. 1987). Acquired causes, on the other hand, usually affect one limb only.

Is there a history of trauma?

Malunion of a fracture of both bones of the forearm (Yasutomi et al. 2002; Tynan et al. 2000; Perron et al. 2001), a missed Monteggia fracture, or a traumatic dislocation of the radial head (Weisman et al. 1999) may all result in limitation of motion of the forearm.

Was the child a large baby at birth and was there a history of a difficult or obstructed labor?

The history suggestive of difficult labor may be obtained in children who have obstetric brachial plexus palsy. Despite the initial weakness of supination noted in many children with obstetric brachial plexus palsy, limitation of both supination and pronation is often encountered later on (Sibinski et al. 2007).

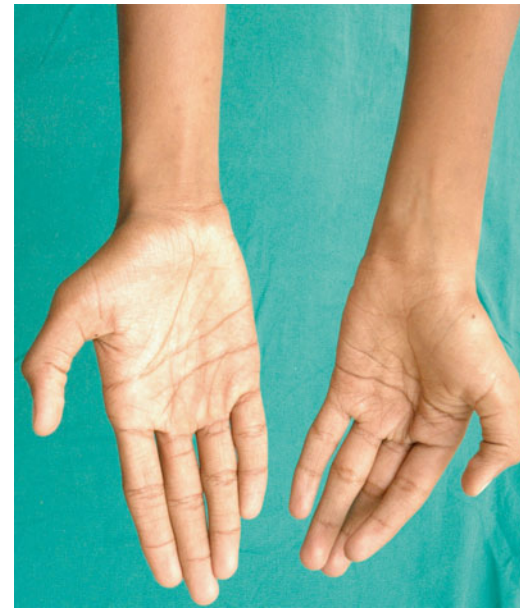

Fig. 28.4 Limited supination of the left forearm is seen in a child with left-sided hemiplegic cerebral palsy

Is there some developmental delay?

Children with hemiplegic cerebral palsy will have some limitation of forearm rotation, particularly supination (Fig. 28.4), and most of these children will have some degree of motor development delay (Kreulen et al. 2007; Yokochi et al. 1992). These children typically start walking at around 18 months, while normal children begin to walk by 14 months of age.

Is there facial dysmorphism and features of any recognized syndrome?

Congenital radioulnar synostosis is often associated with one of several syndromes with a genetic basis (Cleary and Omer 1985; Green and Mital 1979; Rizzo et al. 1997; Crisponi et al. 1997). Some syndromes with thrombocytopenia have been identified in children with radioulnar synostosis, and screening children with radioulnar synostosis for thrombocytopenia may be in order as this has a bearing on treatment of these children (Giuffre et al. 1994; Thompson et al. 2001).

Is there a family history of limitation of motion of the forearm?

A positive family history is often present in hereditary multiple osteochondromatosis. A familial tendency has also been noted in a proportion of cases of congenital radioulnar synostosis.

28.3 Physical Examination

Careful examination is most important for establishing the diagnosis.

28.3.1 Look

Look for facial dysmorphism, growth retardation, and wasting of the muscles of the shoulder, arm, and forearm. Ask the child to perform the following actions with both hands simultaneously, and observe if the shoulder, elbow, forearm, and wrist motions are normal or abnormal:

- With the elbows flexed to 90° and with the arms by the side of the trunk, ask the child to face both palms upward (maximum active supination; Fig. 28.5), and then face both palms downward (maximum active pronation).
- Touch both palms on the opposite shoulder.
- Wipe the opposite cheek.
- Take a mug to the mouth.

Look for prominence of the radial head on the lateral aspect of the elbow.

28.3.2 Feel

Systematically palpate the radius and the ulna throughout their entire lengths, and note if they are of normal relative lengths or if either bone is bowed. Carefully palpate the head of the radius and the lower end of the ulna, and note if the proximal and distal radioulnar joints are dislocated. Palpate the metaphyseal regions of the radius and ulna to see if there are palpable osteochondromata.

28.3.3 Move

Test the passive range of pronation and supination with the elbow held at 90° of flexion.

Fig. 28.5 The child is attempting active supination of both the forearms with the elbows flexed to 90°. The right forearm can be actively supinated, but the left forearm is fixed in pronation; no active supination is possible. The child also has preaxial polydactyly on the right hand

Stabilize the elbow, and pronate and supinate the forearm passively (Fig. 28.6). Making the child hold a pen or pencil in each hand helps in giving a clearer impression of the range of motion of the forearm (Fig. 28.7a, b).

28.3.4 Special Tests

Test the power of muscles on the shoulder, arm, forearm, and wrist by manual muscle testing, and grade the muscles according to the Medical Research Council (MRC) grading.

Test for spasticity by noting the resistance to passive movement of the forearm, elbow, and wrist. If the tone is increased, test the range of motion first by slow passive stretch. Test again by rapidly performing the movement. A demonstrable difference in the range of motion with the range being less on rapid movement than on slow stretching denotes spasticity.

Fig. 28.6 To accurately test the range of passive pronation and supination, the elbow must be stabilized

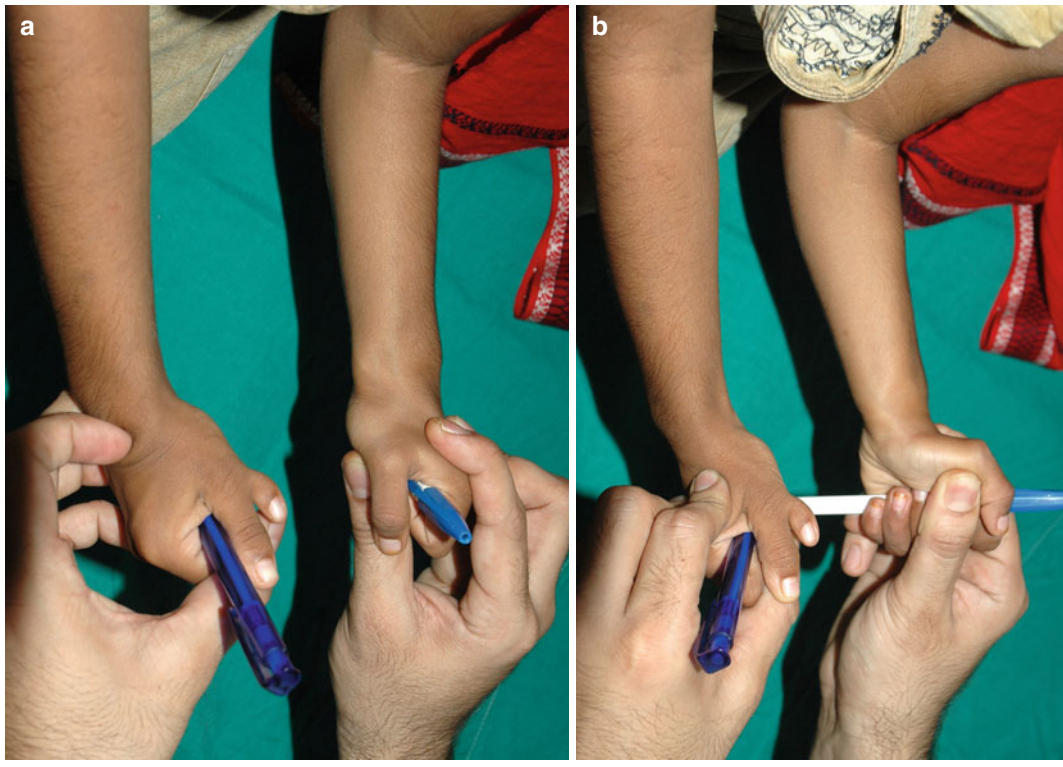

Fig. 28.7 (**a**, **b**) A clearer impression of the range motion of the forearm can be obtained if the child holds a pen or pencil in each hand

28.4 Investigations to Confirm the Diagnosis

Plain Radiographs

Anteroposterior and lateral plain radiographs of the forearm including the elbow and wrist are mandatory. The diagnosis of congenital radioulnar synostosis, radial head dislocation, malunited Monteggia fracture-dislocation, and hereditary multiple osteochondromatosis can all be conclusively made from these radiographs (Fig. 28.8a–d).

28.5 Differential Diagnosis

28.5.1 Congenital Radioulnar Synostosis

Radioulnar synostosis can occur as an isolated anomaly or in association with other skeletal anomalies or as part of a syndrome (Joseph 2000; Cleary and Omer 1985) (Table 28.1). Bilateral involvement occurs in 60 % of cases. Varying degrees of fixed pronation deformity

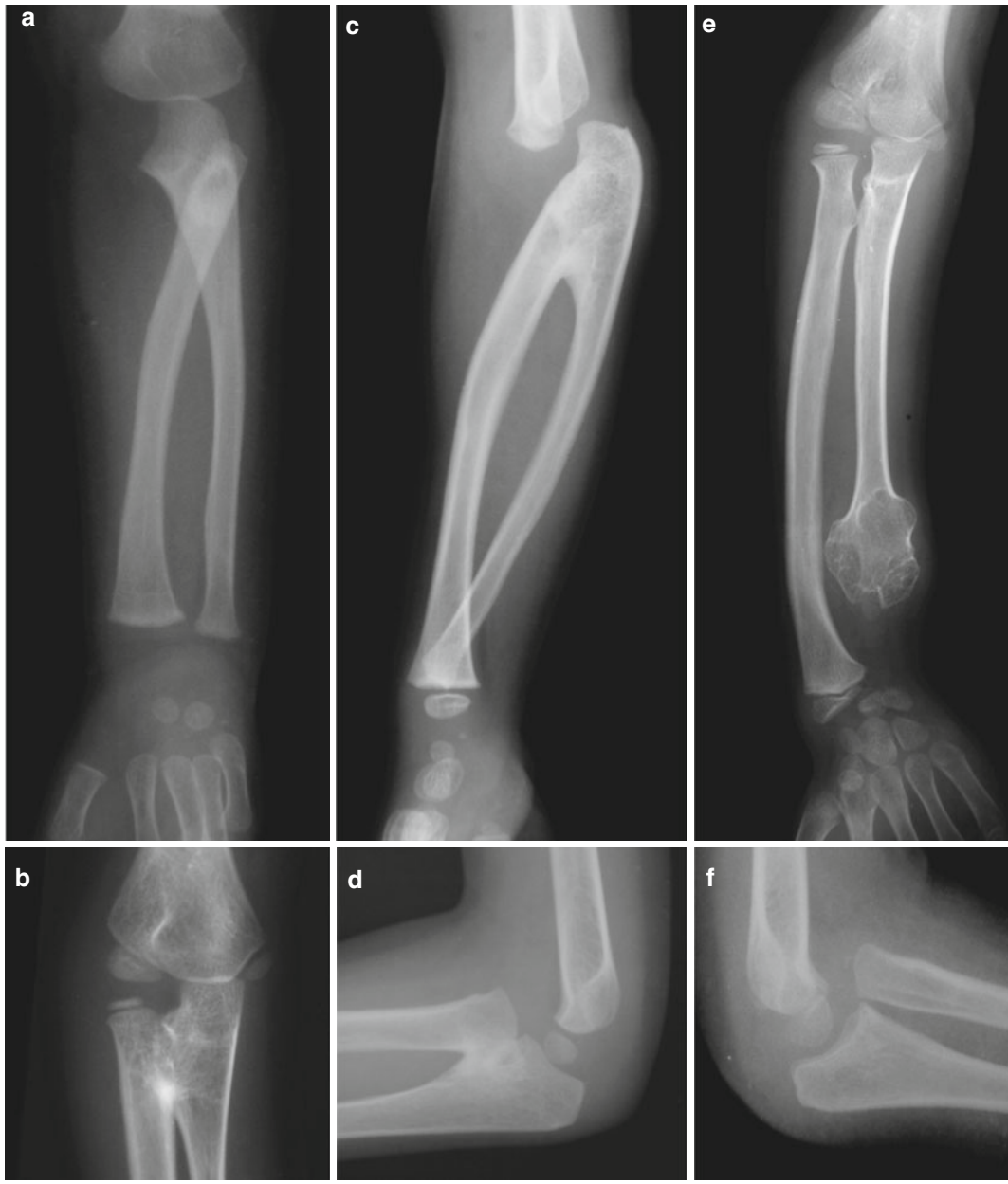

Fig. 28.8 Radiographs of the forearm and elbow of conditions that cause limitation of pronation and supination including congenital radioulnar synostosis (**a**–**c**), radial head dislocation associated with radioulnar synostosis (**d**), hereditary multiple osteochondromatosis (**e**), and radial head dislocation due to a missed Monteggia injury (**f**)

may be encountered. No movement of the radioulnar joint is possible, but exaggerated motion of the wrist and compensatory movement of the shoulder may mitigate the disability (Ogino and Hikino 1987).

28.5.2 Congenital Dislocation of the Radial Head

Congenital dislocation of the radial head is generally bilateral and may occur as an isolated entity or be part of a wide variety of syndromes

(Rizzo et al. 1997) (Almquist et al. 1969) (Table 28.1). Familial aggregation is not uncommon with both autosomal dominant and recessive patterns of inheritance. The direction of dislocation can be anterior (47 %), posterior (43 %), or lateral (10 %); isolated radial head dislocation is more frequently anterior, while those associated with syndromes are more frequently posterior (Reichenbach et al. 1995; Almquist et al. 1969). Often the dislocation is totally asymptomatic though some degree of reduction of pronation and supination is present.

Table 28.1 Skeletal anomalies and syndromes associated with congenital radioulnar synostosis and congenital dislocation of the radial head

	Congenital radioulnar synostosis	Congenital radial head dislocation
	Anomaly or syndrome	
General skeletal anomaly	Hypermobile joints	
Localized skeletal anomaly		
Upper limb anomalies		
Shoulder		Congenital elevated scapula
Elbow	Dislocation of the radial head (Fig. 28.8d)	Radioulnar synostosis Cubital pterygium
Forearm		Ulnar aplasia Below-elbow transverse deficiencies
Hand and wrist	Aplasia of the thumb Symphalangism Carpal coalition Polydactyly (Fig. 28.5) Syndactyly	Aplasia of the thumb Aplasia of ulnar ray Metacarpal fusion Side-to-side phalangeal fusion Cleft hand Thumb duplication
Lower limb anomalies		
Hip	Developmental dysplasia of the hip	
Leg	Tibial aplasia	
Foot	Clubfoot	
Associated syndromes	Acrocephalopolysyndactyly (Carpenter syndrome) Acrocephalosyndactyly (Apert syndrome) Acrofacial dysostosis Arthrogryposis Fetal alcohol syndrome Mandibulofacial dysostosis Mesomelic dysplasia Nievergelt syndrome	Acrocephalopolysyndactyly (Carpenter syndrome) Acrocephalosyndactyly (Apert syndrome) Acro-osteolysis congenita Cornelia de Lange syndrome Chondroectodermal dysplasia Craniostenosis Cleidocranial dysostosis Diastrophic dysplasia Ehlers-Danlos syndrome Larsen syndrome Nail-patella syndrome Nievergelt syndrome Mesomelic dysplasia Rubinstein-Taybi syndrome Silver-Russell syndrome
Chromosomal abnormalities	XXY XXXY XXXX	

Adapted from Joseph (2000)

28.5.3 Cerebral Palsy

Though hemiplegic cerebral palsy may often be easily diagnosed by gait abnormalities and flexion deformity of the wrist and a fixed pronation deformity of the forearm, mild forms may not have any of these typical deformities and may present as inability to perform certain activities of daily living with ease. Some limitation of supination is characteristically noted, and this is usually associated with spasticity of the flexors and pronator muscles of the forearm (Kreulen et al. 2007; Yokochi et al. 1992). The static posture of the wrist and forearm may be normal when the child is seated or recumbent. However, if the child is asked to run, the affected limb will go into flexion and pronation. A careful neurological examination will demonstrate spasticity of muscles. Involvement of the lower limb with loss of selective motor control and spasticity of muscles like the gastroc-soleus may also be noted. The affected lower limb is slightly shorter than the normal side, and the foot and the hand are also smaller.

28.5.4 Obstetric Brachial Plexus Palsy

Limitation of forearm rotation is a common sequel of obstetric brachial plexus palsy. The pattern and severity of limitation of movement of the forearm depend on the location and severity of nerve damage in the first instance (Sibinski et al. 2007; Zancolli and Zancolli 2000). The more severe the initial paralysis and the poorer the recovery of shoulder function, the more the limitation of forearm motion. There is often a disparity between the range of active and passive movements; active movements are more limited than passive movements signifying that there is persistent muscle weakness. The limitation of passive motion is largely on account of contractures of the muscles; the interosseous membrane may also become contracted in the more severe cases. Pronation contractures and supination contractures have been reported (Zancolli and Zancolli 2000).

28.5.5 Hereditary Multiple Osteochondromatosis

Growth abnormalities of bones with osteochondromata are common in children with hereditary multiple osteochondromatosis (Ishikawa et al. 2007; Matsubara et al. 2006). In the forearm, growth retardation of the ulna is usually more pronounced than that of the radius. The location of the osteochondromata seems to determine the extent and pattern of the growth aberration and even the response to treatment by tumor excision (Ishikawa et al. 2007). The degree of growth retardation of the ulna has a bearing on the limitation of pronation and supination; the shorter the ulna, the greater the limitation of forearm motion (Matsubara et al. 2006). The osteochondromata may be small in the very young child as are the growth retardation and the limitation of forearm motion.

28.5.6 Malunited Monteggia Fracture-Dislocation

Monteggia fracture-dislocations in young children may be missed since the ulna may undergo plastic deformation without an overt fracture (Kemnitz et al. 2000). The initial displacement of the radial head and the subtle bowing of the ulna may be overlooked. The child will manifest with limitation of pronation and supination later on. Full-length anteroposterior and lateral radiographs of the forearm including the wrist and the elbow will show that the radial head is dislocated. Bowing of the ulna with its apex directed toward the direction of dislocation of the radial head will be clearly evident in some instances. The ulnar bow sign which detects residual bowing of the ulna will be present in the more subtle cases (Lincoln and Mubarak 1994) (Fig. 28.9).

28.5.7 Malunited Fracture of the Forearm Bones

Malunion of fractures of the radius and ulna often results in limitation of pronation and/or

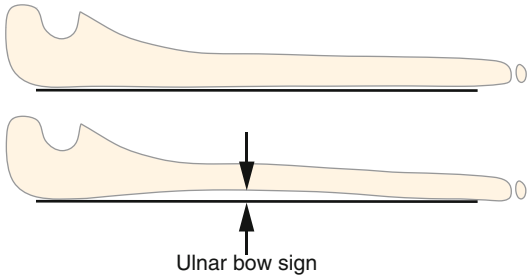

Fig. 28.9 The ulnar bow sign

supination. Angular malunion results in reduction of both pronation and supination, while rotational malunion results in an increase in one movement and reduction in the other depending on the direction of the malunion (Tynan et al. 2000). Minor degrees of malunion may not cause appreciable reduction of pronation and supination. One study based on an experimental model suggests that significant loss of pronation and supination will occur if the angular malunion is more than 14° radially, 7° ulnarward, 5° anteriorly, and 4° posteriorly (Yasutomi et al. 2002; Price et al. 1990).

28.5.8 Rare Cause of Limitation of Pronation and Supination

Osteogenesis Imperfecta Type V (See Chap. 30) (*Congenital brittle bones with redundant callus*)

This form of brittle bone disease tends to form hyperplastic callus following fractures. Radial head dislocation and calcification of the interosseous membrane may develop and will result in loss of pronation and supination (Lee et al. 2006; Glorieux et al. 2000) (Fig. 28.10). The diagnosis can be made on the basis of the history of frequent fractures following trivial trauma, tendency to form hyperplastic callus which is palpable and often has features of inflammation. Radiographs of the forearm may show heterotopic ossification in the interosseous space which is characteristic of the condition (Fig. 28.10).

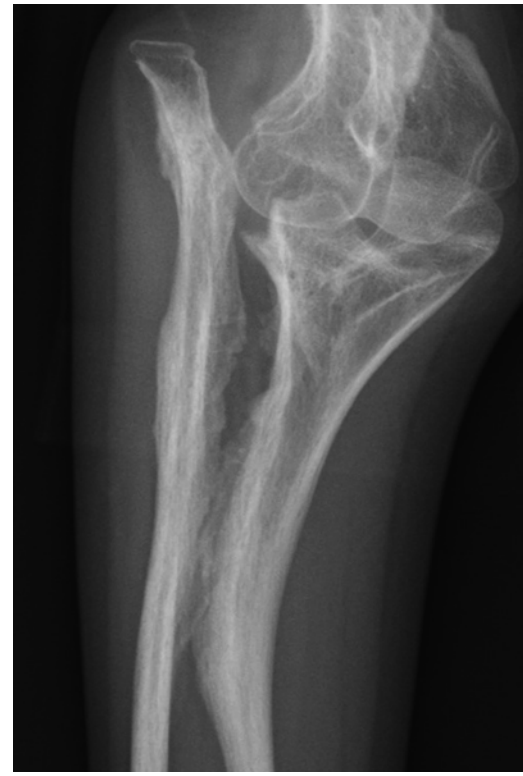

Fig. 28.10 Heterotopic ossification in the interosseous membrane in a child with osteogenesis type V

28.6 Establishing the Diagnosis

An outline of the process of establishing a diagnosis of the cause of diminished movements of a limb in the neonate is shown in Table 28.2.

Table 28.2 An outline of the process of establishing the diagnosis of diminished movement of the forearm in a young child

History					
Family history may be positive	Family history may be positive	Family history often positive	No family history	No family history	No family history
No history of trauma	No history of trauma	No history of trauma	No history of trauma	No history of trauma	History of trauma may be present
–	–	–	Developmental delay (e.g., delayed walking)	Large baby and difficult labor	–
Physical examination					
Fixed pronation deformity of forearm	Usually no fixed deformity	Usually no fixed deformity	Forearm may be in pronation	Forearm may be in pronation or supination	Usually no fixed deformity
Marked compromise of activities that require active supination	–	–	Compromise of activities that require active supination	–	–
Shoulder function normal	Shoulder function usually normal	Shoulder function usually normal	Shoulder function may be compromised (especially involving external rotation)	Shoulder function usually compromised. External rotation often limited	Shoulder function normal
Radius and ulna of normal length	Radius and ulna of normal length	Ulna short Radius bowed	Radius and ulna of normal length	Radius and ulna of normal length	Radius and ulna of normal length
Radial head may be dislocated in some cases	Radial head dislocated	Radial head may be dislocated	Radial head not dislocated	Radial head may be dislocated	Radial head dislocated
No passive or active motion possible from fixed position	Passive and active motions show equal (mild to moderate) reduction in range of motion	Passive and active motions show equal (mild to moderate) reduction in range of motion	Mild reduction of passive supination and moderate reduction of active supination Spasticity of pronator muscles	Reduction of active motion exceeds reduction of passive motion Weakness of muscles of shoulder, arm, and forearm	Passive and active motions show equal (mild to moderate) reduction in range of motion

Investigations					
Plain radiograph: Radioulnar synostosis Radial head may be dislocated in some	Plain radiograph: Radial head dislocation Ulnar bow sign negative	Plain radiograph: Ulna shorter than radius Osteochondroma on distal ulna (frequent), proximal radius or distal radius (less frequent) Radial head may be dislocated	Plain radiograph not indicated	Plain radiograph not indicated	Plain radiograph Ulnar bow sign positive and radial head dislocation confirms old Monteggia injury Negative ulnar bow sign and radial head dislocation suggest traumatic radial head dislocation
Platelet count to be done to exclude thrombocytopenia	–	–	–	–	–
Diagnosis					
Congenital radioulnar synostosis	Congenital radial head dislocation	Hereditary multiple osteochondromatosis	Hemiplegic cerebral palsy	Obstetric brachial plexus palsy	Monteggia fracture-dislocation or traumatic radial head dislocation

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Deformities and Limitation of Movement of the Wrist

29

Benjamin Joseph

29.1 Introduction

Apart from untreated congenital deformities of the wrist (described in Chap. 9) that persist, there are deformities that manifest in the young child or even later either as a consequence of growth disturbance of the growing skeleton or due to the effects of muscle imbalance. The age at which a growth-related deformity becomes apparent depends on the severity of growth inhibition. Consequently, some of the conditions mentioned in this chapter may actually manifest in the older child.

29.2 Questions to Establish a Diagnosis

- **Is there a history suggestive of obstructed labor?**
- **Is there a developmental delay?**
- **Is the forearm short?**
- **Are there bilateral symmetric deformities?**
- **Is there a history of significant trauma to the forearm?**

Is there a history suggestive of obstructed labor?

Such a history may point to obstetric brachial plexus injury which could have resulted in muscle imbalance across the wrist which in turn could have caused a paralytic deformity.

Is there a developmental delay?

A delay of motor development may suggest that the child has cerebral palsy with upper limb involvement where flexion and ulnar deviation deformities are commonly seen.

Is the forearm short?

A disproportionately short forearm is characteristic of mesomelic skeletal dysplasia and may also be seen in hereditary multiple osteochondromatosis. Shortening may also be noted in growth arrest following trauma and in paralyzed limbs.

Are there bilateral symmetric deformities?

Bilateral symmetric deformities are often seen in skeletal dysplasia.

Is there a history of significant trauma to the wrist or forearm months or years prior to noting the deformity?

A history of previous trauma may suggest that the growth plate of the radius or the ulna may have been injured. There is always a time lag between the injury and the appearance of the deformity, and parents often are aware of the progressive nature of the deformity.

29.3 Physical Examination

29.3.1 Look

Observe the wrist from the dorsal aspect, the ulnar aspect, and from the side. Note if the ulnar head is unduly prominent or if it is barely visible. Note if

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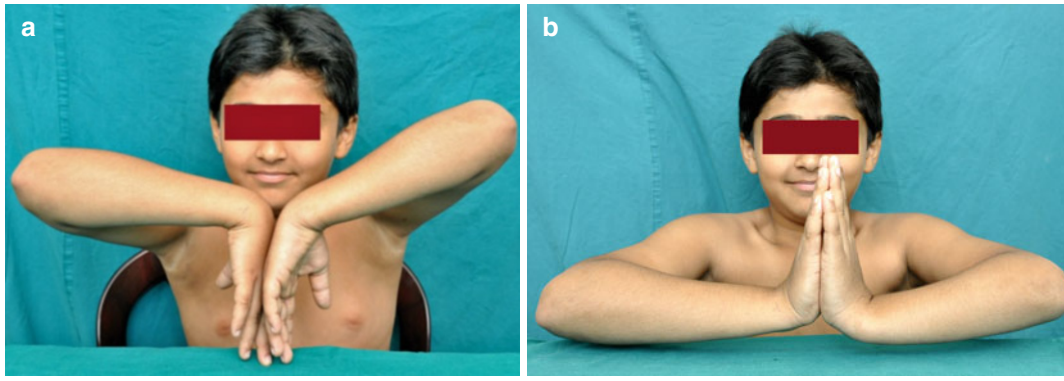

Fig. 29.1 Technique of testing for limitation of wrist flexion: ask the child to hold the dorsal surfaces of both hands together in the midline and lower the elbows as far as possible. Normally, 90° of wrist flexion should be possible such that the forearm is horizontal; flexion is restricted on the left side (**a**). Testing for limitation of wrist extension: hold both palms together in the midline and ask the child to raise the elbows as far as possible. 90° of normal wrist extension is possible on both sides (**b**)

the wrist is deviated either radially or ulnarwards and check if the forearm is short or bowed. Observe the stature of the child; if the stature is short, observe if the body proportions are altered.

29.3.2 Feel

Palpate the ulna and radius through their entire lengths; note if the relative lengths of these bones are normal, if the bones are bowed, and if there are palpable bony swellings in the metaphyseal regions of either bone. Note if the superior and inferior radioulnar joints are stable or if they are dislocated.

29.3.3 Move

Check the movements of the wrist including flexion, extension, ulnar, and radial deviation. A simple way to detect limitation of wrist flexion is to ask the child to hold the dorsal surfaces of both hands together in the midline and lower the elbows as far as possible. Normally, 90° of wrist flexion should be possible such that the forearm is horizontal (Fig. 29.1a). Similarly test if there is limitation of wrist extension by holding both palms together in the midline and ask the child to raise the elbows as far as possible. Normally 90° of wrist extension should be possible (Fig. 29.1b). Check if the ranges of pronation and supination are normal or reduced. Ninety degrees of pronation and supination should be possible from the mid-prone position (Fig. 29.2a–c). Check the tone of the muscles and test the power of the muscles crossing the wrist.

29.4 Investigations to Confirm the Diagnosis

Plain Radiographs

Plain anteroposterior and lateral radiographs of the affected wrist and forearm including the elbows are essential if a bony abnormality is suspected. Bowing of the radius, abnormal inclination of the distal articular surface of the radius, a wedged appearance of the distal radial epiphysis, and dislocation of the distal radioulnar joint may be seen when there is growth inhibition of the radius. A short ulna relative to the radius with disruption of the distal radioulnar joint is a feature of growth inhibition of the ulna. Abnormal volar or dorsal tilting of the radial articular surface may be observed in children with a Madelung deformity.

MRI Scan

If growth plate damage or a soft tissue tether to normal growth is suspected a MRI scan is indicated; these lesions can be identified on the scan.

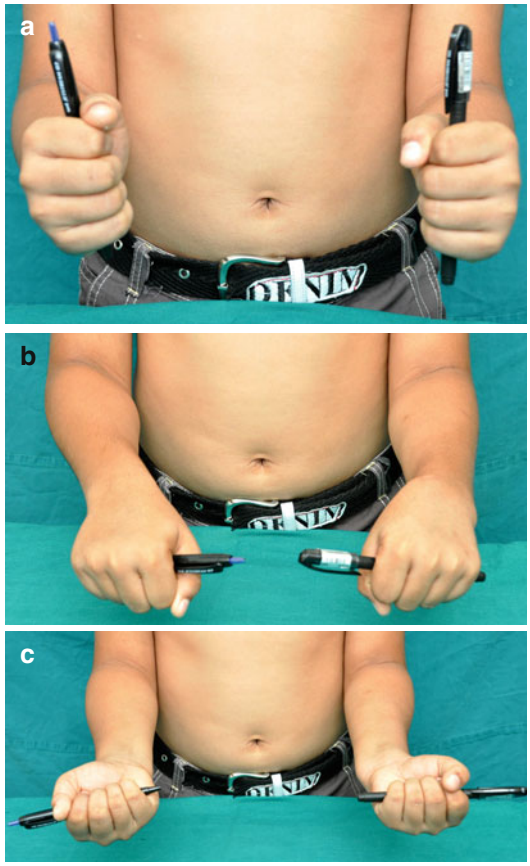

Fig. 29.2 Technique of testing the ranges of pronation and supination: 90° of pronation (**b**) and supination (**c**) should be possible from the mid-prone position (**a**)

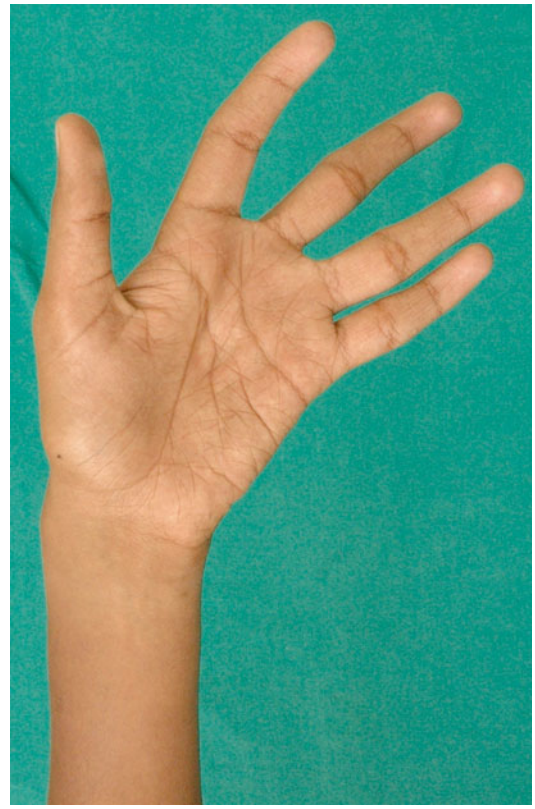

Fig. 29.3 Ulnar deviation of the wrist in a child with cerebral palsy. The flexor carpi ulnaris muscle was spastic

29.5 Differential Diagnosis

29.5.1 Obstetric Brachial Plexus Palsy

A flexion deformity of the wrist may develop in children who have paralyzed wrist extensors and functioning wrist flexors, and an extension deformity may develop if the pattern of paralysis is reversed. Ulnar deviation is also often encountered in obstetric brachial plexus palsy (Chuang et al. 2002).

29.5.2 Cerebral Palsy

Flexion and ulnar deviations of the wrist are characteristically seen in children with upper limb involvement in cerebral palsy (Carlson et al. 2006). The wrist flexion develops on account of spasticity or contracture of the wrist and finger flexors, while ulnar deviation is on account of spasticity or contracture of the flexor carpi ulnaris muscle (Fig. 29.3).

29.5.3 Hereditary Multiple Osteochondromatosis

Growth inhibition of the ulna and ulnar deviation of the wrist is a feature of hereditary multiple osteochondromatosis (Fig. 29.4). The growth inhibition appears to be related to the location of the osteochondroma on the distal ulna (Stieber and Dormans 2005; Vanhoenacker et al. 2001).

29.5.4 Skeletal Dysplasia

Mesomelic skeletal dysplasia can affect the growth of the radius or ulna with ulnar or radial

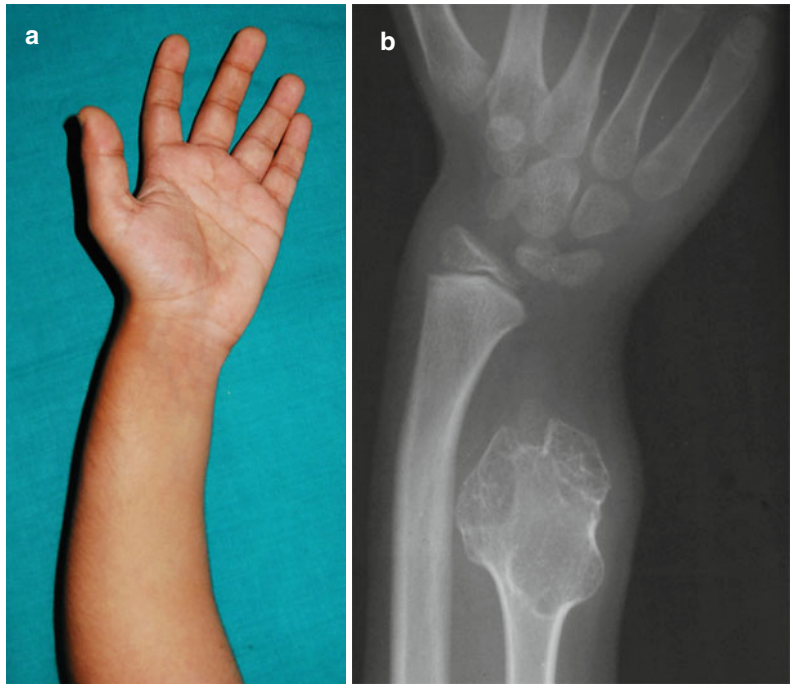

Fig. 29.4 Ulnar deviation of the wrist in a child with hereditary multiple osteochondromatosis (**a**). The radiograph shows that the ulna with an osteochondroma on the distal end is short (**b**)

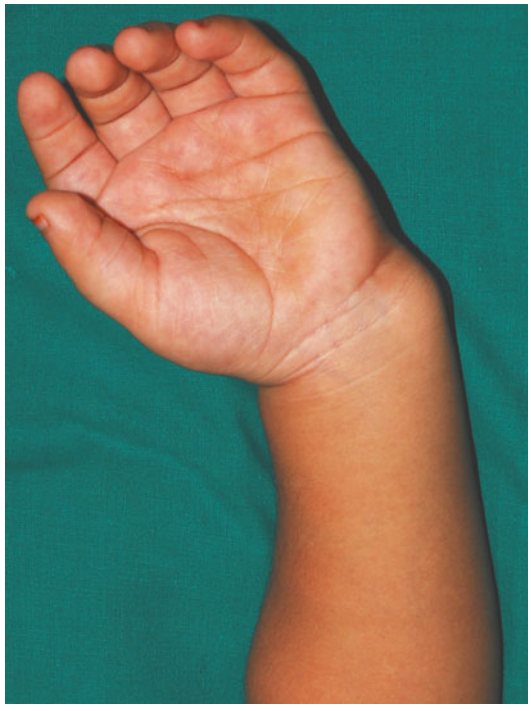

Fig. 29.5 Radial deviation of the wrist in a child with a mesomelic form of skeletal dysplasia with predominant growth inhibition of the radius. The head of the ulna is prominent, and the inferior radioulnar joint is dislocated

deviation of the wrist (Fig. 29.5). In the Reinhardt-Pfeiffer dysplasia, the ulna is short and the wrist is in ulnar deviation (Bhatia and Joseph 2000).

29.5.5 Leri-Weill Dyschondrosteosis and Turner Syndrome

Leri-Weill dyschondrosteosis is an inherited dysplasia with moderate dwarfism, short forearms, and changes in the distal radius resembling the Madelung deformity (see below) (Berdon et al. 1965; Binder et al. 2001; Tauber et al. 2004). The dwarfism and the forearm bowing is less pronounced than that seen in Langer mesomelic dysplasia (see Chap. 9). Similar wrist changes have been noted also in Turner syndrome which has other characteristic features such as gonadal dysgenesis, cubitus valgus, and short fourth metatarsals. It is now established that mutations in the short stature homeobox-containing gene (SHOX) are responsible for the wrist deformity in these distinctly separate conditions (Grigelioniene et al. 2001).

29.5.6 Madelung Deformity

The initial manifestation of the Madelung deformity is an undue prominence of the ulnar head associated with some painful limitation of wrist and forearm motion. Later deformities of the wrist may become evident (Zebala et al. 2007) (Fig. 29.6). Though the initial symptoms may only appear around 12 years of age, the underlying abnormality of the soft tissues and growth plate is present earlier and can be clearly identified on MRI scans (Cook et al. 1996). A consistent characteristic feature is the presence of a stout anomalous ligament extending from the lunate to the metaphysis of the radius. In addition, the volar radiotriquetral and the short radiolunate ligament may be hypertrophied (Cook et al. 1996). Narrowing of the physis or a physeal bar bridging the distal metaphysis of the radius to the epiphysis is seen later. It is presumed that the anomalous ligament acts as a tether leading to the development of the physeal bar (Vickers 2000). Since there is a strong genetic basis for this condition, a family history is often present. Children of an affected parent may benefit from early imaging and surgical intervention as soon

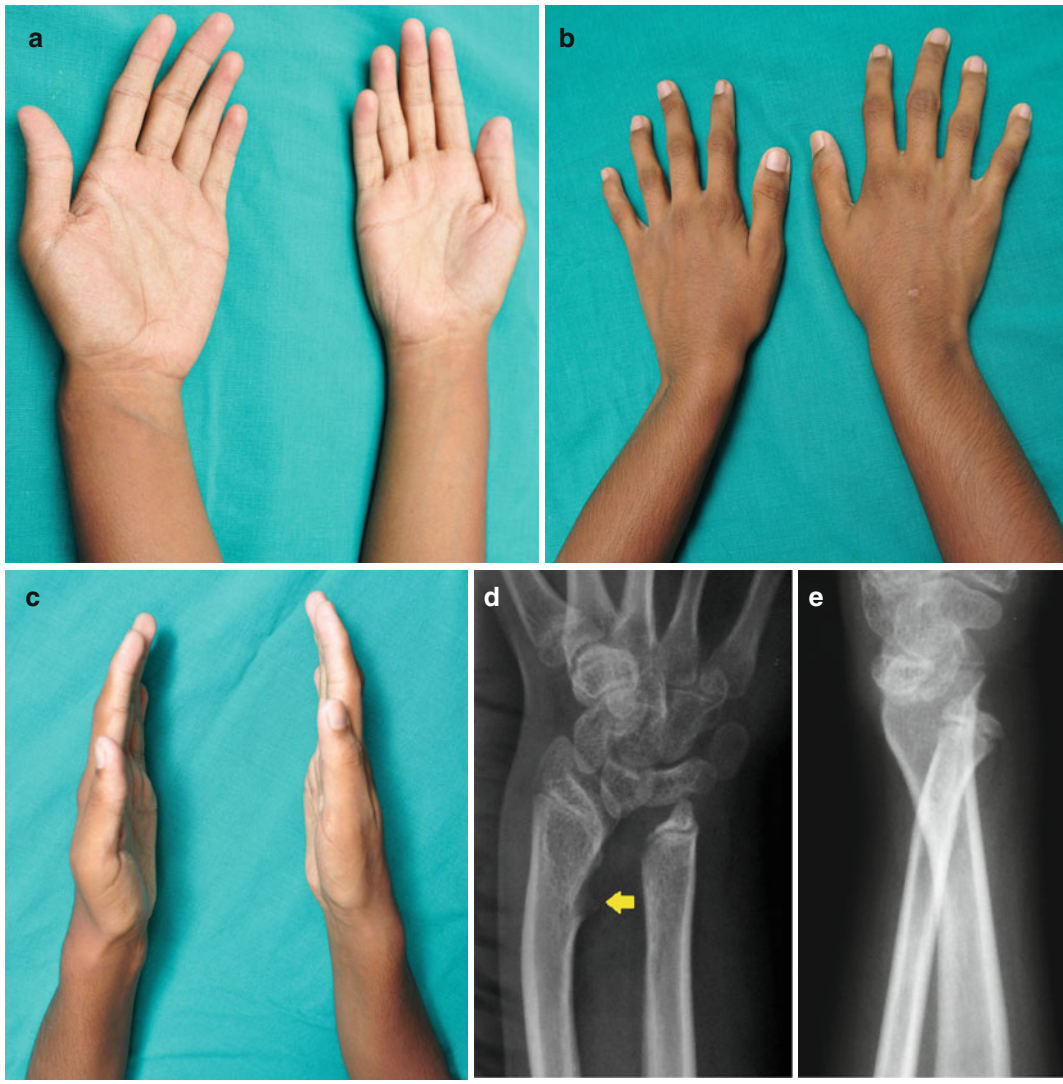

Fig. 29.6 Appearance of the wrist of a girl with Madelung deformity (**a**–**c**); the left wrist is more severely affected. Radiograph of the wrist (**d**, **e**) shows the characteristic features; they include the abnormal inclination of the radial articular surface, inferior radioulnar dislocation, and the site of attachment of the anomalous ligament (*arrow*)

Table 29.1 Site of physeal bar in Madelung deformities

Type	Deformity	Site of physeal bar
Madelung deformity	Ulnar and volar tilt of radial articular surface Dorsal prominence of ulnar head Instability of inferior radioulnar joint	Ulnar and volar part of the distal radial physis
Reverse Madelung deformity	Ulnar and dorsal tilt of radial articular surface Volar prominence of ulnar head Instability of inferior radioulnar joint	Ulnar and dorsal part of the distal radial physis
Chevron carpus	No deformity of the wrist Wedge-shaped carpus Stable inferior radioulnar joint	Ulnar and central part of the distal radial physis

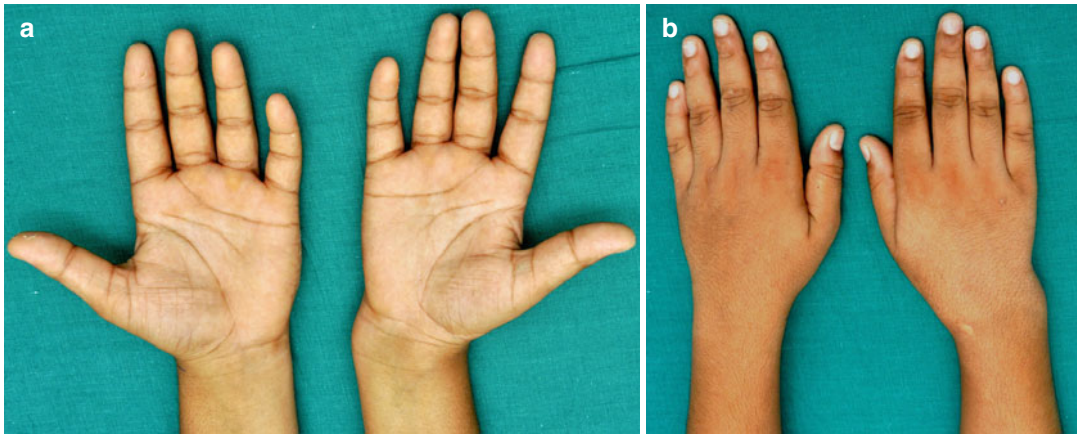

Fig. 29.7 (**a**, **b**) Posttraumatic growth arrest of the distal radial physis leading to a valgus deformity

as the physeal bar appears (Vickers 2000). The site of the physeal arrest can vary, and accordingly the wrist deformity varies (Table 29.1). Accurate identification of the site of the physeal bar is imperative if surgery is being planned.

29.5.7 Growth Arrest of Distal Radial Physis

Premature arrest of growth at the distal radial physis can occur following trauma or infection; the linear growth of the radius is retarded, while the ulna continues to grow. A valgus deformity will develop and progress (Fig. 29.7).

29.5.8 Rare Conditions

Wrist deformities may occur in Ollier's disease and dysplasia epiphysealis hemimelica (Trevor's disease) (Oestreich et al. 2002).

29.6 Establishing the Diagnosis

An outline of establishing the cause of ulnar or radial deviation deformity of the wrist is shown in Table 29.2, and the outline of diagnosis of Madelung deformity and its variants is shown in Table 29.3.

Table 29.2 An outline of the process of establishing the diagnosis of the cause of radial or ulnar deviation of the wrist in a child

History						
No family history	No family history	No family history	Family history often present	Family history often present	No family history	Family history often present
Developmental delay	Difficult labor	History of trauma months or years earlier	–	–	History of trauma months or years earlier	–
Physical examination						
Ulnar deviation of wrist	Ulnar deviation of wrist	Ulnar deviation of wrist	Ulnar deviation of wrist	Ulnar deviation of wrist	Radial deviation of wrist	Radial deviation of wrist
Relative lengths of radius and ulna normal	Relative lengths of radius and ulna normal	Ulna short relative to radius	Ulna short relative to radius	Ulna short relative to radius	Radius short relative to ulna	Radius short relative to ulna
Forearm slightly shorter than on normal side	Forearm shorter than normal side	Forearm length normal	Forearm short on both sides	Forearm short	Forearm length normal	Forearm short on both sides
Normal stature	Normal stature	Normal stature	Proportionate mild short stature	Disproportionate dwarfism	Normal stature	Disproportionate dwarfism
–	–	–	Osteochondromata present	–	–	–
Spasticity of flexor carpi ulnaris	Muscle weakness and muscle imbalance	–	–	–	–	–
Investigations						
No specific investigation indicated	No specific investigation indicated	Plain radiograph	Plain radiograph	Plain radiograph and skeletal survey	Plain radiograph	Plain radiograph and skeletal survey
Diagnosis						
Cerebral palsy	Obstetric brachial plexus palsy	Traumatic growth arrest of distal ulna	Hereditary multiple osteochondromatosis	Mesomelic dysplasia with predominance of ulnar involvement	Traumatic growth arrest of distal radius	Mesomelic dysplasia with predominance of radial involvement

Table 29.3 Diagnosis of Madelung deformity and its variants

History		
Usually prominence of ulna first noted around the age of 12 years	Dwarfism and forearm deformity noted at birth	Wrist deformity noted around the age of 12 years
Wrist pain is often an early symptom	Pain is not an early symptom	Pain seldom an early symptom
Physical examination		
Stature normal	Moderate dwarfism	Some stunting of growth
Elbow normal	Elbow normal	Cubitus valgus
Forearm length near normal	Forearm short	Forearm length near normal
No relative shortening of fourth metatarsal	No relative shortening of fourth metatarsal	Short fourth metatarsals
–	–	Cardiac and renal anomalies common
–	–	Delayed sexual maturation due to gonadal dysgenesis
Investigations		
Plain radiograph	Plain radiograph and skeletal survey	Plain radiograph
–	–	Chromosomal studies
Diagnosis		
Isolated Madelung deformity	Leri-Weill dyschondrosteosis	Wrist changes in Turner syndrome

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Brittle Bones and Frequent Fractures

30

Benjamin Joseph

30.1 Introduction

Frequent fractures following trivial trauma or during normal daily activities occur in a group of conditions where the bones are brittle. The most common of these conditions is osteogenesis imperfecta (Plotkin 2004). Various types of osteogenesis imperfecta have been described; the severity, the frequency of fractures, and other clinical features vary in each of the types (Sillence et al. 1979; Sillence 1988; Glorieux et al. 2000).

All conditions with congenital brittle bones result from genetic abnormalities (Plotkin 2004). In most cases of osteogenesis imperfecta, the underlying genetic abnormality is a mutation in one of the type I procollagen genes (COL1A1 and COL1A2); in the rarer forms of osteogenesis imperfecta, these genes are normal (Plotkin 2004; Sillence 1988). Plotkin suggests that brittle-bone disease with mutations of the procollagen genes alone should be considered as true osteogenesis imperfecta (OI) and all other conditions be grouped as "syndromes resembling osteogenesis imperfecta" (SROI). The author has used the classification suggested by Plotkin in this chapter.

Another important, though unfortunate, cause of repeated fractures is non-accidental injury (NAI). It is vital that the clinician tries to differentiate NAI from brittle-bone disease, but this may be exceedingly difficult to do, and it is possible that the two may coexist (Chapman and Hall 1997; Kasim et al. 1995; Smith 1995; Marlowe et al. 2002; Ablin and Sane 1997; Steiner et al. 1996; Bulloch et al. 2000).

30.2 Questions to Establish a Diagnosis

- **Is there a positive family history of similar symptoms?**
- **Is the sclera blue, the dentition normal, are there features of generalized ligament laxity, and is the stature normal?**
- **What is the pattern of fractures (which bones and the location of fractures in the bones)?**
- **Are there features suggestive of non-accidental injury?**

Is there a positive family history of similar symptoms?

A positive family history of bone fragility and repeated fractures would suggest that there is an underlying genetic abnormality. Pedigree charting may give some insight into the inheritance pattern. However, a family history may not be present in several instances of genetically determined brittle-bone disease.

Is the sclera blue, the dentition normal, are there features of generalized ligament laxity, and is the stature normal?

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_30

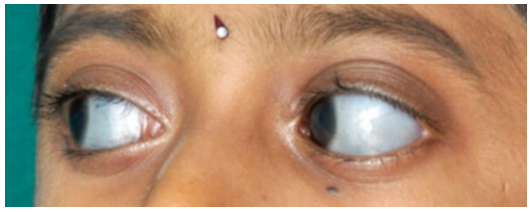

Fig. 30.1 Blue sclera in a child with osteogenesis imperfecta

Blue sclera (Fig. 30.1), abnormal dentition, generalized ligament laxity, and short stature are seen in some, but not all, forms of osteogenesis imperfecta.

What is the pattern of fractures (which bones and the location of fractures in the bones)?

Fractures in osteogenesis usually involve the diaphysis of the long bones of the extremities often leading to bowing of the limbs (Fig. 30.2a–d).

Fractures in non-accidental injury are often in the metaphyseal region of the long bones (Fig. 30.3a–c) and the posterior part of the ribs (Fig. 30.3d). Fractures in different stages of healing may be seen (Fig. 30.3e, f), and this finding has been regarded as highly presumptive of non-accidental injury.

Are there features in the history and physical examination that are suggestive of non-accidental injury?

Though often it may be difficult to establish with any degree of certainty that the fracture is the result of non-accidental injury, the features in the history that point to this diagnosis are listed here (Loder and Bookout 1991; Carty 1993).

- The alleged mechanism of injury that the parents describe does not match the severity of the injury.
- The parents or guardians give conflicting histories, or there are inconsistencies in the history when repeated.
- There is an unexplained delay in seeking medical attention.
- There is a record of poor compliance with medical advice on a previous occasion.

The features that suggest non-accidental injury on physical examination include:

- Failure to thrive.
- Soft tissue bruising or injuries especially on the face, perineum, and genitalia, sharply demarcated round marks suggestive of cigarette burns, and linear ecchymoses are all highly suggestive of non-accidental injury.

30.3 Physical Examination

30.3.1 Look

Measure the height of the child, and plot the height on a gender-specific growth chart for children of the same ethnic group. Examine the shape of the head and see if it is disproportionately large and note the shape of the face. A relatively large head and a distinctive triangular facial shape are often seen in osteogenesis imperfecta.

Look for features of malnourishment and soft tissue injuries that may suggest that the injury is inflicted. Look for microcephaly, proptosis, joint deformities, and short femora and humeri; these may be seen in some types of syndromes resembling osteogenesis imperfecta (SROI).

30.3.2 Feel

Feel the bones of the limbs to see if they are angulated, bowed, or flattened; these features may often be seen in the more severe forms of osteogenesis imperfecta. Palpate the radial head and determine if it is dislocated; radial head dislocation is characteristic of a form of SROI that has been labeled as type V OI. Another feature of this form of SROI is hyperplastic callus at the site of previous fractures. This may be palpated as a hard, painful, and warm swelling over the bone sometimes leading to an erroneous diagnosis of osteomyelitis or osteosarcoma.

30.3.3 Move

Test for ligament laxity as shown in Fig. 23.3. Joint laxity or hypermobility of the joints of the extremities may be seen in osteogenesis imperfecta.

Passively move the joints if there are obvious deformities to confirm the presence of joint contractures that may be seen in Bruck syndrome, a

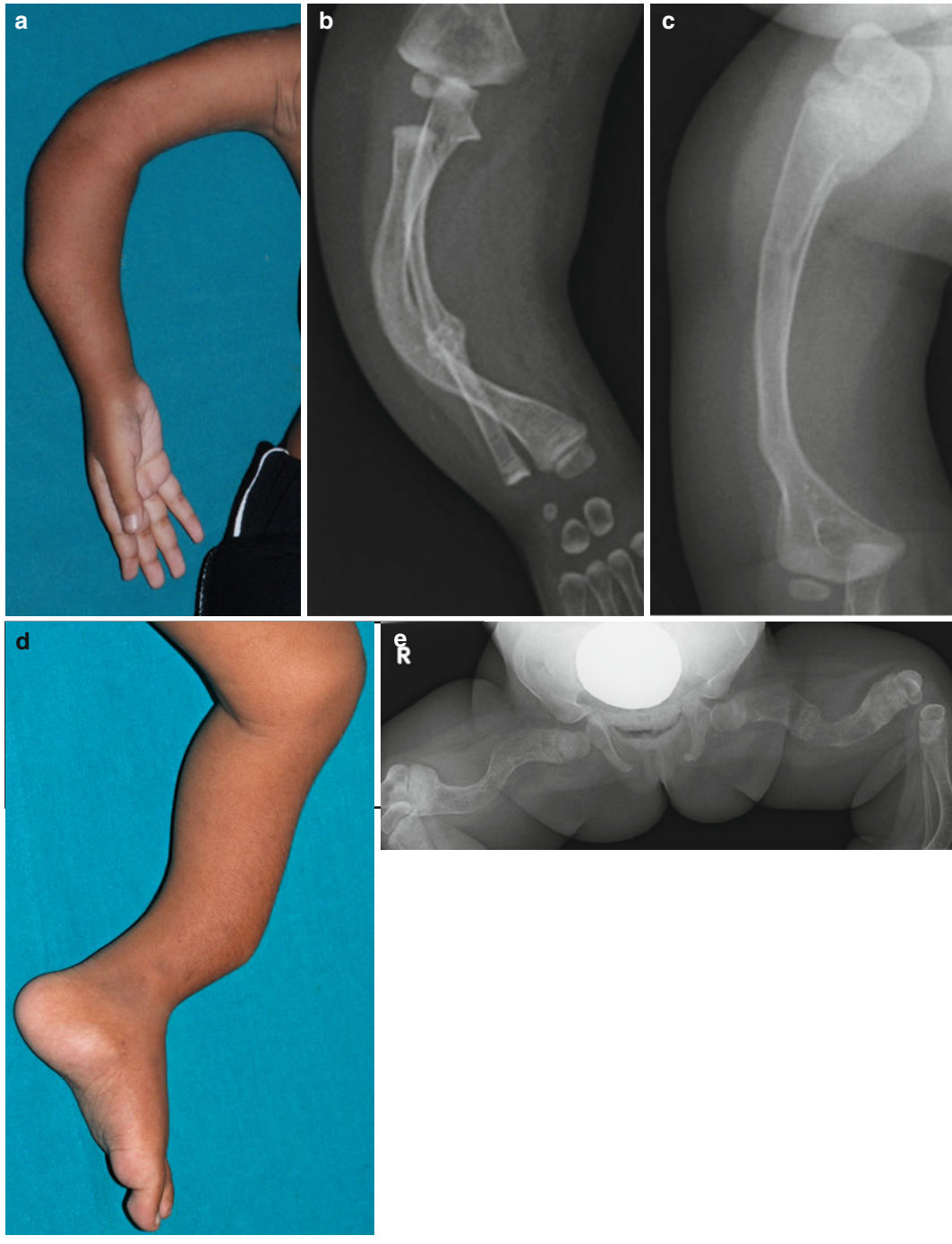

Fig. 30.2 Examples (**a**–**e**) of bowing of the lower and upper extremities following repeated fractures in children with osteogenesis imperfecta

form of SROI (Viljoen et al. 1989; Leroy et al. 1998). Limitation of motion of the radioulnar joints is a characteristic feature of the form of SROI also referred to as type V OI (Glorieux et al. 2000).

30.3.4 Special Tests

Ophthalmological examination including examination of the fundus is essential to exclude cataracts, optic atrophy, and pseudoglioma as these features may be seen in types of SROI.

30.4 Investigations to Confirm the Diagnosis

30.4.1 Plain Radiographs

Osteogenesis Imperfecta

Fractures in Osteogenesis Imperfecta

Most fractures occur in the diaphysis of the long bones. Metaphyseal fractures are very rare, but they can occur.

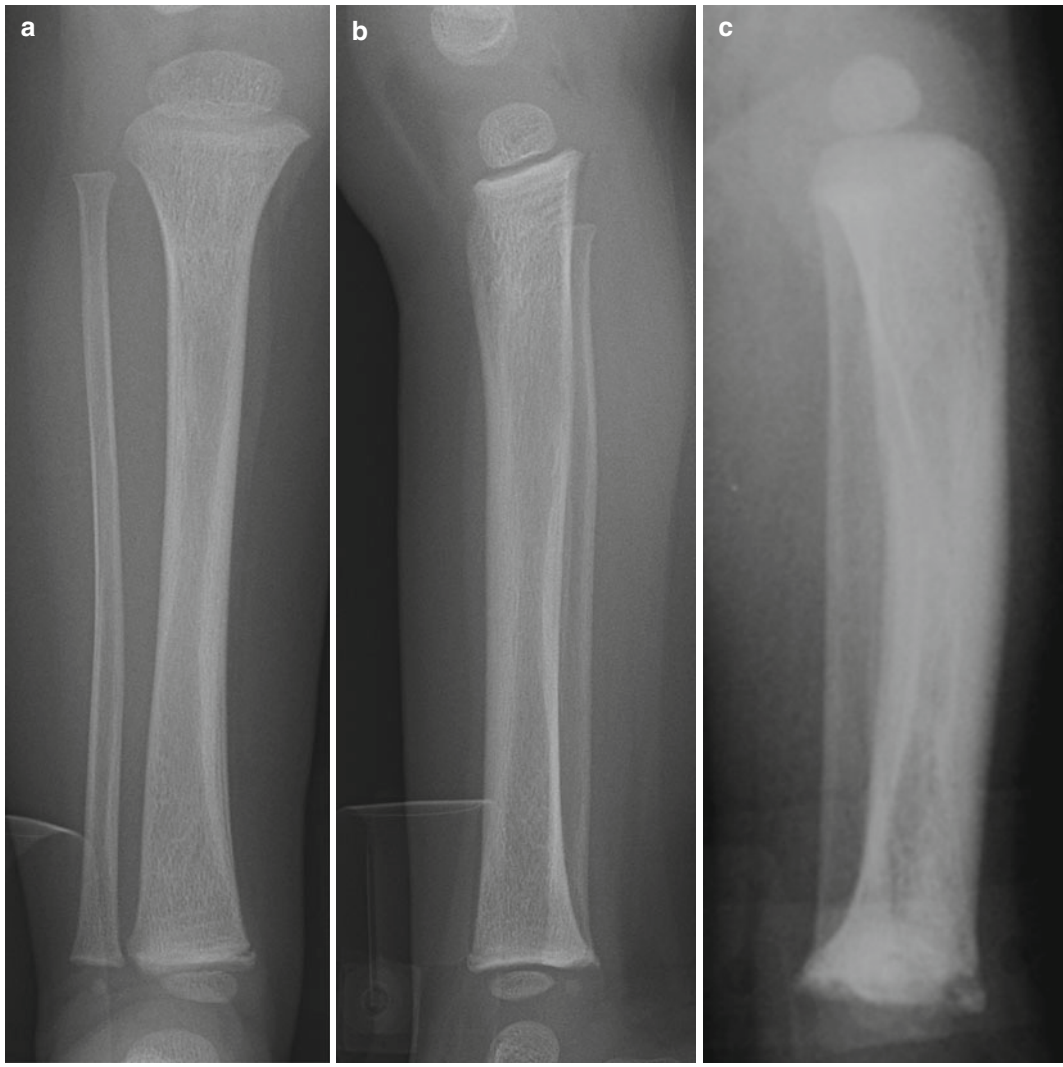

Fig. 30.3 Metaphyseal fractures and fractures in different stages of healing are frequently seen in non-accidental injury. An acute distal tibial metaphyseal corner fracture (**a**, **b**), a distal tibial metaphyseal fracture that is healing (**c**), rib fractures (**d**), and a healed proximal tibial fracture (**e**, **f**) in non-accidental injury

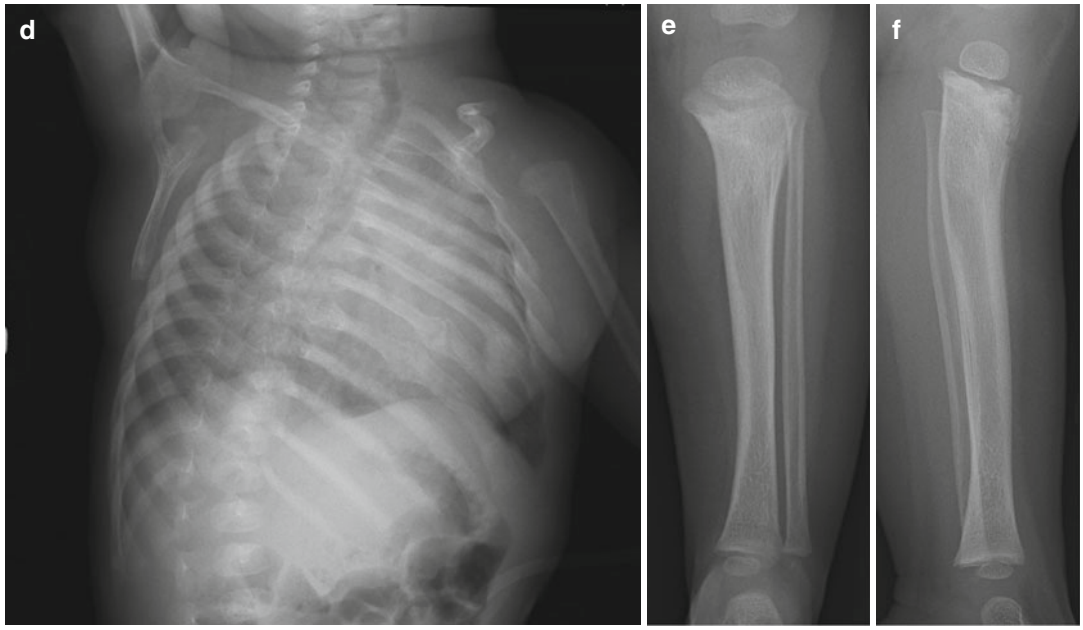

Fig. 30.3 (continued)

Vertebral fractures may occur in severe forms of osteogenesis.

Rib fractures are seen in the most severe forms of osteogenesis imperfecta.

Skull fractures are not usually seen.

Associated Features

Osteopenia is characteristically present, and bowing of the long bones of the extremities is a very common feature.

Wormian (intra-sutural) bones are often present in the skull, and this is considered a feature of osteogenesis imperfecta (Fig. 30.4). They occur more frequently in the more severe forms of the disease (Semler et al. 2010).

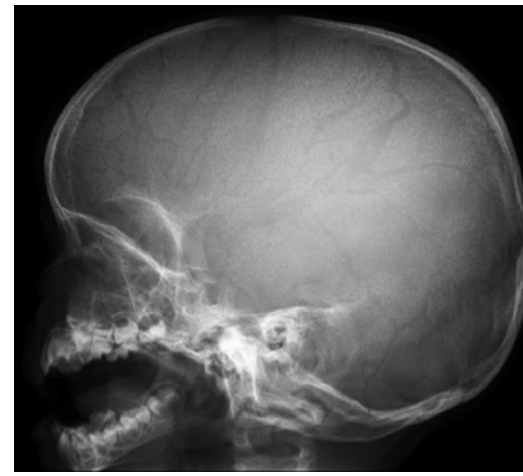

Fig. 30.4 Radiograph of the skull of a child with osteogenesis showing wormian bones

Non-accidental Injury

If non-accidental injury is even remotely suspected from the history and interaction with the child and the caregivers, a skeletal survey is mandatory. This would include the following radiographs:

- AP and lateral of the skull (a Townes view may be included if an occipital fracture is suspected)
- Lateral cervical and thoracolumbar spine
- Chest radiograph
- Abdomen
- Left and right oblique views of the ribs
- Both the humeri, forearms, and hands
- Both the femora, tibia, and fibula
- Dorsoplantar views of the feet

Fractures in Non-accidental Injury

Metaphyseal fractures of the long bones are most frequently seen, and they often have a characteristic "bucket-handle" appearance. They are seen in children less than 2 years of age and may involve the distal femur, the proximal tibia, the distal tibia, or the proximal humerus. Diaphyseal fractures are often spiral. Epiphyseal separations are not infrequent.

Rib fractures are frequently seen, and they are commonly present in the posterior part of the ribs. Injuries of the costochondral junctions may also be seen. There may be overlying bruising. Rib fractures may be missed, and so it may be prudent to repeat the radiograph after 2 weeks, by which time the healing fractures would be clearly evident.

Fractures of the scapula and spinous processes of the vertebrae are also highly suggestive of non-accidental injuries.

Skull fractures are often seen, and they are frequently depressed fractures involving the bones other than the parietal bone (parietal bone fractures are more frequently seen after accidental injury). Fractures that cross sutures or are bilateral are suggestive of non-accidental injury. Subconjunctival hemorrhages may be present if the child has been shaken violently.

Characteristically fractures in different stages of healing (a synchronous fractures) will be present if the child has been subjected to abuse for some time. However, this will not be present if the child is seen after the first episode of non-accidental injury.

Associated Features

Osteopenia is usually not seen unless the child is malnourished.

30.4.2 CT Scan/MRI

If non-accidental injury is strongly suspected, imaging of the skull and brain may be needed to reveal intracranial hemorrhage.

30.5 Differential Diagnosis

30.5.1 Osteogenesis Imperfecta

The four types of osteogenesis imperfecta (Types I–IV) vary in severity; the clinical features of the four types are listed in Table 30.1.

30.5.2 Syndromes Resembling OI (SROI)

In all these syndromes, no mutations in COL1A1 and COL1A2 have been detected so far

- Congenital brittle bones with redundant callus
- Congenital brittle bones with mineralization defect
- Congenital brittle bones with rhizomelia
- Congenital brittle bones with craniostenosis and ocular proptosis
- Congenital brittle bones with congenital joint contractures
- Osteoporosis-pseudoglioma syndrome
- Congenital brittle bones with optic atrophy, retinopathy, and severe psychomotor retardation
- Congenital brittle bones with microcephaly and cataracts

Congenital Brittle Bones with Redundant Callus

Originally this form of brittle-bone disease was designated as Type V OI (Glorieux et al. 2000). The characteristic feature of this syndrome is that hyperplastic callus forms when the fractures heal (Fig. 30.5a). Rapid initial growth of the mass of callus which is associated with pain and warmth may cause the clinician to suspect infection or sarcomatous change. The swelling may remain for several years. Radial head dislocation and calcification of the interosseous membrane between the radius and ulna and consequent inability to pronate or supinate the forearm may be present (Fig. 30.5b).

Table 30.1 Features of the four common types of osteogenesis imperfecta (listed in order of severity from left to right)

Type I OI	Type IV OI	Type III OI	Type II OI
Severity: Mild to moderate	*Severity*: Moderate	*Severity*: Severe	*Severity*: Lethal frequently Very few survive more than a few days
Stature: Normal often	*Stature*: Short	*Stature*: Very short	*Stature*: Very short
Fracture frequency: Variable (few to frequent)	*Fracture frequency*: Frequent	*Fracture frequency*: Very frequent Fractures may be present at birth	*Fracture frequency*: Intrauterine fractures invariable
Ambulatory status: Usually fully ambulant	*Ambulatory status*: Generally able to ambulate with or without walking aids	*Ambulatory status*: Frequently wheelchair bound (few walk with walking aids)	*Ambulatory status*: The few that survive are always wheelchair bound
Face: May be normal or triangular	*Face*: Variable	*Face*: Triangular	*Face*: Triangular
Sclera: Most have blue sclera	*Sclera*: White usually Blue sclera may be present at birth and fade with time	*Sclera*: Blue sclera may be present at birth and fade with time	*Sclera*: Variable
Dentition: Dentinogenesis imperfecta (DI) not common but may be present	*Dentition*: DI present in one group DI absent in one group	*Dentition*: DI common	*Dentition*: May not survive long enough to evaluate teeth
Bowing of the long bones: Usually not seen	*Bowing of the long bones*: Often present	*Bowing of the long bones*: Present and severe	*Bowing of the long bones*: Present and severe
Spine: Usually not affected	*Spine*: Vertebral fractures Scoliosis may be present	*Spine*: Vertebral fractures Scoliosis frequent	*Spine*: Vertebral fractures Scoliosis frequent

Congenital Brittle Bones with Mineralization Defect

This condition, initially designated as Type VI OI, is clinically indistinguishable from moderate or severe forms of osteogenesis imperfecta (Glorieux et al. 2002). The diagnosis currently can be made only on the basis of bone biopsy.

Congenital Brittle Bones with Rhizomelia

Originally designated as Type VII OI, this syndrome is characterized by frequent fractures and short femora and humeri.

Congenital Brittle Bones with Craniostenosis and Ocular Proptosis (Cole-Carpenter Syndrome)

There are very few reports of this syndrome in the literature. Multiple metaphyseal fractures in early childhood, hydrocephalus, craniostenosis, and very short stature are features of this syndrome (Cole and Carpenter 1987).

Congenital Brittle Bones with Congenital Joint Contractures (Bruck Syndrome)

The children are born with joint contractures including popliteal pterygia. Multiple fractures occur on account of brittle bones (Leroy et al. 1998; Viljoen et al. 1989). The radial head may be dislocated, and wormian bones may be present.

Osteoporosis-Pseudoglioma Syndrome

This syndrome has been described in a few families (Beighton 1986). The children have features of mild to moderate fragility of bones and blindness due to vitreous hyperplasia and secondary glaucoma.

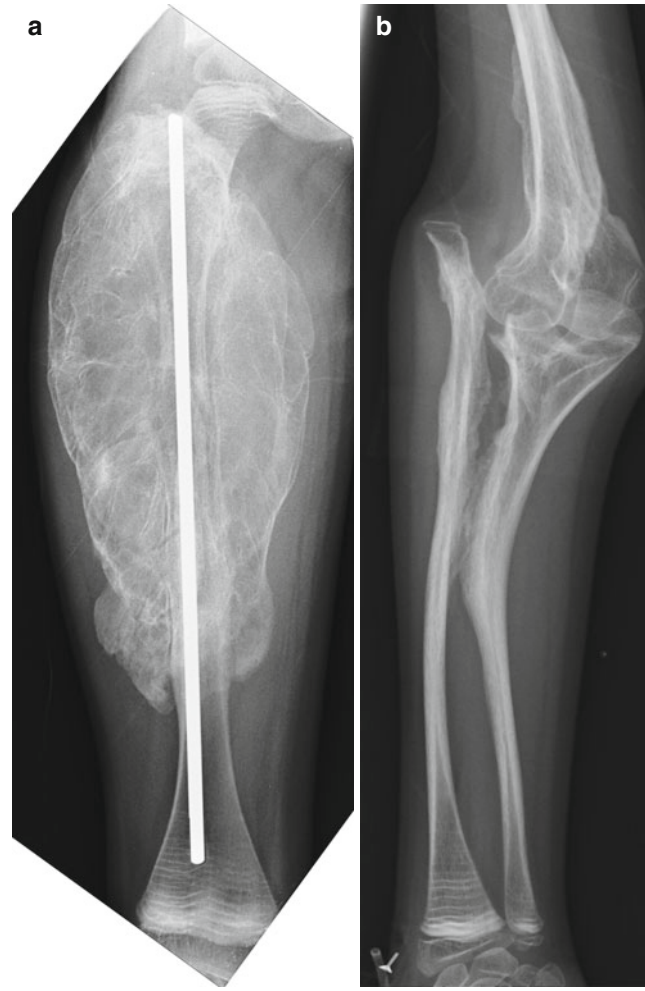

Fig. 30.5 Hyperplastic callus that developed after intramedullary rodding of the femur in a child with brittle-bone disease with redundant callus (**a**). The child also had bilateral radial head dislocation and ossification of the interosseous membrane of the forearm (**b**). Courtesy: Professor Sharaf Ibrahim, Malaysia

Congenital Brittle Bones with Optic Atrophy, Retinopathy, and Psychomotor Retardation and Congenital Brittle Bones with Microcephaly and Cataracts

These are two rare forms of brittle bone disease with ocular involvement (al Gazali et al. 1994).

30.5.3 Osteopetrosis and Pycnodysostosis

Osteopetrosis and pycnodysostosis are inherited conditions with brittle bones. These conditions are characterized by bone that is densely sclerotic unlike osteogenesis and syndromes resembling osteogenesis where the hallmark is osteopenia. The frequency of fractures is less than that encountered in osteogenesis imperfecta. Fracture healing may be delayed in osteopetrosis and pycnodysostosis.

Pycnodysostosis, an autosomal recessive condition with short stature, brittle bones, short fingers, and open fontanelles, is very similar to osteopetrosis. Acro-osteolysis involving the terminal phalanges is a characteristic distinguishing feature (Fig. 30.6).

30.5.4 Metabolic Disorders Leading to Multiple Fractures

Vitamin D Deficiency

Vitamin D deficiency leading to rickets in the premature infant and toddler can occasionally

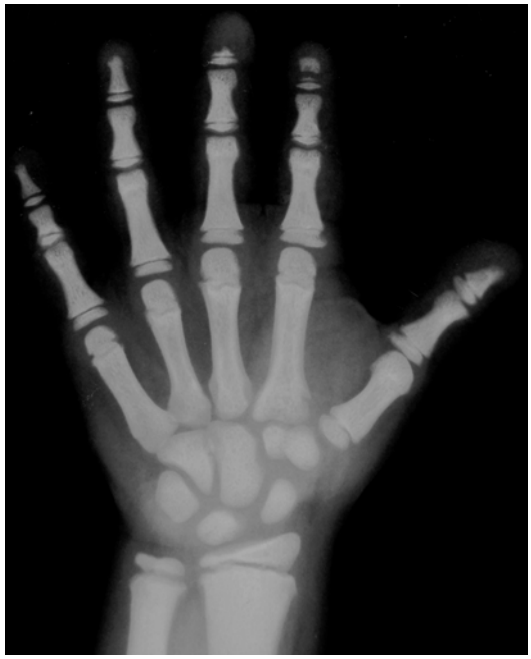

Fig. 30.6 Sclerotic bone and acro-osteolysis of the terminal phalanges in pycnodysostosis

present with repeated fractures (Keller and Barnes 2008). Estimation of serum levels of calcium, phosphates, and vitamin D and radiologic changes in the growth plates and metaphyses of the long bones will help in establishing the diagnosis.

Copper Deficiency

Copper deficiency also has been implicated as a cause of multiple fractures (Chapman 1987). Nutritional deficiency of copper can occur particularly in premature and malnourished infants. If the copper deficiency is severe enough to cause fractures, psychomotor retardation and hypotonia will be present. Measurement of serum copper and ceruloplasmin levels are needed to establish the diagnosis of copper deficiency. Menkes kinky hair syndrome is an x-linked recessive condition associated with copper deficiency. Decreased muscle tone, seizures, developmental delay, and brittle fuzzy sparse hair are the characteristics of this condition.

Vitamin C Deficiency

Scurvy, a deficiency of ascorbic acid, may present with multiple fractures including metaphyseal fractures that may resemble the corner fractures seen in non-accidental injury. Plasma ascorbic acid levels may be estimated.

30.6 Establishing the Diagnosis

The orthopedic surgeon who is confronted with a child with multiple fractures must be aware of the various conditions that may cause such fractures to distinguish them from non-accidental injury. An attempt has been made to include most of these conditions in this chapter. In addition, a clear knowledge of the spectrum of fracture patterns encountered in non-accidental injury is essential (Fig. 30.7). The summary of the pattern of fractures provided in this chapter may be supplemented by the detailed reviews of Loder and Bookout (1991) and Carty (1993) when confronted with a suspected case of non-accidental injury.

An outline of establishing the diagnosis of the underlying cause of repeated fractures in children is shown in Table 30.2.

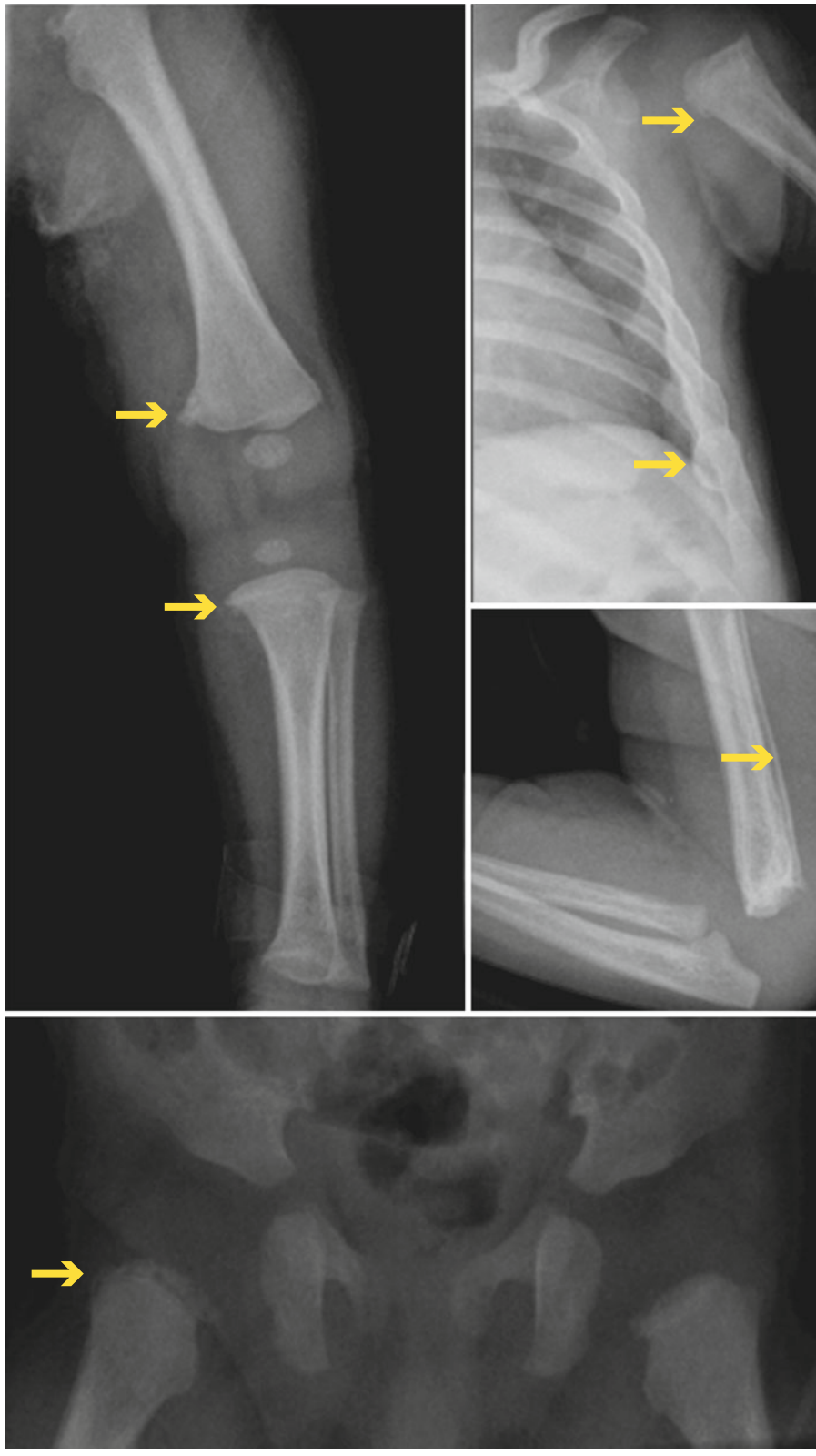

Fig. 30.7 Pattern of fractures seen in a child who suffered non-accidental injuries. *Arrows* point to the various fractures seen on the radiographs

Table 30.2 Establishing the diagnosis of frequent fractures in toddlers and young children

History		
Family history of brittle bones may be positive	No family history	Family history of brittle bones may be positive
Cause of alleged injury consistently stated	Discrepancies in the history regarding the cause of the alleged injury	Cause of alleged injury consistently stated
Usually no delay in seeking treatment	Delay in reporting to hospital following the trauma	Usually no delay in seeking treatment
Good compliance with treatment in the past	Previous history of noncompliance with treatment recommendation	Good compliance with treatment in the past
Physical examination		
Short stature, large head, triangular face, blue sclera, hypermobile joints	No abnormal shape of the skull or face, no blue sclera	Face and skull normal, no blue sclera
Bowing of the femur, tibia, humerus, or forearm	No bowing of the long bones	Bowing of the long bones
No failure to thrive	Failure to thrive	No failure to thrive
No soft tissue injuries	Burn marks or soft tissue injuries in one or more of these regions: Face Perineum Genitalia	No soft tissue injuries
–	–	Microcephaly, proptosis, joint deformities with contractures, disproportionately short humeri and femora, limited motion of the radioulnar joints, dislocated radial heads, or palpable bony swellings in the region of previous fractures
Working diagnosis: Osteogenesis imperfecta	Working diagnosis: Non-accidental injury	Working diagnosis: A syndrome resembling osteogenesis imperfecta
Investigations		
Plain radiographs Osteopenia Wormian bones in skull Bowing of the bones Diaphyseal fractures Fractures of the vertebrae Fractures of the ribs	Plain radiographs Fractures in different stages of healing Metaphyseal “bucket-handle” or “corner” fractures Fractures of the posterior ribs, scapula, or spinous processes of vertebrae Skull fractures crossing sutures	Plain radiographs Osteopenia Diaphyseal fractures Hyperplastic callus at previous fracture site Radial head dislocation Calcification of the interosseous membrane of the forearm
MRI and CT scan not indicated	MRI or CT scan of brain: Intracranial hemorrhage	MRI and CT scan not indicated
Genetic sequencing: Mutation in one of the type I procollagen genes (COL1A1 or COL1A2)	Genetic sequencing: No genetic mutation	Genetic sequencing: No mutation in type I procollagen genes (COL1A1 or COL1A2). Other genetic mutations may be present.
Diagnosis		
Osteogenesis imperfecta (OI) (Diagnostic features of the specific types of OI are outlined in Table 30.1)	Non-accidental injury	Syndrome resembling osteogenesis imperfecta (SROI) (Features of specific types of SROI are presented at the end of the chapter)

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The Child with a Painful Limp

31

James Robb

31.1 Introduction

By school age, children will have acquired an adult walking pattern but will still have a faster cadence (steps per minute) and slower velocity than an adult, and any deviation in the gait from the adult pattern should be regarded as a limp. In this age group, a painful limp must be investigated carefully as the causes include potentially serious conditions such as trauma, infection, inflammation, and tumor. A careful history may often point to the diagnosis. Children in this age group are usually good historians.

31.2 Questions to Establish a Diagnosis

- **What is the site of pain?**
- **What is the duration of the painful limp?**
- **Was the onset acute or insidious?**
- **Is there a history of antecedent trauma?**
- **Was the onset of the limp associated with fever?**
- **What are the relieving or exacerbating factors?**
- **Does the pain disturb sleep?**
- **Is there associated morning stiffness or does the limp worsen as the day goes on?**
- **Is there history of medication for any disease prior to the onset of the limp?**
- **Is the child unwell?**

What is the site of pain?

This is perhaps the most important question; accurate localization of pain can often help in eliciting the relevant clinical signs, in choosing the appropriate imaging, and in making an accurate diagnosis. However, it is important to be aware that referred pain from the hip may be felt in the knee and referred pain from the thoracolumbar spine may be felt in the groin.

What is the duration of the painful limp?

If the child has been limping for some time in spite of pain, the underlying condition may not be serious; a limp of recent onset is more worrisome.

Was the onset acute or insidious?

Pain and a limp of acute onset are typically seen following trauma or if there is an infection. Pain associated with a tumor may be more insidious in onset.

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Is there a history of antecedent trauma?

A history of trauma in most instances will facilitate a diagnosis; however, a child will seldom be able to identify the repetitive trauma that leads to a stress fracture. Occasionally the history of trauma may be misleading as it may just have drawn attention to some other pre-existing pathology.

Was the onset of the limp associated with fever?

If fever was present at the onset of symptoms, acute septic arthritis and acute osteomyelitis must be ruled out. The onset of JIA may also be associated with fever.

What are the relieving or exacerbating factors?

Pain unrelieved by rest may be seen in children with bone tumors and acute sepsis.

Does the pain disturb sleep?

Pain severe enough to disturb sleep suggests that there is serious underlying pathology.

Is there associated morning stiffness or does the limp worsen as the day goes on?

Morning stiffness is a feature of JIA and worsening of limp as the day goes on may point to an underlying neurological condition.

Is there history of medication for any disease prior to the onset of the limp?

Steroids used for treatment of leukemia or other childhood diseases are known to cause avascular necrosis of the femoral head. Antiepileptic medication can cause rickets.

Is the child unwell?

The cause of a painful limp in a child who is well is likely due to a less serious underlying disease than if the child is unwell. Musculoskeletal infection or malignancies are more probable causes in the ill child.

31.3 Physical Examination

31.3.1 Look

Observation of an Antalgic Gait

A painful limp or antalgic gait invokes mechanisms to minimize the pain of bearing weight on the affected limb. There is a shortened stance time on the affected side as the child unloads the affected limb quickly. An antalgic gait may originate from the spine or lower limbs. Pain originating from the spine may produce a different gait pattern; the child may walk more slowly but symmetrically or refuse to walk at all.

31.3.2 Feel

Systematically palpate the hip, knee, and ankle joint lines and ensure that there is no tenderness. Palpate the femur, tibia, and fibula throughout their entire lengths. Palpate the heel, the midfoot, and forefoot. Palpate the iliac fossae and the iliac wings. Palpate the posterior elements of the thoracic and lumbar spine. Note if there is in any point tenderness in any of these regions.

31.3.3 Move

Gently move the hips, knees, ankles, and subtalar joints and note if attempted movement produces pain. Check the active movements of the lumbar and thoracic spine. Flexion of the lumbar spine is assessed with the child standing and bending forward as far as possible without bending the knees. Thoracic spine movements are tested with the child sitting and rotating the trunk without moving the buttocks off the seat.

31.3.4 Special Tests

Test for Detecting Reduced Mobility of the Lumbar Spine

Place an object on the floor and ask the child to pick it up. Normally the child would bend forward and pick up the object. If there is a painful condition of the spine, the child will flex the hips and knees to avoid having to bend the spine (Fig. 31.1, Video 31.1).

Screening Tests for Detecting Limitation of Abduction and Internal Rotation of the Hip

Simple screening tests to detect limitation of abduction and internal rotation of the hip that can be applied in a busy clinic were initially devised for Perthes' disease (Joseph et al. 1988).

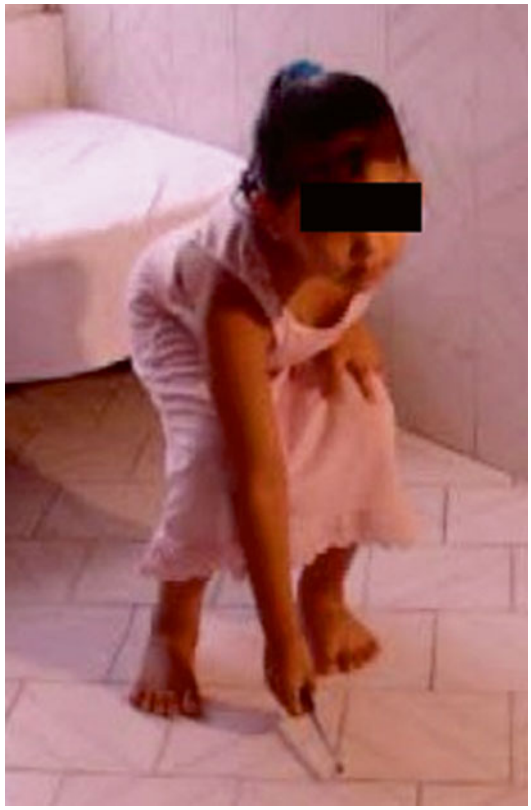

Fig. 31.1 A child with leukemic deposits in the spine attempting to pick up an object from the floor. She flexes her hips and knees to avoid having to bend the spine

These tests were found to be highly sensitive though they are not specific for Perthes' disease.

Test for Limited Abduction

The child is asked to stand in front of any vertical object (e.g., door, tall cupboard) and asked to spread the legs apart as widely as possible. The test is considered to be positive if:

(a) The torso tilts to one side; the edge of the vertical object serves as the plumb line against which to compare the alignment of the torso. The hip opposite to the side to which the torso tilts has limited abduction (Fig. 31.2a).
(b) The torso does not tilt to either side but the sum of abduction of both hips is less than 60°. Both hips are then considered to have limited abduction.

Test for Limited Internal Rotation

The child is seated on a tall stool with the legs hanging over the edge of the stool. The child is asked to keep the knees together and move the feet as far apart as possible. Limitation of internal rotation can be readily noted (Fig. 31.2b).

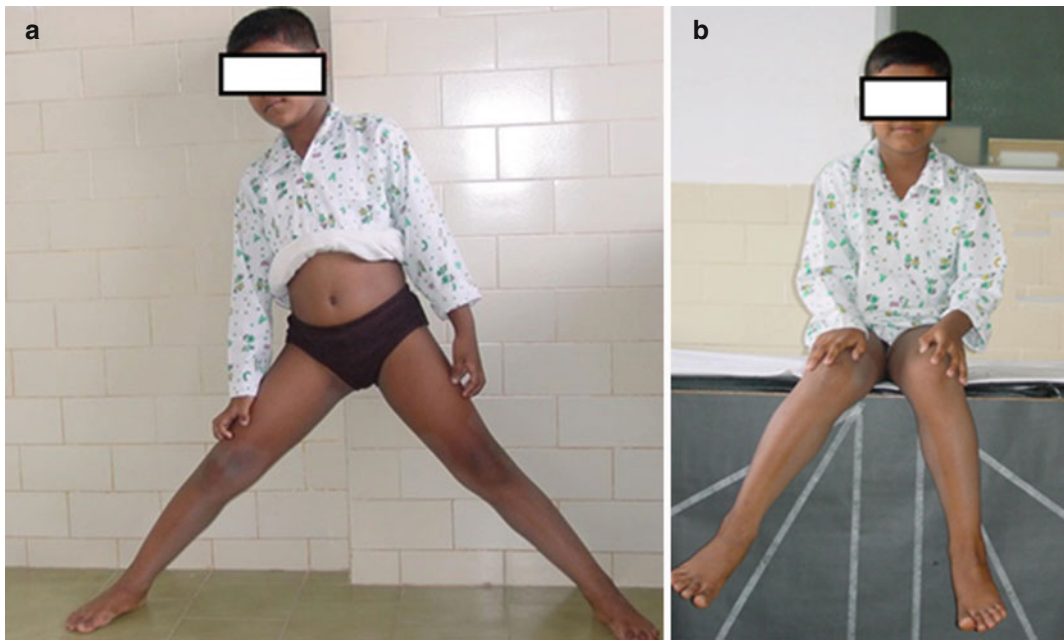

Fig. 31.2 Screening tests to demonstrate limitation of abduction of the hip (**a**) and limitation of internal rotation (**b**)

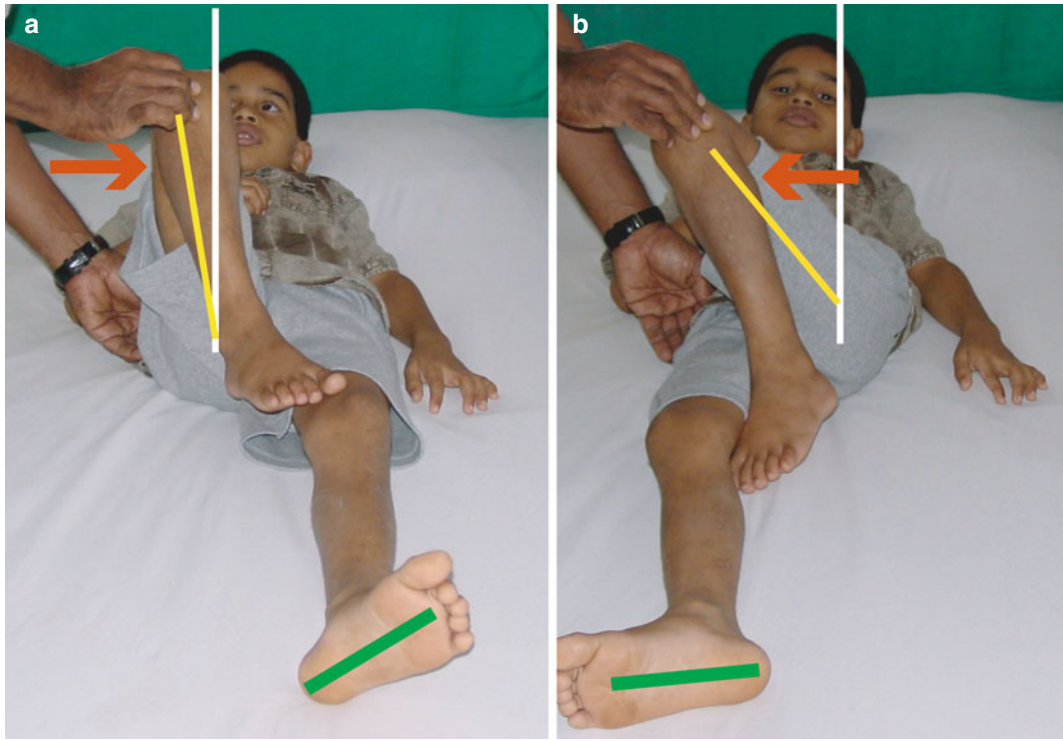

Fig. 31.3 (**a**, **b**) The flexion-adduction test: the left hip can be adducted beyond the neutral position; there is limitation of adduction in the right hip. The boy had transient synovitis of the right hip

Screening Test for Detecting Intra-articular Pathology in the Hip

The Flexion-Adduction Test (Woods and Macnicol 2001)

The child lies supine and the examiner flexes one hip to 90° and then attempts to adduct the hip. If the hip cannot be adducted beyond the neutral position, it indicates intra-articular pathology (Fig. 31.3a, b). The test is very sensitive and may be positive before restriction of other movements is evident.

31.4 Investigations to Confirm the Diagnosis

31.4.1 Imaging

Radiography

Plain radiographs (at least two orthogonal views) of the region from where the pain appears to be originating should be obtained. In many situations the diagnosis or bony alterations that point to a diagnosis may be evident on this first radiograph itself. The radiographs may show a fracture or stress fracture, new bone formation (infection, malignancy, healing fracture), resorption (malignancy, osteomyelitis), sclerosis (drug-induced avascular necrosis, Perthes' disease, osteoid osteoma), or widening of the growth plate and metaphyseal changes (rickets). Additional views may help to identify, for example, a tarsal coalition (oblique view of the foot) or an osteochondritis of the knee (tunnel view). However, plain radiographs may not show early bone changes and only give limited information on soft tissues.

Ultrasound

Ultrasound is specifically indicated in a few situations such as when a joint effusion or osteomyelitis is suspected. Even small effusions can be detected on the scan, but it may not be able to differentiate between a synovial effusion and pus of septic arthritis. Subperiosteal collection of pus

in osteomyelitis can be demonstrated clearly on the ultrasound scan and the scan can guide a needle for aspiration.

MRI Scan
MRI can provide further non-bony detail of infection and tumors and is helpful in identifying the early stages of avascular necrosis of a bone. It is also the imaging modality that can facilitate the diagnosis of pelvic osteomyelitis and pyomyositis.

Isotope Scan
Isotope scans with bone-seeking radioisotopes are particularly useful in demonstrating increased or decreased vascularity and increased bone turnover (osteoblastic activity). Osteoid osteoma, osteomyelitis, and malignant bone tumors will all show an increased uptake of the isotope, while the uptake is reduced in regions where the blood flow is diminished (e.g., Perthes' disease).

31.4.2 Laboratory Investigations

CRP will be raised in infection and inflammatory conditions. RF, ANF, and HLA-B27 are not always positive in JIA, and further specialist help should be sought if this, or other childhood inflammatory disorder, is suspected. A full blood count and film will help to exclude leukemia where, typically, the child is anemic and thrombocytopenic. Decreased levels of serum calcium, phosphate, and vitamin D with elevated levels of alkaline phosphatase and parathormone will be seen in children with rickets.

31.5 Differential Diagnosis

31.5.1 Trauma

The diagnosis is usually straightforward in children with a limp after acute trauma; however, stress fractures are more difficult to diagnose. Stress fractures may be seen in young athletes and present as unexplained pain and limp after activity. If there is no abnormality on a plain film initially, a fracture is suspected, and the child is otherwise well, it is worth repeating the radiograph 2 weeks later to see if a periosteal reaction has appeared (Fig. 31.4). An isotope scan will show increased uptake in the region.

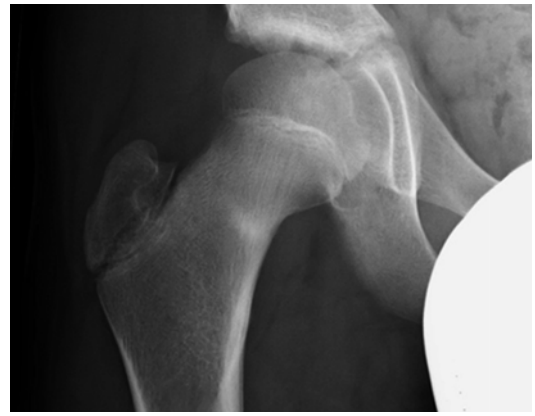

Fig. 31.4 Healed stress fracture of the femoral neck in a young athlete

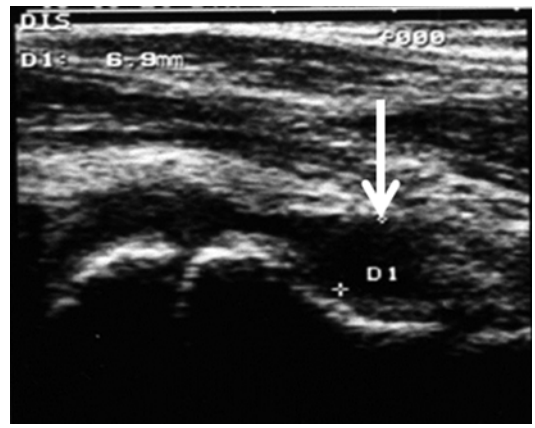

Fig. 31.5 Ultrasound of hip. The capsule (*arrow*) is distended anteriorly by an effusion

31.5.2 Infection

Septic Arthritis of the Hip
Children presenting with septic arthritis of the hip are usually unwell, febrile, and reluctant to move the hip on the affected side which typically is held in flexion and abduction. ESR and CRP are raised and ultrasound will also demonstrate a hip joint effusion (Fig. 31.5). Children presenting with transient synovitis typically have a limp with or without pain and are usually afebrile.

They hold the affected hip in flexion and abduction, and on examination there is loss of adduction and internal rotation. The diagnosis is made from a positive ultrasound; the child does not have a markedly elevated ESR or CRP and the condition is self-limiting.

One clinical challenge is to distinguish between the two conditions. Caird et al. (2006) have shown that if a child has five predictive factors (oral temperature >38.5 °C, elevated CRP, elevated ESR, refusal to bear weight, and an elevated WBC), there is a 98 % likelihood of them having septic arthritis. Those with four factors have a 93 % chance and those with three an 83 % chance. CRP >20 mg/l was a strong independent risk factor for septic arthritis. More recently, Singhal et al. (2011) have shown that only two determinants (weight-bearing status and CRP >20 mg/l) were independent in differentiating septic arthritis from transient synovitis. Individuals with neither predictor had a <1 % probability of septic arthritis, but those with both had a 74 % probability of septic arthritis (Fig. 31.6).

Septic Arthritis of the Knee and Ankle

The child presents with similar symptoms to those described above. The knee, or ankle, is warm and may be swollen and the child is reluctant to bear weight on the affected side or have the joint moved passively. Important differential diagnoses are osteomyelitis of bones adjacent to the joint and JIA as, in both conditions, the child may be febrile and have an elevated CRP (Fig. 31.7). The diagnosis is made on culture of the aspirate or if negative on the clinical response to antibiotics. Sepsis requires prompt treatment.

Osteomyelitis

In acute osteomyelitis (Peltola and Pääkkönen 2014), the child typically presents with a limp or inability to bear weight on the affected side. They may be afebrile in the earlier stages and it is important to establish if the child is immune-compromised and if they have been immunized against *H. influenzae*. There may be localizing warmth and tenderness in subcutaneous bones, where cellulitis is an important differential diagnosis, and the child may be reluctant to move the adjacent joint. CRP, ESR, and WBC are usually elevated (Fig. 31.8a–c).

Plain radiographs in the early stages will not show bony changes, but may show soft tissue swelling. Osteomyelitis usually affects the metaphysis of a long bone. Ultrasound is very useful to identify a subperiosteal collection and to distinguish the condition from septic arthritis. It may take 7–14 days, depending on the age of the child, before a periosteal reaction is seen on a plain radiograph. Pelvic osteomyelitis can be difficult to diagnose in the early stages, and important differentials are septic arthritis of the hip, osteomyelitis of the proximal femur, pyomyositis, intra-abdominal sepsis, trauma, and Ewing's sarcoma. MRI is helpful in establishing a diagnosis (Fig. 31.9).

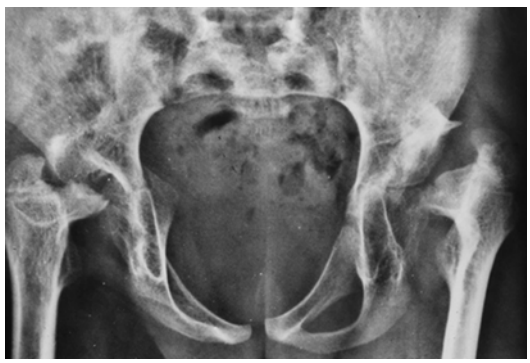

Fig. 31.6 End-stage destruction of the hips after bilateral septic arthritis

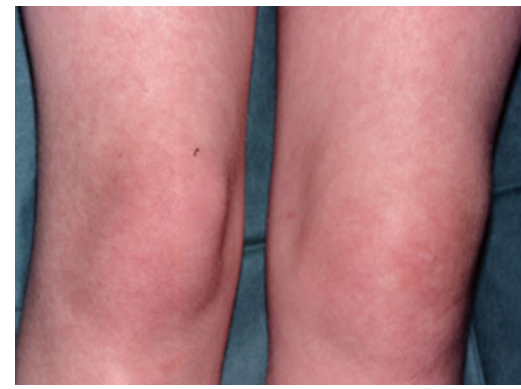

Fig. 31.7 Effusion of the left knee. This girl had morning stiffness and a persistent swelling of her left knee for several weeks and was subsequently diagnosed with oligoarthritis

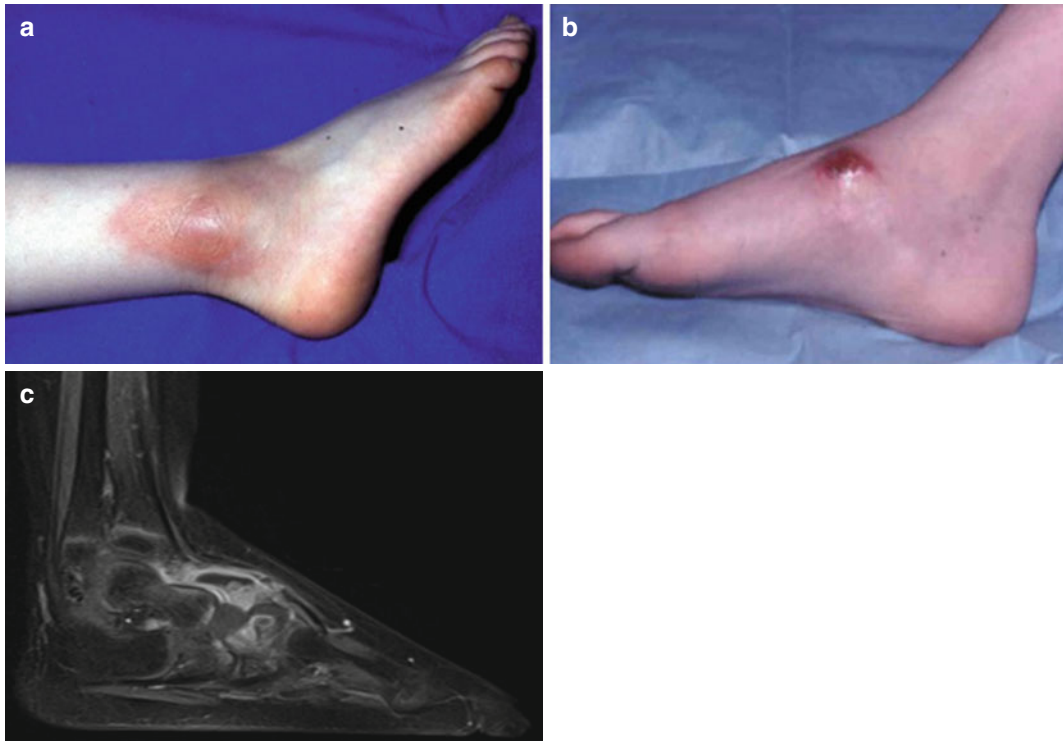

Fig. 31.8 Cellulitis or osteomyelitis? The diagnosis in (**a**) was cellulitis although the differential would include osteomyelitis of the fibula and septic arthritis of the ankle. In (**b**) the diagnosis was osteomyelitis of the navicular confirmed on MRI (**c**)

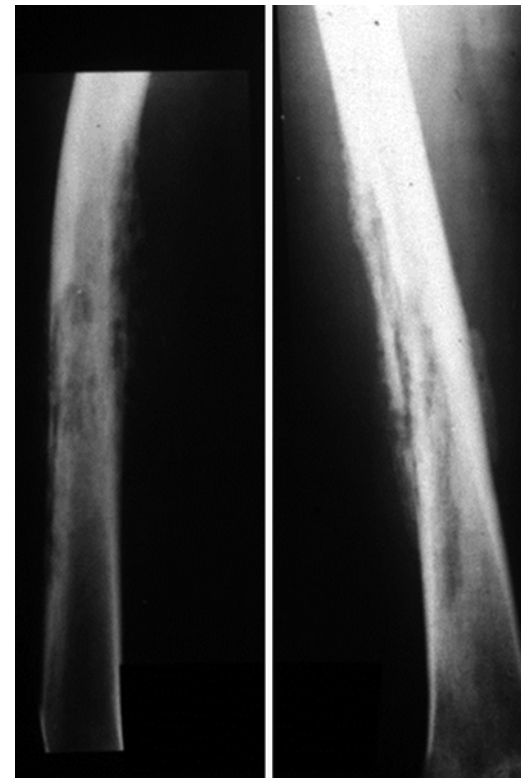

Fig. 31.9 Extensive osteomyelitis of the femur involving the diaphysis. An important differential is Ewing's sarcoma as new bone formation, sclerosis, and lysis may be seen in both conditions

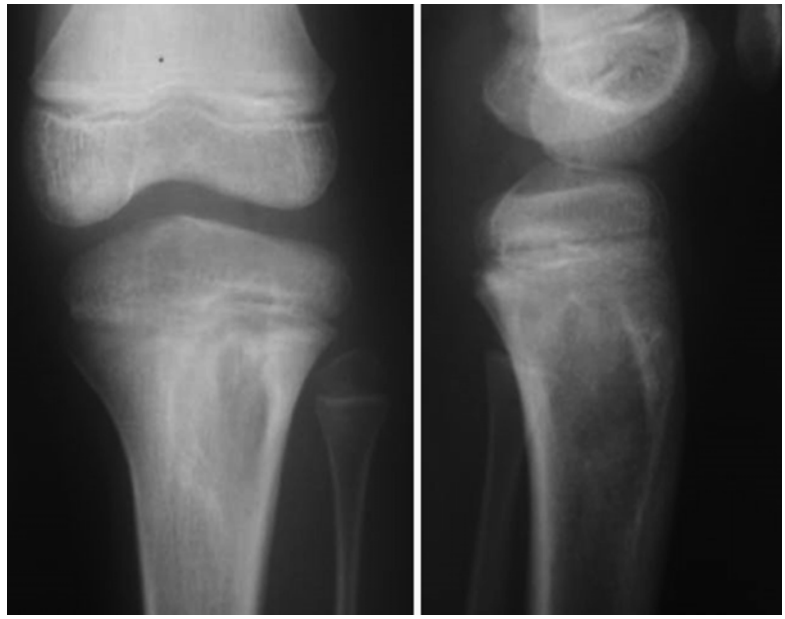

Fig. 31.10 Subacute osteomyelitis of the proximal tibial metaphysis

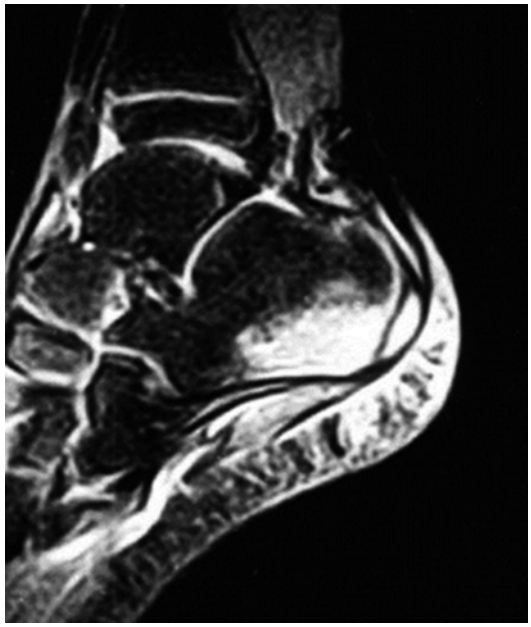

Fig. 31.11 Osteomyelitis of the calcaneus. The child had stood on a rusty nail 10 days earlier and had persisting heel pain

In subacute osteomyelitis, the child usually has symptoms for more than a month and presents with an unexplained limp and radiological changes of osteomyelitis. Chronic osteomyelitis is usually associated with late-presenting open fractures or after inadequate management of acute osteomyelitis. Pus discharging from a sinus, exposed subcutaneous bone, and a sequestrum are late clinical features (Fig. 31.10).

Penetrating Wounds

Penetrating wounds of the foot can cause infection with atypical organisms, particularly *P. aeruginosa* (Fig. 31.11).

Tubercular Arthritis and Osteomyelitis

M. tuberculosis is a cause of chronic infection of the bone, joints, and spine and is associated with immune-compromised patients. TB of joints is most often seen in the knee and hip; the ankle (Fig. 31.12) is less frequently affected.

31.5.3 Osteochondrosis

Perthes' Disease

The onset is usually between 4 and 10 years of age and boys are more commonly affected than girls. Children usually first present with a limp rather than pain; pain is often a later feature. Restricted abduction and internal rotation in extension are typical findings. A Trendelenburg

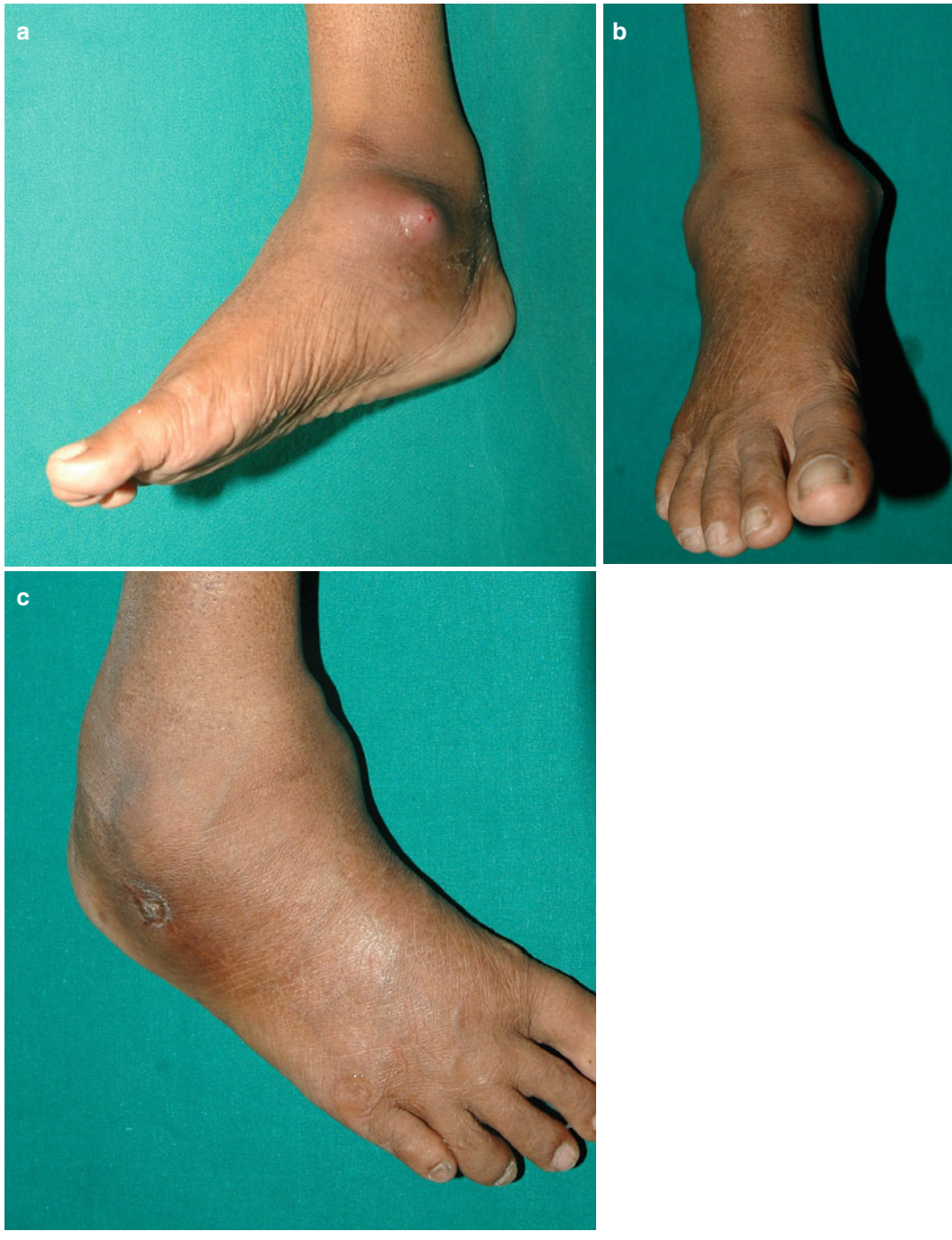

Fig. 31.12 (**a–c**) TB of the ankle

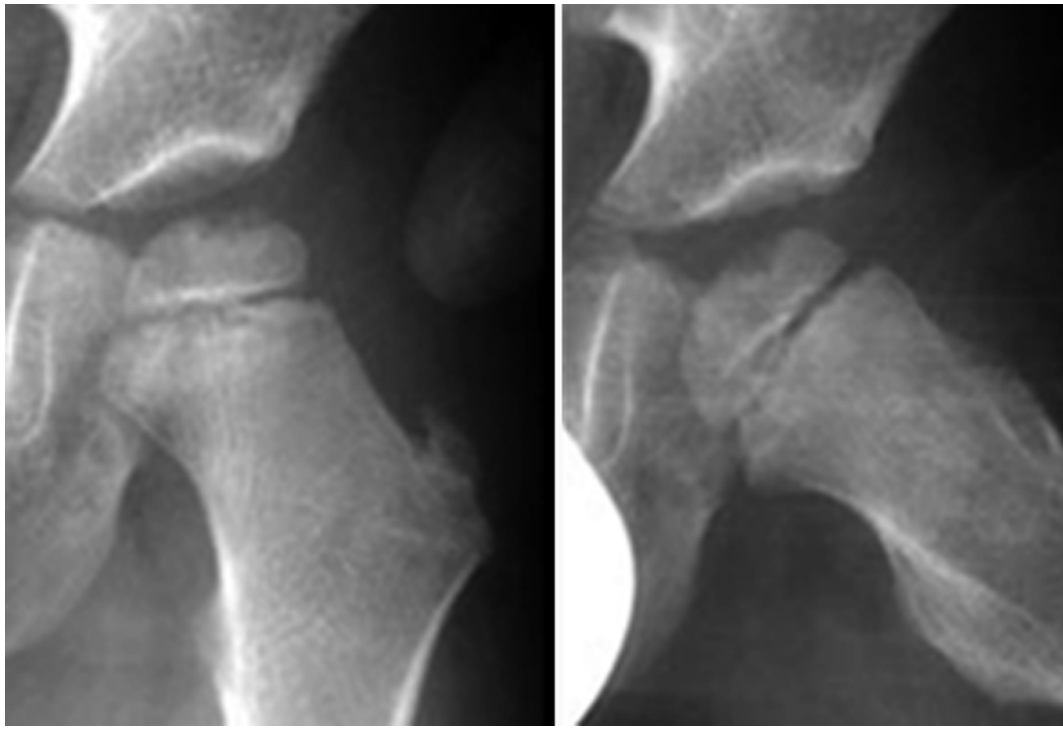

Fig. 31.13 Perthes' disease

gait pattern will not appear unless there are structural changes in the hip that affect the abductor lever arm (usually after the disease has progressed a great deal). The diagnosis is made on pelvic radiography (Fig. 31.13). Management is based on the concept of "containment" of the femoral head within the acetabulum aiming to produce, if possible, a spherical, congruous joint free of arthritis (Shah 2014).

Traction Apophysitis

Usually this affects the calcaneus or the tibial tuberosity and is seen in active children or young athletes. Pain comes on after activity and the child may limp. In the heel there may be tenderness along the calcaneal apophysis, and it is important to distinguish this from pain arising from the Achilles tendon or its insertion. At the knee there is tenderness at the tibial tuberosity which may enlarge and, if unilateral, the tuberosity on the affected side may be more prominent. This is best seen by having the child bend both knees and viewing the tuberosities from the side (Fig. 31.14).

The diagnosis of calcaneal or tibial apophysitis is a clinical one as radiographs are not diagnostic.

Osteochondritis Dissecans (OCD) of the Knee

The child may complain of mechanical knee pain that worsens with activity and is relieved by rest. They may limp and avoid full extension of the knee and may have intermittent knee effusions. The most common site is the lateral side of the medial femoral condyle. The juvenile type begins before closure of the growth plates and has a good prognosis (Fig. 31.15).

31.5.4 Neoplasia

Benign Bone Tumors

In this age group, unicameral bone cyst (often in the calcaneus), osteochondroma, Langerhans cell

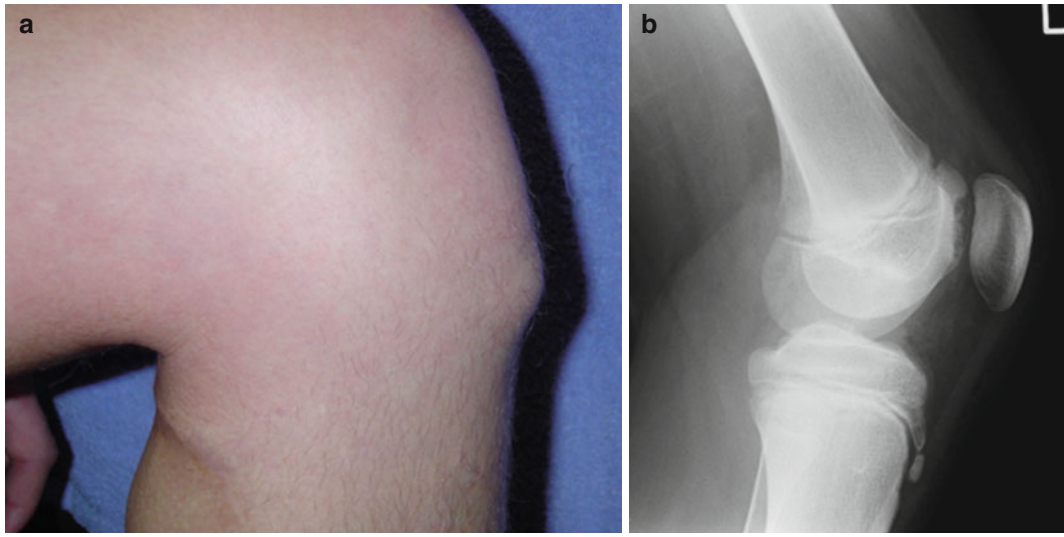

Fig. 31.14 Traction apophysitis of the tibial tuberosity, "Osgood-Schlatter disease." The tibial tuberosity is prominent (**a**) and there is a small ossicle on the lateral radiograph (**b**)

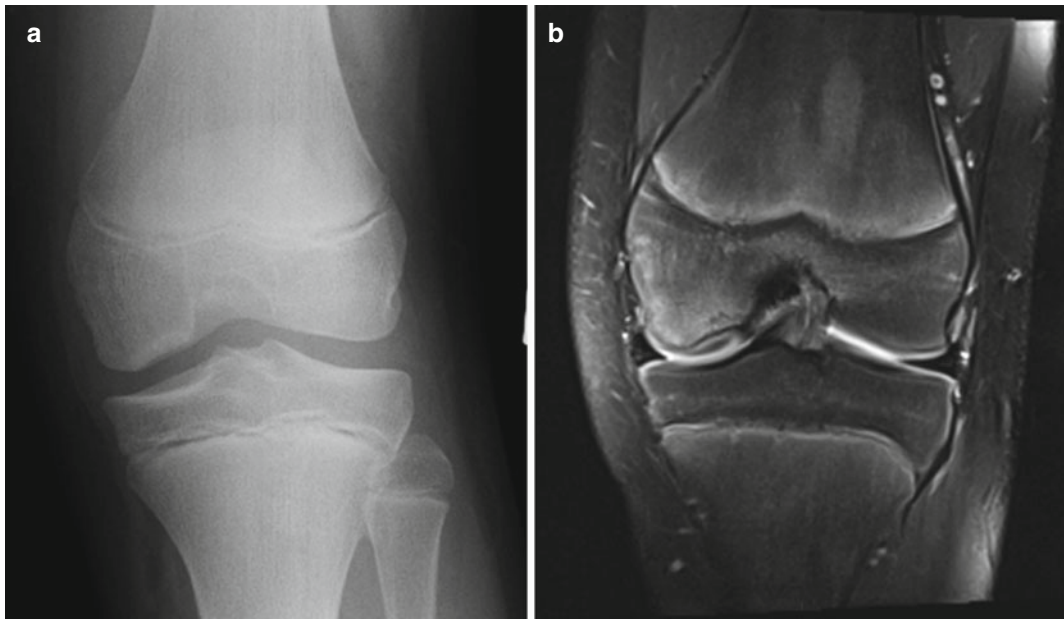

Fig. 31.15 Juvenile type of OCD of the medial femoral condyle (**a**). The MRI shows that the articular cartilage overlying the lesion is intact (**b**)

histiocytosis (LCH), and osteoid osteoma should be considered. Microfractures from a unicameral bone cyst in the lower limb may cause pain and a limp (Fig. 31.16). Rubbing of an osteochondroma against muscle bellies or tendons may be uncomfortable and inhibit movement. The swelling is palpable and has typical radiological appearances (Fig. 31.17). LCH can affect any location in a bone and the child presents with a painful radiolucent lesion that may be associated with a

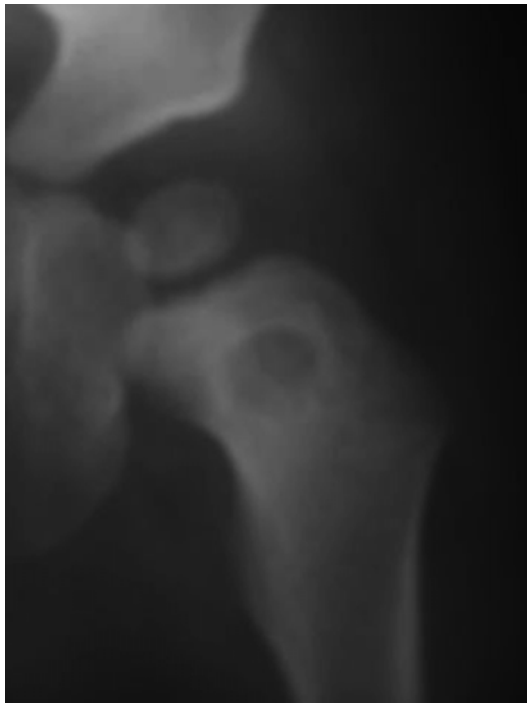

Fig. 31.16 Unicameral cyst of the femoral neck. The child presented with an unexplained limp

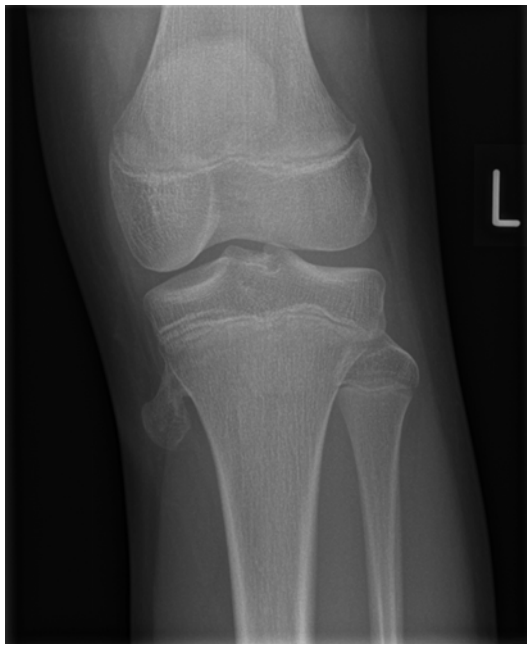

Fig. 31.17 Solitary osteochondroma of the proximal tibia

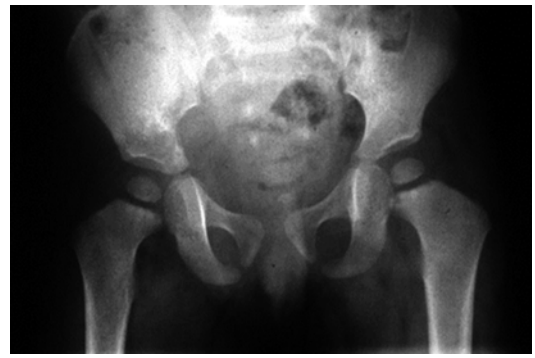

Fig. 31.18 LCH of the right supra-acetabular region. The child presented with an unexplained limp

periosteal reaction (Fig. 31.18). The child may have non-osseous lesions and should be evaluated for diabetes insipidus. In the spine it causes vertebra plana which, if painful, can cause a gait disturbance. The osseous lesions are generally self-limiting. A child with an osteoid osteoma may present with night pain, an important "red flag" symptom. There are typical appearances on plain radiography, and on MRI there may be bone edema (Fig. 31.19).

Malignant Bone Tumors

Two malignant tumors of bone should be considered in this age group: Ewing's sarcoma and osteosarcoma (Schwab et al. 2013). The child may present with unexplained bone pain (see "red flag" symptoms) and, if the condition affects a subcutaneous bone, tenderness and swelling. Radiological changes are not specific for either condition but typically will show periosteal new bone formation and lysis and in late stages the child may present with a pathologic fracture (Fig. 31.20).

Leukemia

Leukemia is the most common malignancy of children, bone pain is common, and the child appears unwell at presentation. A painful limp may be the presenting symptom (Tuten et al. 1998) (Video 31.2).

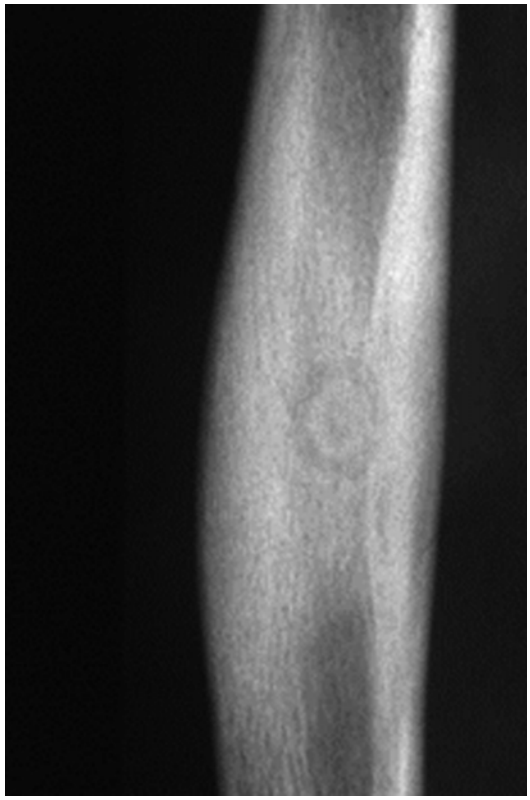

Fig. 31.19 Osteoid osteoma of the femoral diaphysis. There is a central lucency surrounded by sclerotic bone

31.5.5 Chronic Arthritis

JIA may affect any synovial joint in the lower limbs and the synovitis, effusion, joint deformity, and associated pain may cause a limp. Attempts have been made to classify gait patterns in children with JIA and correlate them with the clinical features (Fairburn et al. 2002). The four patterns that were identified were pattern I (near normal), pattern II (adaptive changes for lower limb pain), pattern III (adaptive changes for lower limb deformity), and pattern IV (adaptive changes for a combination of lower limb pain and deformity).

31.5.6 Metabolic Bone Disease

Rickets can present as bone pains and a painful limp before deformities become evident. The child may not be able to localize the site of pain very clearly. There may be a history of limited exposure to sunlight, extensive skin lesions, or antiepileptic medication, all of which can interfere with vitamin D metabolism. The metaphyseal regions of long bones may be widened and tender.

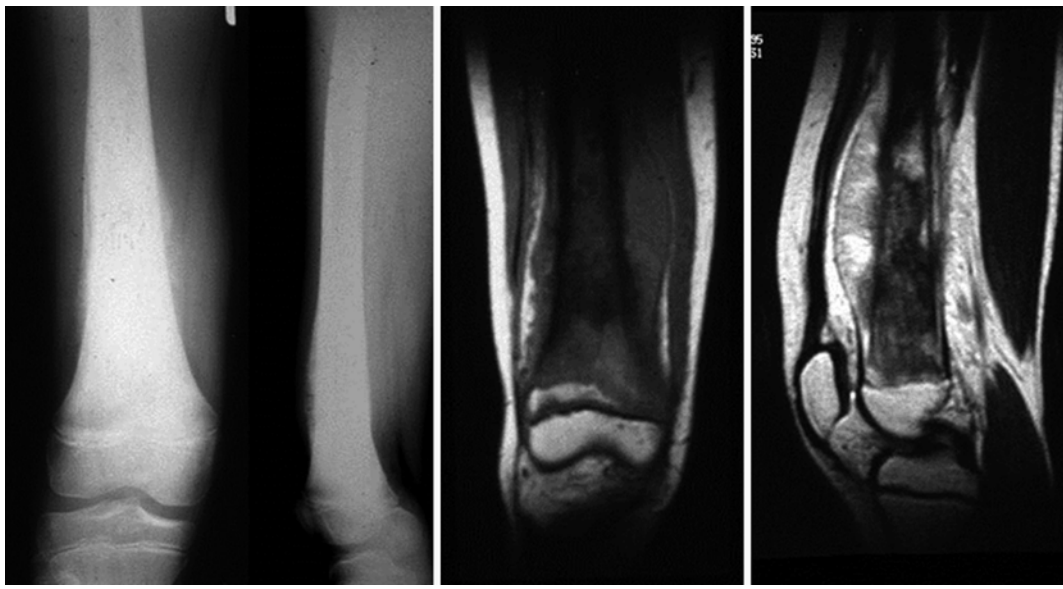

Fig. 31.20 Osteosarcoma of the distal femur. The extent of the lesion and soft tissue component on MRI is much greater than the plain radiographs might suggest

31.5.7 Neuromuscular Conditions

Muscle weakness and paralytic deformities are the common causes of a limp in children with neuromuscular disorders. However, pain in the lower limbs may develop in these conditions and aggravate the preexisting limp.

Neuromuscular causes to be considered in this age group are Duchenne muscular dystrophy (DMD), where the boy develops anterior foot pain due to equinus, the hereditary motor and sensory neuropathies (HMSN) where peroneal muscle weakness causes excessive loading on the lateral side of the foot, and Friedrich's ataxia, characterized by a broad-based ataxic gait pattern, and cerebral palsy (CP). Crouch (flexed knee gait) in CP produces patellofemoral pain and, if longstanding, an elongation of the patellar tendon, which may also be painful at its origin or insertion. Cavus feet are often seen in neuromuscular conditions and children may limp and complain of pain over the metatarsal heads.

31.5.8 Other Regional Causes of Painful Limp

Pain of Spinal Origin

Referred pain from the spine may cause a limp (Fig. 31.21). In the young athlete, spondylolisthesis (Fig. 31.22) may be associated with restriction of spinal movement and short hamstrings. Severe spondylolisthesis may cause nerve root irritation or compression. Discitis (Fig. 31.23)

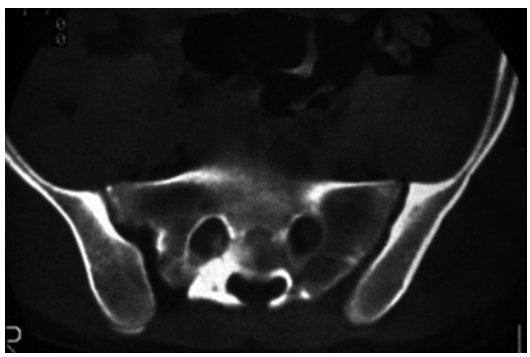

Fig. 31.21 Osteoid osteoma of the sacrum showing extensive sclerosis

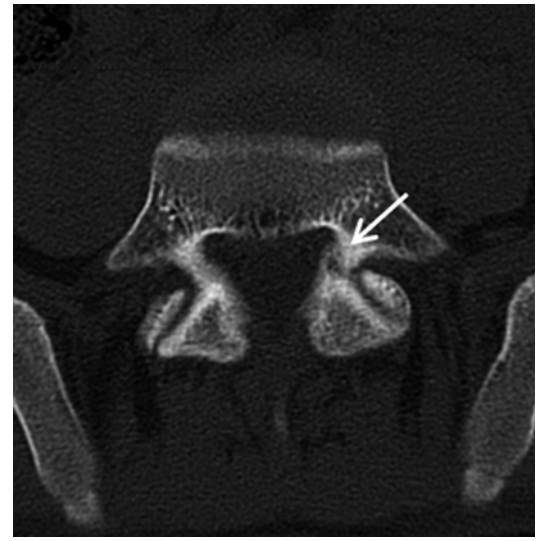

Fig. 31.22 CT slice through L5 showing a healing spondylolysis (*arrow*)

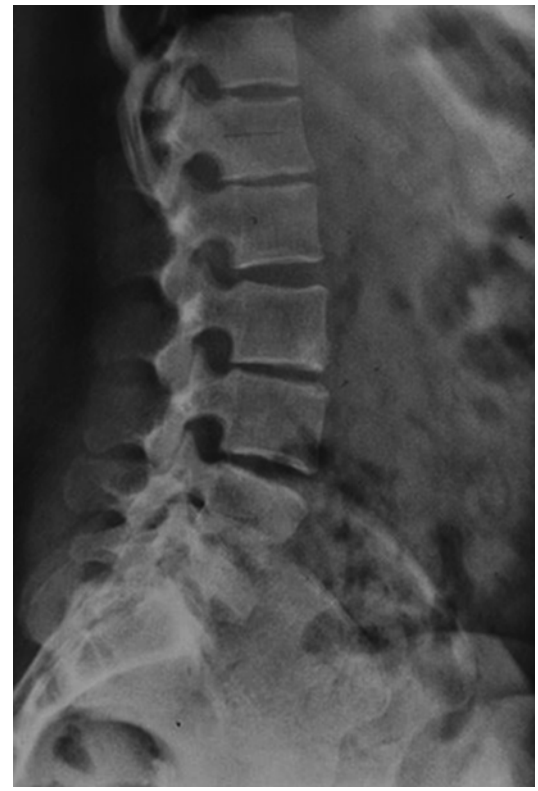

Fig. 31.23 Healing vertebral osteomyelitis at L4/5. There are loss of vertebral body height of L5, narrowing of the disk space, and destruction of the contiguous end plate of L4

and vertebral osteomyelitis may also be associated with a disturbance of gait.

Pain in the Foot

Tarsal Coalition

A calcaneonavicular cartilaginous bar starts to ossify between the ages of 8 and 12 years and may produce pain and a limp. The child may have restricted mid-foot movement and the bar may be apparent on an oblique radiograph of the foot (Fig. 31.24) or if still cartilaginous may be shown on MRI or CT. Talocalcaneal bars are best visualized on CT or MRI and become symptomatic a couple of years later. The medial longitudinal arch is lost and the hindfoot is in valgus. Pain and a limp are often noted in these children.

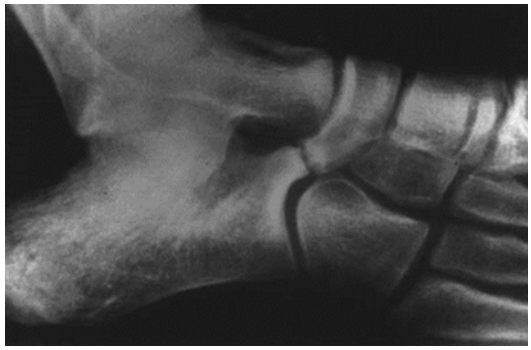

Fig. 31.24 Calcaneonavicular bar shown on an oblique radiograph of the foot

Accessory Navicular

The child may complain of pain at the insertion of the tibialis posterior tendon or from rubbing of the prominence against a shoe. There is a prominence over the medial aspect of the navicular which may be tender and the diagnosis can be confirmed radiologically (Fig. 31.25).

31.5.9 Rarer Causes of Painful Limp

Psychogenic Limp

Children with psychological problems may present with a "painful limp" (Tripathy et al. 2014). The gait pattern may be bizarre and sometimes inconsistent with the pattern varying between examinations. The child may hold the limbs stiff and resist passive movement. There may be symptoms of psychological distress and further specialist evaluation may reveal an underlying cause.

Complex Regional Pain Syndrome (CRPS)

Children with CRPS type 1 affecting the lower limb often limp (Wilder et al. 1992). The diagnosis of CRPS is made on clinical grounds; the characteristic features include pain, sensory disturbances (hyperesthesia or dysesthesia), vasomotor disturbances (hyperemia, pallor, or cyanosis of the extremity), and sudomotor

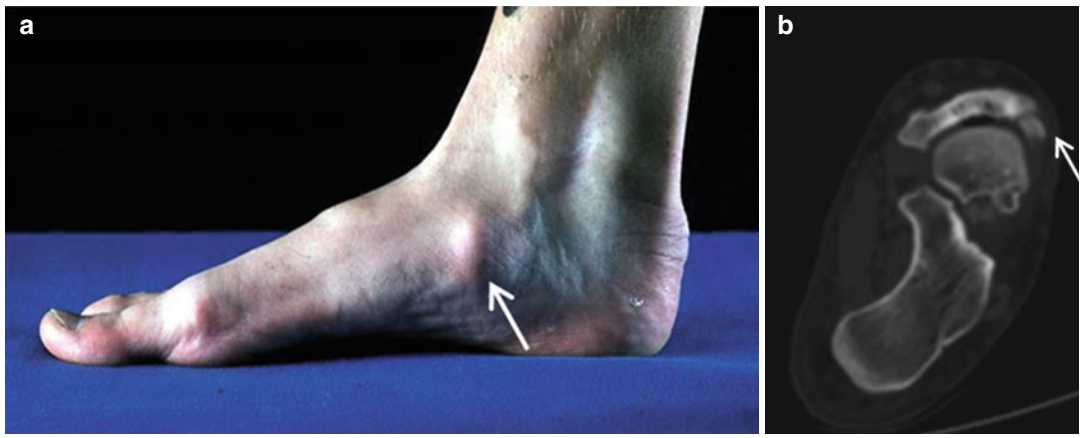

Fig. 31.25 Accessory navicular (*arrow*) shown clinically (**a**) and on CT (**b**)

Table 31.1 Recommended investigations based on the clinical situation in a child with a painful limp

Clinical situation	Choice of investigation
	Initial investigation
Site of pain clearly specified by child and localizing signs point to same site	Anteroposterior and lateral plain radiographs of the affected limb centered on that region
Site of pain not specified by child but there are localizing signs on physical examination	Anteroposterior and lateral plain radiographs of the affected limb centered on site of physical signs
Site of pain not specified by child and no localizing signs are elicited	Anteroposterior and lateral radiographs of both lower limbs (include the pelvis in the AP view)
Infection suspected	Complete blood count, ESR, CRP, blood culture + Plain radiographs
Inflammatory arthritis suspected	Complete blood picture, ESR, CRP, rheumatoid factor, antinuclear antibodies, HLA-B27
Metabolic bone disease suspected	Serum calcium, phosphate, vitamin D, alkaline phosphatase estimation
	Further investigation
Hip effusion suspected	Ultrasound to confirm hip effusion and to facilitate aspiration if septic arthritis is a probability
Site of pain not clearly specified No localizing signs elicited Plain radiographs do not show abnormality	Isotope bone scan
Vertebral or pelvic osteomyelitis or pelvic pyomyositis suspected	MRI scan
Pathology identified on plain radiographs but extent of intramedullary and soft tissue involvement not clear	MRI scan

disturbances (excessive sweating or dryness of the skin). Unusual patterns of pain can also occur (Agarwal and Joseph 2006).

31.6 Establishing the Diagnosis

It is important that the appropriate investigations are done without doing several unnecessary ones. A rational approach would be to go in a sequential manner, selecting the most appropriate investigation for the particular clinical situation (Sawyer and Kapoor 2009). Table 31.1 shows what investigations are needed for the most common conditions. Outlines of the approach to establishing a diagnosis are shown in Tables 31.2 and 31.3.

Table 31.2 Establishing the diagnosis of the cause of a painful limp in a child who looks ill

History				
Fever at onset	Fever at onset	Fever at onset	Fever may or may not have been present at onset	Fever may or may not have been present at onset
Site of pain clearly localized by the child (over the joint)	Site of pain clearly localized by the child (over the bone)	Site of pain clearly localized by the child (over the back)	Site of pain not clearly localized by the child	Site of pain often not localized by the child
Physical examination				
Febrile/toxic	Febrile/toxic	Febrile/toxic	Febrile/toxic	Febrile
Gait: May be unable to bear weight on the limb Antalgic gait if able to walk	Gait: May be unable to bear weight on the limb Antalgic gait if able to walk	Gait: May be unable to walk Shuffling gait with stiff back (not typical antalgic gait)	Gait: Antalgic gait	Gait: May be antalgic or shuffling gait
Warm swollen joint (if superficial)	Warmth and swelling over the metaphysis of the affected bone (if superficial)	No demonstrable warmth or swelling on the back	No demonstrable warmth or swelling	No localized warmth or swelling
Tenderness in the joint line	Tenderness over the metaphysis	Tenderness over the spine and paraspinal region	Tenderness may be present in the iliac fossa Often tenderness cannot be elicited	Tenderness may occasionally present on the back or a limb
Fixed deformities due to muscle spasm	Fixed deformities not present	Loss of lumbar lordosis Paraspinal muscle spasm present	Fixed flexion deformity will be present if there is psoas spasm	Deformities are usually not present
Passive movement grossly restricted due to pain	Gentle passive movement of the adjacent joint possible	Movements of the spine are grossly limited	Some movements of the hip may be restricted due to muscle spasm (varies with the location of infective focus)	Movements of a joint or the spine may or may not be restricted
		Signs of nerve root irritation may be present		
		Loss of sphincter control due to an epidural abscess formation can develop (*this is a surgical emergency*)		
Working diagnosis: Septic arthritis	Working diagnosis: Acute osteomyelitis	Working diagnosis: Vertebral osteomyelitis	Working diagnosis: Pelvic osteomyelitis or pelvic pyomyositis	Working diagnosis: Leukemia

(continued)

Table 31.2 (continued)

Investigations				
WBC count: Increased with neutrophilia (>12,000/mm^3)	WBC count: Increased with neutrophilia	WBC count: Increased with neutrophilia	WBC count: Increased with neutrophilia	WBC count: Neutropenia Platelet count: decreased Anemia
ESR raised (>40 mm)	ESR raised	ESR raised	ESR raised	ESR raised
CRP raised (>2 mg/dL)	CRP raised	CRP raised	CRP raised	CRP raised
Blood culture: Occasionally positive	Blood culture: Occasionally positive	Blood culture: Occasionally positive	Blood culture: Occasionally positive	-
Plain radiograph May not show any abnormality	Plain radiograph May not show any abnormality Rarefaction in the metaphysis may be present	Plain radiograph Rarefaction or destruction of vertebral body	Plain radiograph May not show any abnormality	Plain radiograph May not show any abnormality
	MRI Bone destruction, edema, subperiosteal collection of pus	MRI Vertebral destruction, Epidural abscess may develop	MRI Destructive changes in one of the pelvic bones or fluid collection in relation to muscles in the pelvis	
Joint aspiration and synovial fluid Gram's stain and culture: Positive culture not always obtained	Subperiosteal fluid aspiration, Gram's stain and culture: Positive culture not always obtained			Bone marrow aspiration cytology
Diagnosis				
Septic arthritis	Acute osteomyelitis (lower limb bone)	Vertebral osteomyelitis	Pelvic osteomyelitis/pelvic pyomyositis	Leukemia

Table 31.3 Establishing the cause of a nontraumatic painful limp in a child who is not ill

History			
Pain on moving the joint	Pain on exertion (not caused by any specific movement) or pain at rest	Pain on walking	Pain in the back aggravated by movement
	History of repetitive stress may be present	History of repetitive stress may be present	
Physical examination			
Swelling of the joint (evident in superficial joint but not in hip)	Swelling +/– over the bone	Swelling +/– over the end of the tendon or over the site of bony attachment of the tendon	No swelling of the spine
Warmth over the joint (evident in superficial joint but not in hip)	Warmth +/– over the bone	Warmth +/– over the tendon	No warmth over the spine

Table 31.3 (continued)

Joint line tenderness	Tenderness over the bone	Tenderness over the tendon or the bony attachment of the tendon	Tenderness may be present
Effusion	No effusion	No effusion in adjacent joint	–
Synovial hypertrophy	No synovial hypertrophy	Tenosynovium may be hypertrophic	–
Muscle spasm	No muscle spasm	No muscle spasm	Paraspinal muscle spasm may be present Hamstrings may be tight
Decreased range of motion (may be limitation of extremes of certain movements or may be throughout the range of movement)	Range of motion of adjacent joint usually not affected	Pain on stretching the tendon (by passively moving the limb in the direction opposite to the movement produced by the muscle of the affected tendon	Pain on moving the lumbar spine
		Pain on actively contracting the muscle of the tendon against manual resistance	
Working diagnosis: Pathology in the joint	Working diagnosis: Pathology in the bone	Working diagnosis: Pathology in tendon or at the site of attachment of the tendon	Working diagnosis: Pathology in the vertebral column
Investigations			
Plain radiograph Not helpful in many instances but will show changes in intra-articular epiphysis if present	Plain radiograph often diagnostic	Plain radiograph may be diagnostic if the bone is diseased or avascular	Plain radiograph may show: Narrowing of disk space and para-diskal irregularities (discitis) Defect in pars interarticularis Altered alignment of the vertebra (listhesis)
MRI confirmation may be used if epiphyseal avascularity is suspected on plain radiograph	MRI confirmation occasionally required	MRI confirmation seldom required	MRI confirmation to be considered if root irritation signs are present
Ultrasound useful to confirm effusion in hip (not needed in superficial joint)		Ultrasound may show changes in and around tendon	
Diagnosis			
Intra-articular pathology as listed below: *Hip*: Transient synovitis Perthes' disease JIA Reactive arthritis *Knee*: JIA Reactive arthritis Osteochondritis dissecans Hemophilic arthropathy *Ankle* JIA	*Bone pathology as listed below*: Stress fracture Osteoid osteoma Simple bone cyst Subacute osteomyelitis Rickets	*Tendon pathology or osteochondritis as listed below*: *Knee*: Osgood-Schlatter *Ankle and foot*: Sever Kohler Accessory navicular *Tendinitis*: Achilles tendon Tibialis posterior Peroneus	*Spinal pathology as listed below*: Discitis Spondylolysis Spondylolisthesis

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The Painful Hip

32

Randall T. Loder

32.1 Introduction

The diagnoses of a painful hip in a child can range from straightforward conditions with a good prognosis to severe conditions with a poor prognosis, either for the future of the hip or the life of the patient. It is important to remember that hip pathology can manifest not just as hip pain but also as knee pain. Hence, any child complaining of pain in the hip or knee must have a thorough examination of the hip as outlined in this chapter. Several conditions that occur in younger children may also occur in adolescents; this chapter will only focus on the school-going age child.

32.2 Questions to Establish a Diagnosis

History
- **Are the symptoms unilateral or bilateral?**

Unilateral involvement
- **Was the onset gradual or abrupt?**
- **Are there associated constitutional symptoms (fever, chills, weight loss)?**
- **Can the child still walk or not?**
- **Does the pain occur at night or all the time?**
- **Is the pain relieved by NSAIDs?**
- **Are there any other associated medical conditions?**

Bilateral involvement
- **Are other joints also painful?**
- **Are there deformities of other joints, altered body proportions, or dwarfism?**

Physical Examination
- **Is there loss of hip motion?**
- **Does the loss of motion affect only movements in some planes or is it global?**
- **Are there fixed deformities?**

Are the symptoms unilateral or bilateral?

This is an important question that helps to separate conditions that seldom affect both hips from conditions where bilateral involvement is commonplace. Bilateral symptoms are rare with fractures, infections, and neoplasms and uncommon with Perthes' disease but often seen in hemoglobinopathies and skeletal dysplasias.

Was the onset gradual or abrupt?

Abrupt acute onset heralded by injury or fever and constitutional symptoms suggests that the cause of the symptoms is a fracture, dislocation, or an articular or peri-articular infection. If the onset was gradual, noninfective inflammatory conditions, neoplasms, and Perthes' disease are more likely possibilities.

Can the child still walk or not?

The ability to ambulate is frequently lost with fractures and acute septic arthritis and other

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forms of advanced arthritis on account of pain.

Are there associated constitutional symptoms?

Fevers and chills are frequently associated with infections and weight loss with tubercular infection and malignant neoplasms.

Does the pain occur at night?

The classic history for an osteoid osteoma is night pain; pain associated with Perthes' is frequently relieved by rest and aggravated by activity.

Is the pain relieved by NSAIDs?

Osteoid osteoma pain is often relieved by NSAIDs and to some extent for other inflammatory processes (e.g., inflammatory arthropathies).

Are there any other associated medical conditions?

Children with the various epiphyseal dysplasias frequently have hip involvement. Those with hemoglobinopathies also can have hip involvement.

Are other joints also painful and are there deformities of other joints, altered body proportions, or dwarfism?

Pain in other joints and deformities of other joints suggest a generalized condition such as juvenile chronic arthritis. Altered body proportions and dwarfism may indicate that the child has a form of skeletal dysplasia.

32.3 Physical Examination

32.3.1 Look

The need for a careful general physical examination cannot be overemphasized. Note the body habitus; is the child emaciated or ill? Note the stature and body proportions. Inspect other joints of the body and note if they are swollen or deformed. If the child can stand, note the posture; note if there is exaggerated lumbar lordosis (possibly due to a fixed flexion deformity of the hip). Note if there is a tilt of the pelvis (possibly due to a fixed abduction or adduction deformity of the hip) and note if there is limb length inequality. Confirm these findings when the child is examined recumbent. Note if there is wasting of the gluteal and thigh muscles; this suggests that the underlying problem is not of very recent onset. Observe the position in which the limb lies; in particular, note if the limb lies flexed, abducted, or adducted and internally or externally rotated.

32.3.2 Feel

Note if the local temperature over the joint is increased and if there is tenderness directly over the joint in Scarpa's triangle. Also systematically palpate the greater trochanter, the gluteal region, the iliac bone, the pubis, and the inguinal fossa for tenderness or fullness.

32.3.3 Move

The most important component of the physical examination is noting the range of motion of the hip. Careful documentation of the range of hip motion in three planes can often help in arriving at a provisional diagnosis in many instances (Rao and Joseph 2001; Sankar et al. 2012).

Screening Tests for Detecting Limitation of Motion of the Hip

Before actually measuring the range of motion, a few simple tests to demonstrate limitation of hip motion can be initially employed (Table 32.1; Figs. 31.2 and 31.3) (Frick 2006; Perry and Bruce 2010; Rao and Joseph 2001; Woods and Macnicol 2001). These tests are very useful in a busy clinic. The added benefit of some of these tests is that the child does not have to lie down on the examination couch and the clinician does not have to touch the child both of which can be stressful for anxious children.

Apart from giving an impression of the range of all movements of the hip except extension (squat test, flexion; hip abduction-torso tilt test, abduction; sit and split test, internal rotation; log roll test, internal and external rotation; flexion-adduction test, adduction), these screening tests may be sufficiently sensitive to diagnose or exclude hip disease. Woods and Macnicol observed that the flexion-adduction test is an early and sensitive test to diagnosis hip disease (Woods and Macnicol 2001). The hip abduction-torso tilt test was noted to be an

Table 32.1 Simple screening tests to demonstrate limitation of range of hip motion in a child

Test	What to look for	Interpretation
The squat test:		
Ask the child to squat on the ground Not possible if child cannot bear weight on the limb Child does not have to lie on examination couch Child does not have to be handled by clinician Provides information about the knee and ankle in addition to the hip	If the child can squat normally without any discomfort	The range of hip flexion, knee flexion, and ankle dorsiflexion is normal
	If the child cannot adopt the squat posture	There is limitation of either hip or knee flexion (due to pain or contracture)
	If the child squats with one foot forward	There is either limitation of flexion of the affected knee or limitation of ankle dorsiflexion (due to pain or contracture)
	If the child can only squat with the heel off the ground	There may be a contracture of the gastroc-soleus
	If the child can only squat with the hips widely abducted	There is probably a contracture of the gluteus maximus
The hip abduction-torso tilt test:		
Child standing Ask the child to stand with hips as widely apart as possible. The interpretation of the test is easier if the child stands in front of a door or pillar, the edge of which can serve as the plumb line (Fig. 31.2a) Not possible if child cannot bear weight on the limb Child does not have to lie on examination couch Child does not have to be handled by clinician	The torso (trunk) remains vertical, and the sum of abduction of both hips equals or exceeds 60°	There is normal symmetric range of abduction of both hips
	The torso tilts to one side	There is limitation of abduction of the hip opposite to the side to which the torso tilts
	The torso remains vertical, and the sum of abduction of both hips is less than 60°	There is symmetric reduction of hip abduction
Sit and split test:		
Child seated on a chair or stool Ask the child to sit with knees together and keeping the knees together move the feet apart as widely as possible (Fig. 31.2b) Possible if child cannot bear weight on the limb Child does not have to lie on examination couch Child does not have to be handled by clinician	Both feet move apart symmetrically with legs at an angle of at least 30° from the vertical	Normal internal rotation of both hips
	Both feet move apart symmetrically with legs at an angle of at less than 30° from the vertical	Decreased range of internal rotation in both hips (bilateral disease)
	Reduction of movement of symptomatic hip	Decreased range of internal rotation of symptomatic hip
Log-roll test:		
Child lying supine First, gently log-roll the asymptomatic limb; then log-roll the painful limb Note if rotations are equal on both sides and if the test causes pain	Log-rolling the limb causes severe pain	The underlying pathology is severe (possible septic arthritis or a peri-articular fracture)
	Log-rolling not painful but motion restricted on the symptomatic side	There is limitation of rotations of the hip
Flexion-adduction test		
Child lying supine Flex the hip to 90° and attempt to adduct the flexed hip	The hip can be adducted such that the thigh moves beyond the mid-vertical line	Intra-articular hip pathology unlikely
	The thigh cannot be adducted up to the mid-vertical line (Fig. 31.3)	Limitation of hip adduction in flexion suggests that there is some intra-articular pathology

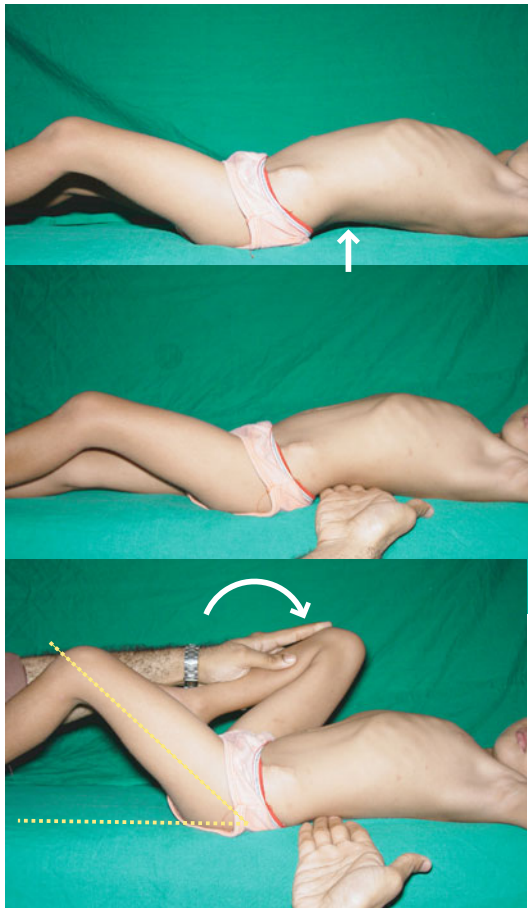

Fig. 32.1 The Thomas test to demonstrate and quantify flexion deformity of the hip. When the child is recumbent, the exaggerated lumbar lordosis is seen (*straight arrow*). The hand of the examiner is placed under the lumbar spine, and the normal hip is flexed till the lumbar lordosis is obliterated (*curved arrow*). The angle the thigh makes with the couch is the degree of flexion deformity (*yellow dotted lines*)

extremely sensitive screening test to detect limitation of hip abduction in Perthes' disease (Joseph 2002). It follows that if all these screening tests are normal, it is highly probable that the hip is normal.

Screening Test to Exclude Serious Hip Pathology

When the child is recumbent, ask the child to raise the limb with the knee extended and note if the child can do this without complaining of pain. Then offer some resistance and see if the child can raise the limb against the resistance offered (Frick 2006). The muscle contraction needed to raise the limb against resistance will load the hip quite significantly; a child who can do this test without any discomfort is unlikely to have major intra-articular pathology (Frick 2006).

Identification and Measurement of Fixed Deformities

Before proceeding to measure the passive range of motion of the hip, fixed deformities, if present, need to be identified and measured (Figs. 32.1 and 32.2). The fixed deformities that may be encountered include flexion, abduction, adduction, and internal and external rotation. These deformities are identified on the basis of clinical findings outlined in Table 32.2.

Measurement of the Range of Motion

A visual estimate of the range of motion is most widely used in clinical practice. Measurement with a linear goniometer or with a fluid-level goniometer increases the accuracy of quantifying joint motion considerably (Rao and Joseph 2001). The use of a goniometer is recommended if the clinician intends to sequentially monitor the range of motion either to document progression or resolution of the disease process.

The pattern of restriction of motion varies with the underlying pathology in the hip, and knowledge of these patterns is helpful in arriving at a diagnosis (Fig. 32.3). In the majority of children with early Perthes' disease, there is selective reduction of abduction and internal rotation, the other movements remaining almost normal (Fig. 32.3b). In situations such as transient synovitis, tubercular synovitis, or early non-virulent septic arthritis, there is an effusion in the hip which distends the joint capsule. Pain is experienced when the distended capsule is stretched further by adducting, internally rotating, or extending the hip (during these movements, the iliofemoral, pubofemoral, and

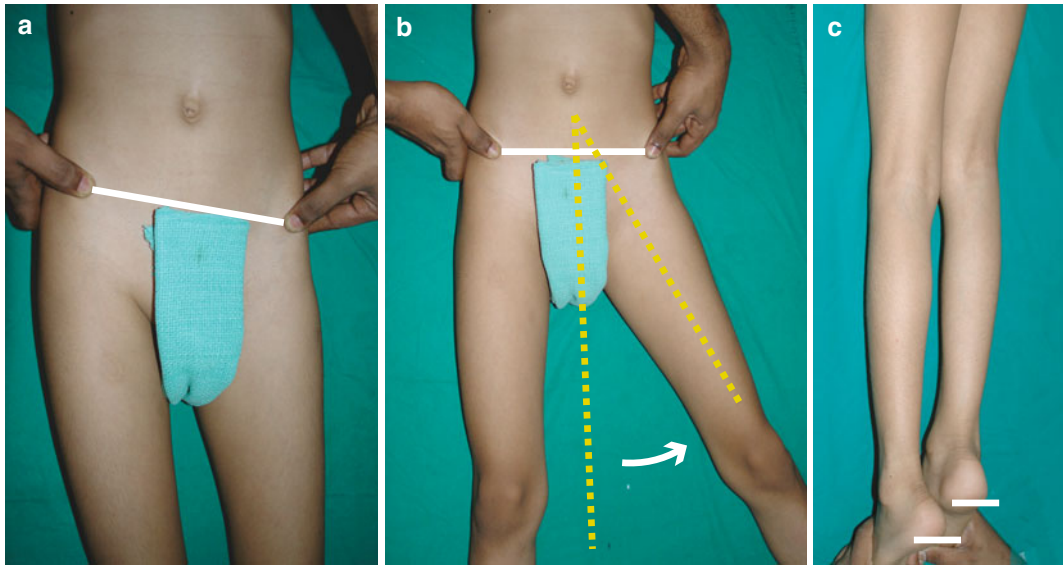

Fig. 32.2 An abduction deformity of the hip is suspected when the pelvis is tilted such that the anterior superior iliac spine (ASIS) on the symptomatic side is at a lower level (**a**). The symptomatic hip is abducted till the pelvis is "squared" (i.e., both ASIS at same level). The angle to which the limb is abducted is the degree of fixed abduction deformity (**b**). On account of the pelvic obliquity, there is apparent lengthening of the affected limb (**c**)

Table 32.2 Diagnosis and measurement of fixed deformities of the hip

Fixed deformity	Signs observed when child lies supine with the lower limbs parallel	How to measure the extent of deformity
Fixed flexion deformity	Exaggerated lumbar lordosis Forward pelvic rotation (Fig. 32.1)	*Thomas test* Flex the asymptomatic hip till the lumbar lordosis is obliterated Ask the patient to avoid actively flexing the symptomatic hip Measure the angle the thigh makes with the couch
Abduction deformity	Pelvic tilt (ASIS of affected side lower) Apparent lengthening of the limb (Fig. 32.2a, c)	Square the pelvis by abducting the symptomatic hip till the left and right ASIS are at the same level Measure the angle that the abducted limb makes with the long axis of the trunk (Fig. 32.2b)
Adduction deformity	Pelvic tilt (ASIS of the affected side higher) Apparent shortening of the limb	Square the pelvis by adducting the symptomatic hip till the left and right ASIS are at the same level Measure the angle that the adducted limb makes with the long axis of the trunk
Internal or external rotation deformity	Patella faces excessively medially or laterally	Bring the child's knees to the edge of the couch with the child lying supine with thighs parallel. Let the legs hang down over the edge of the couch Note if the leg lies vertically. If the leg is deviated away from the mid-vertical line, the hip is in internal rotation while if the leg is deviated towards the mid-line the hip is externally rotated The angle that the leg makes with the vertical is the degree of deformity

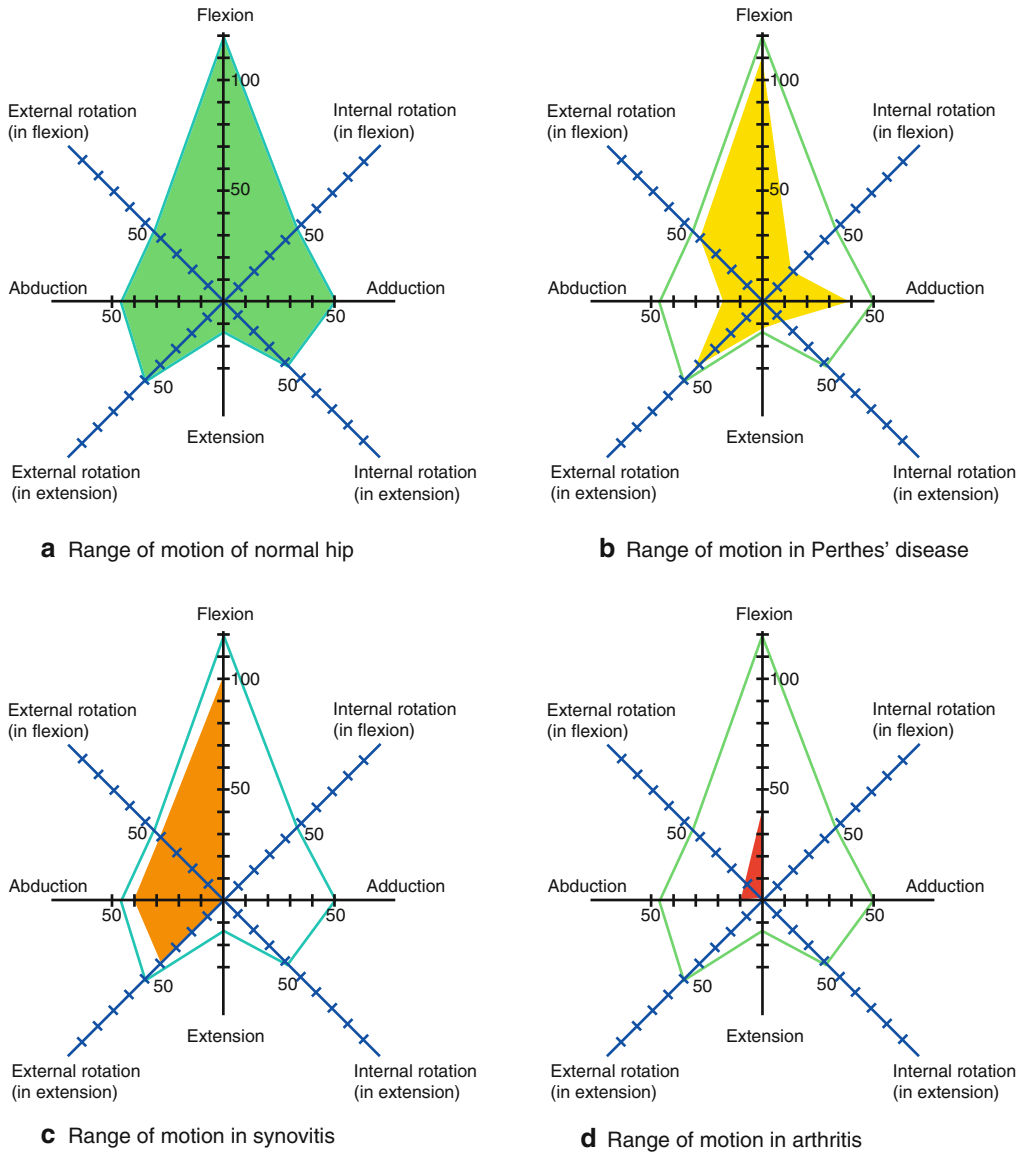

Fig. 32.3 The pattern of restriction of hip motion varies with the underlying pathology in the hip, and knowledge of these patterns is helpful in arriving at a diagnosis. (**a**) Range of motion of normal hip. (**b**) Range of motion in Perthes' disease. (**c**) Range of motion in synovitis. (**d**) Range of motion in arthritis

ischiofemoral ligaments get taut). The hip adopts a position of flexion, abduction, and external rotation. This "position of ease" relaxes the capsule which now can accommodate the maximum volume of synovial fluid. Consequently there is selective reduction of the range of extension, adduction, and internal rotation (Fig. 32.3c). Severe global restriction of motion (in all planes) may occur on account of severe pain of an acutely inflamed hip of septic arthritis, after other forms of arthritis with extensive cartilage destruction or due to extreme capsular contracture (Fig. 32.3d).

Gait

If the child can walk, observe if the gait is antalgic and if there a Trendelenburg component indicating hip abductor weakness.

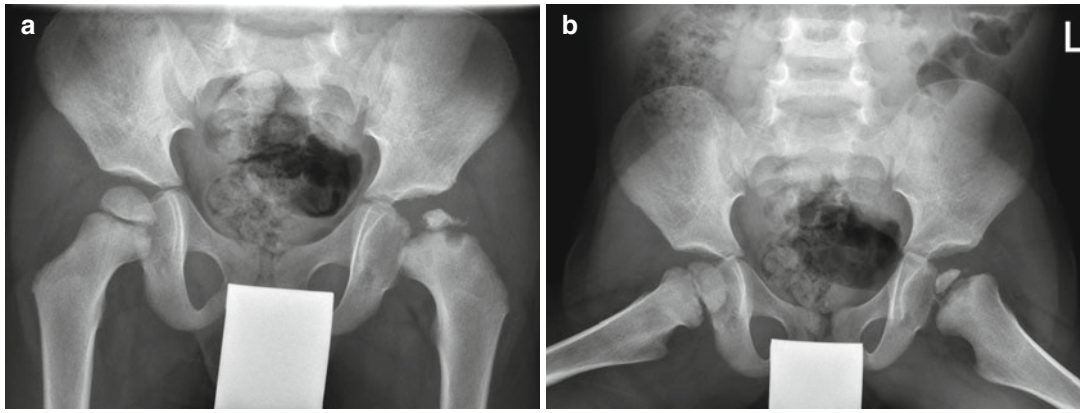

Fig. 32.4 Radiographs of a 5-year-old boy with Perthes' disease as seen on the AP (**a**) and frog lateral (**b**) views; note the epiphyseal collapse, sclerosis, and metaphyseal cysts

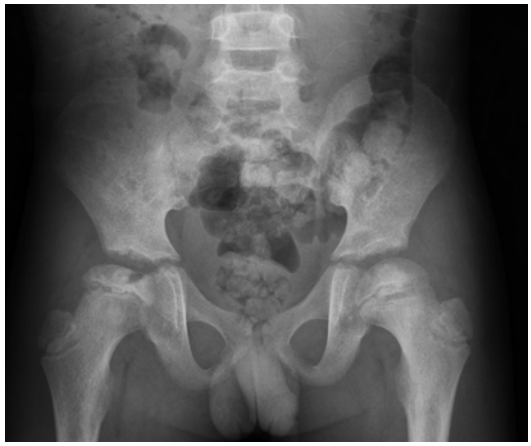

Fig. 32.5 Radiograph of an 8-year-old boy with sickle cell anemia and bilateral avascular necrosis

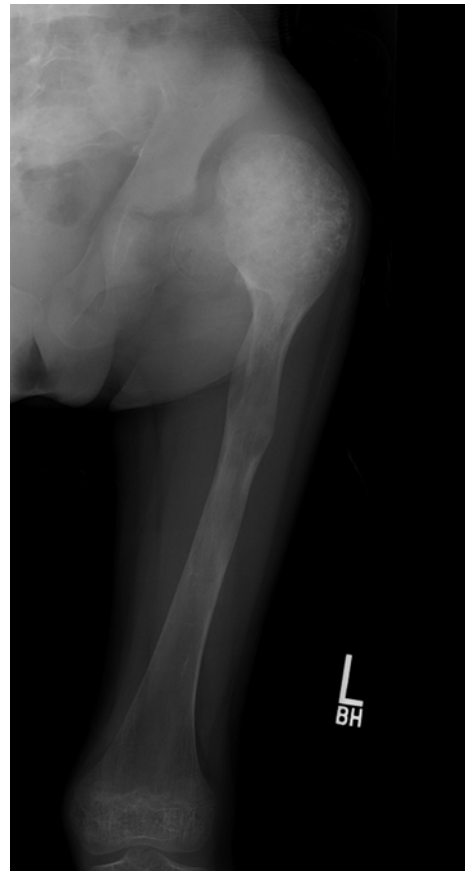

Fig. 32.6 AP radiograph of an 11-year-old child with hip pain and coxa vara with proximal femoral involvement due to fibrous dysplasia

32.4 Investigations to Confirm the Diagnosis

32.4.1 Plain Radiographs

Anteroposterior and frog lateral radiographs of the pelvis are mandatory when a child complains of hip pain (with the exception of situations where positioning for a frog lateral view is too painful) (Nnadi et al. 2002). Conditions like Perthes' disease, avascular necrosis associated with hemoglobinopathy or steroid intake, skeletal dysplasia, stress fractures, and cystic lesions of the femoral neck may be diagnosed on these radiographs (Figs. 32.4, 32.5, and 32.6). However, there may be very few subtle changes such as peri-articular osteopenia or no changes at all on the pelvic radiograph in transient synovitis or early tubercular synovitis.

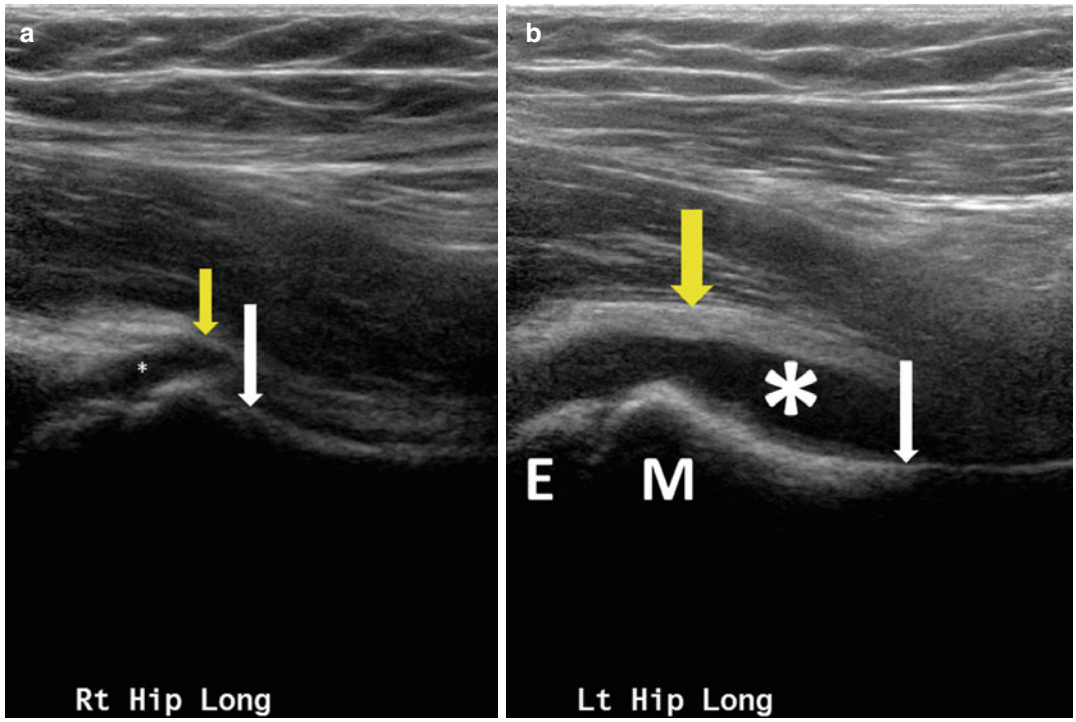

Fig. 32.7 A hip ultrasound of a child with toxic synovitis. The right hip is the normal hip, and the left hip is the one with toxic synovitis. The joint fluid is marked by the *asterisk*, the hip capsule by the *yellow arrow*, and the anterior metaphyseal cortex by the *white arrow*. *E* epiphysis, *M* metaphysis. (**a**) The normal hip with minimal joint fluid (*small asterisk*). (**b**) The irritable hip with a large effusion (*large asterisk*)

32.4.2 Ultrasound

Ultrasound examination is a valuable and sensitive imaging modality that can detect even small effusions in the hip (Fig. 32.7).

32.4.3 MRI Scan

When the plain radiograph is inconclusive, MR imaging may give more information. This modality is particularly useful for diagnosis of pelvic osteomyelitis, pyomyositis, and avascular necrosis

32.4.4 Isotope Bone Scan

Isotope bone scans are useful when the exact location of the pathology cannot be found on clinical exam and plain radiographs. A hot or cold bone scan will then allow for further focused imaging, e.g., MRI or CT scan. The difficulty with isotope scans is that they are sensitive but give little information regarding the anatomic findings of the pathologic process (Kocher et al. 2006).

32.4.5 Laboratory Investigations

The complete blood counts with differential, ESR and CRP, are required if concerned about sepsis; hemoglobin electrophoresis/sickle cell preparation if concerned about a hemoglobinopathy. In septic processes, the white blood count may or may not be elevated, but in typical pyogenic septic processes, it is elevated after several days. The CRP rises rapidly, usually within 6–12 h with any infectious process; thus it is an important laboratory study. The ESR takes longer to rise in an infectious process and is now used less commonly if the CRP is available. The CRP also drops quickly once appropriate therapy has been

instituted and can aid the clinician in this aspect as well (Kocher et al. 2006). Hemoglobin electrophoresis will demonstrate a hemoglobinopathy if present, e.g., sickle cell and thalassemia.

32.5 Differential Diagnosis

32.5.1 Septic Arthritis of the Hip

Septic arthritis of the hip is most commonly due to bacterial infection and may be an isolated process or in conjunction with an osteomyelitis or pyomyositis. There may also be other systemic septic processes, e.g., pneumonia and bacterial endocarditis. This is the most important condition to diagnose or exclude in the child with a painful hip, since if left untreated, the pyogenic process will rapidly destroy the articular cartilage and can also result in epiphyseal necrosis and frank hip dislocation (Samora and Klingele 2013). The child presents with significant limp and limitation of hip motion, often unable to walk, and will typically have a high temperature and elevated ESR/CRP and white count and appear systemically ill. The Kocher (Kocher et al. 1999) criteria have recently been described to assist in the diagnosis of this condition. If at all concerned about a septic hip, the next immediate step that must be taken is aspiration of the hip, with synovial fluid analysis. If the fluid is turbid and the child meets the Kocher criteria, then the diagnosis is confirmed, and therapy can be immediately instituted. Ultrasound will demonstrate effusion in the joint; a MRI scan will also demonstrate the effusion, and there will often be other changes in the femoral metaphysis (early osteomyelitis) or adjacent musculature (myositis) (Hammond and Macnicol 2001; Karmazyn et al. 2006, 2007; McPhee et al. 2007; Robben 2004). The Kocher criteria (Kocher et al. 1999, 2004) are a history of a temperature >38.5 °C, not able to bear weight, and erythrocyte sedimentation rate >40 mm/h and a serum white blood cell count >12,000 cells/mm^3. The probability of a child having a septic arthritis of the hip is 40 % if two of the criteria are met, 93.1 % for three, and 99.6 for all four. However, clinical judgment must be entertained, as the applicability of such criteria to other populations may not be as accurate (Luhmann et al. 2004).

32.5.2 Transient Synovitis

Transient synovitis is typically due to an interaction with antigens and antibodies from an upper respiratory infection which occurred in the recent past, either bacterial or viral. Physical examination will demonstrate mild limitation in hip motion; the child can usually walk but with a limp. The temperature is not elevated, and if there are any elevations in WBC or ESR/CRP, they are minimal. A hip ultrasound will demonstrate increased fluid in the involved hip (Fig. 32.7a, b). Aside from an effusion, a MRI scan will be negative (Do 2000; Nouri et al. 2013).

32.5.3 Perthes' Disease

Perthes' disease is an idiopathic osteonecrosis of the proximal femoral epiphysis, typically between the ages of 4 and 8 years in Caucasians but often older in Indo-Mediterranean peoples (Joseph et al. 1988; Loder and Skopelja 2011b). The child will often present with knee pain and a history of intermittent limp (Herring 2008). The child will demonstrate limitation of hip motion, especially internal rotation and abduction, along with an antalgic gait; there may be associated quadriceps atrophy. The child will otherwise appear healthy and afebrile, and laboratory studies will be negative. Early in the course of the disease, plain radiographs may be normal or simply demonstrate increased joint space, which actually represents a slightly smaller proximal femoral epiphysis. Later the typical changes of epiphyseal collapse and fragmentation will be seen (Fig. 32.4a, b). MRI scans are rarely needed in the diagnosis of this condition.

32.5.4 Tuberculosis of the Hip

In the early stages of tuberculosis of the hip, the features are those of synovitis (Campbell and

Hoffman 1995; Moon et al. 2012). The hip lies in flexion, abduction, and external rotation. This is the stage of apparent limb lengthening. If untreated, the disease will progress, early cartilage destruction occurs, and the hip becomes more painful. Hip flexor and adductor muscle spasm supervenes, and then fixed adduction and flexion deformities develop. This stage is referred to as the stage of apparent shortening. Further destruction of the femoral head or pathologic subluxation of the femoral head with acetabular destruction can result in true shortening. At this stage the hips remains flexed and adducted with gross painful limitation of motion.

32.5.5 Juvenile Idiopathic Arthritis (JIA)

There are three types of onset of JIA: polyarticular (five or more joints affected at initial presentation), oligoarticular (<5 joints affected), and systemic (fever, rash, lymphadenopathy, and hepatosplenomegaly). Oligoarticular onset is the commonest pattern of onset, accounting for about 50 % of cases; hip involvement at the onset is distinctly uncommon in this form of JIA (Malleson and Beauchamp 2001). The hip is commonly affected in severe destructive polyarticular juvenile idiopathic arthritis with up to 50 % of children having hip involvement (Malleson and Beauchamp 2001; Spencer and Bernstein 2002). Children with oligoarticular JIA are prone to develop uveitis, and since it is asymptomatic, it must be screened for as soon as a diagnosis of JIA is made.

32.5.6 Stress Fracture of the Neck of the Femur

Though stress fractures of the femoral neck in children are not common, reports of this entity have appeared with regular frequency in the literature (Fiévez et al. 2012; Lehman and Shah 2004; Maezawa et al. 2004; Scheerlinck and De Boeck 1998). Compression and tension stress fractures of the femoral neck have been reported. In the majority of instances, the fracture is incomplete and not displaced but can very occasionally become complete and then displace (Scheerlinck and De Boeck 1998).

32.5.7 Osteomyelitis (Proximal Femur and Pelvis)

These children will present nearly the same as a child with a septic hip, and it is difficult to differentiate the two until an MRI scan is performed. An early osteomyelitis without an abscess may initially be treated with IV antibiotics alone, thus the importance to establish the diagnosis early (Fig. 32.8).

Juvenile Slipped Capital Femoral Epiphysis

Slipped capital femoral epiphysis in the school-age child is rare; they typically present with groin/thigh/knee pain, an intermittent limp, and usually with no history of trauma. The patient will demonstrate reduced hip motion, especially internal rotation. Plain radiographs will confirm the diagnosis. It is important to diagnose this condition, as they are frequently associated with metabolic conditions which also need to be treated (e.g., hypothyroidism) (Loder and Greenfield 2001; Loder and Skopelja 2011a; Loder et al. 1995) (Fig. 32.9).

32.5.8 Osteoid Osteoma

Osteoid osteoma is an intensely inflammatory condition pathologically described as a small nidus with an intense osteoblastic reaction around the nidus. They can cause significant pain, especially at night. The classic diagnostic hallmark is relief by aspirin. They are very small and frequently not seen on plain radiographs. This is where an isotope bone scan is extremely helpful, as the osteoid osteoma is intensely hot on such a scan. Once the local has been localized, a CT scan of the area will confirm the diagnosis (Frassica et al. 1996; Kitsoulis et al. 2006) (Fig. 32.10).

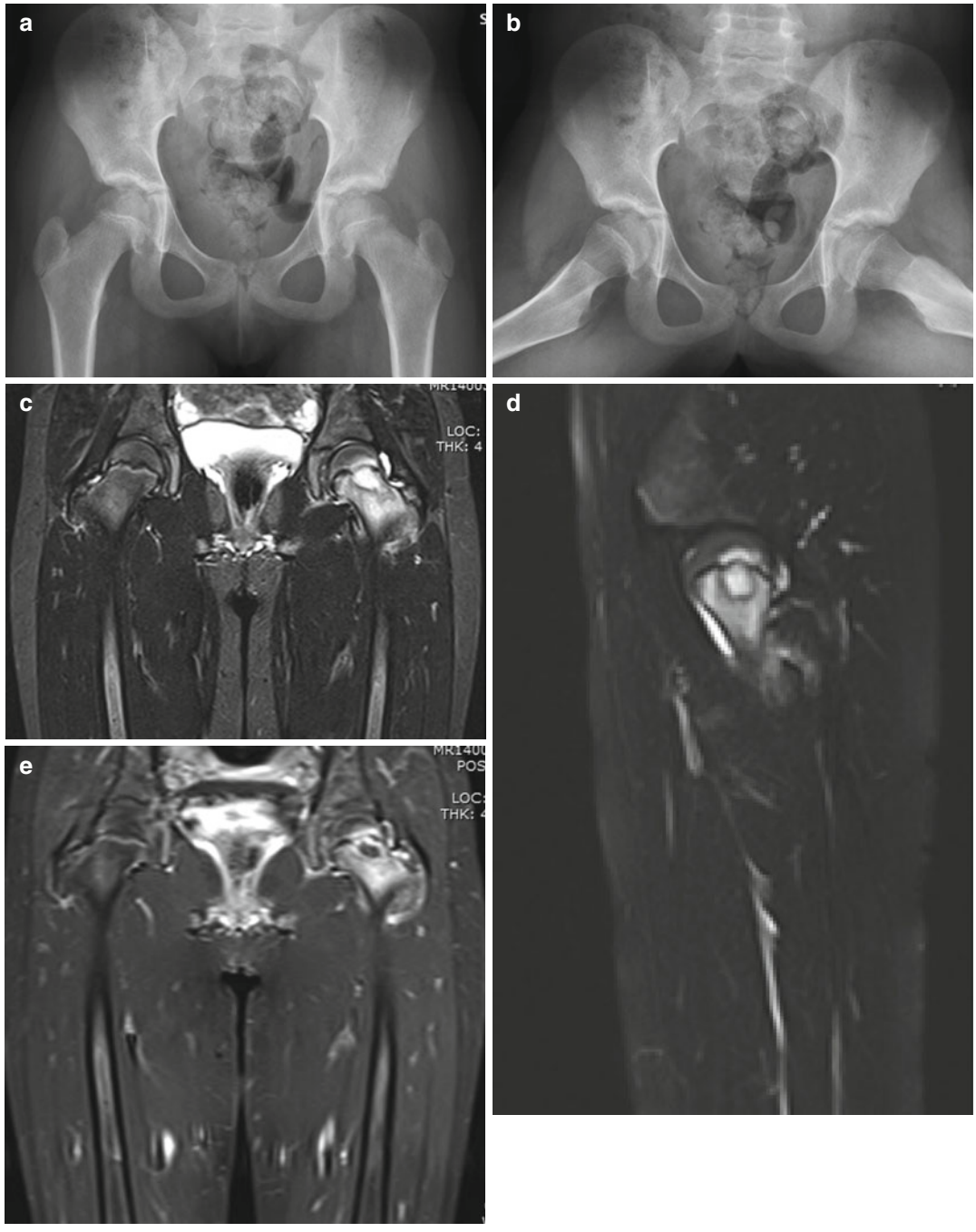

Fig. 32.8 Proximal femoral subacute osteomyelitis in a 5-year-old girl. Plain AP (**a**) and frog lateral (**b**) radiographs of the pelvis demonstrate a very subtle lucency in the proximal (*left*) femoral metaphysis just distal to the physis. MRI demonstrates osteomyelitis. The coronal STIR (**c**) and sagittal T2 (**d**) images demonstrate the edema in the lesion and surrounding bone including the epiphysis; the sagittal T1 fat suppression after gadolinium contrast (**e**) demonstrates no vascularity in the abscess

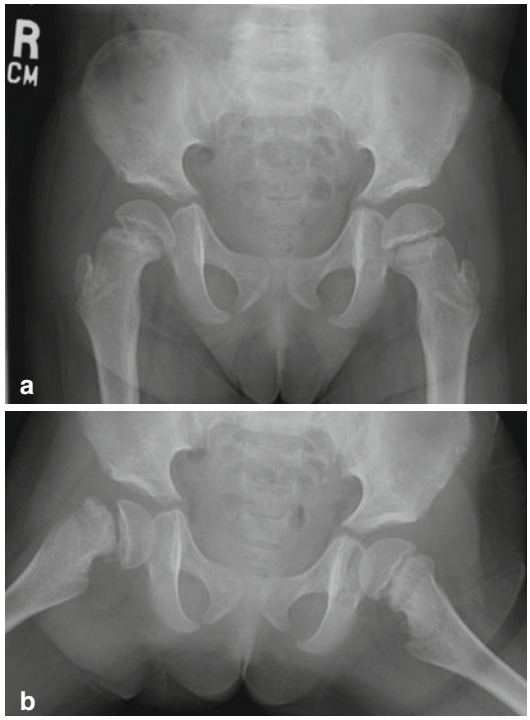

Fig. 32.9 A juvenile SCFE in a 7-year-old child who had been given growth hormone secondary to growth hormone deficiency resulting from total body radiation for a malignancy. Note the moderate SCFE of the right hip. (**a**) AP view. (**b**) Frog lateral view

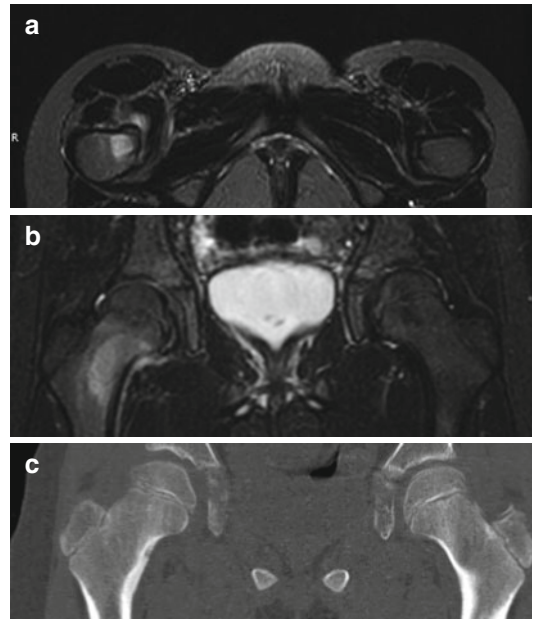

Fig. 32.10 An osteoid osteoma of the femoral neck in an 8-year-old girl with a 2-month history of right thigh and groin pain without any fevers. It was relieved by NSAIDs. Axial (**a**) and coronal (**b**) MRI sequences demonstrate the intense inflammatory response. Coronal CT scan reconstruction demonstrates the cortical thickening around the lucent lesion with its small ossific nidus (**c**)

32.5.9 Pyomyositis

These children will present nearly the same as a child with a septic hip, and it is difficult to differentiate the two until an MRI scan is performed. An early pyomyositis without an abscess may initially be treated with IV antibiotics alone, thus the importance to establish the diagnosis early (Kern et al. 2006; Lambertucci et al. 2001; Robben 2004; Wong-Chung et al. 2004).

32.5.10 Overuse Injuries (Muscle Strains, Apophysitis, Avulsion Injuries)

These are rare in the school-age child and more common in the adolescent (see Chap. 36).

32.6 Establishing the Diagnosis

An approach to the diagnosis of the cause of hip pain in children in the school-going age is shown in Tables 32.3, 32.4, and 32.5

Table 32.3 An outline of diagnosis of the cause of *unilateral hip pain of acute onset* in a child

History			
Prodromal symptoms may or may not have been present	Often prodromal symptoms (upper respiratory) present a few days prior to onset of hip pain	No prodromal symptoms	No prodromal symptoms
Usually no history of trauma (but occasionally may be present)	No history of trauma	History of severe trauma	History of trivial trauma
Fever (often high-grade with chills)	No fever	No fever	No fever
Unable to bear weight on the limb usually (in the very early stage the child may be able to bear weight on the limb)	May or may not be able to bear weight on the limb	Unable to bear weight on the limb	Unable to bear weight on the limb
Physical examination			
Febrile	Not febrile	Not febrile	Not febrile
Increased warmth over the joint	No increased local warmth	No increased local warmth	No increased local warmth
Tenderness over the joint line ++	Tenderness over the joint line ±	Tenderness over the joint line ++	Tenderness over the joint line ++
Fixed deformities often present Flexion, abduction, external rotation initially Flexion, adduction, and internal rotation later	Fixed deformities may be present Flexion, abduction, external rotation	Fixed deformity present External rotation deformity or Flexed, adducted and internally rotated	Fixed deformity present External rotation deformity
Apparent lengthening will be present if there is an abduction deformity Apparent shortening will be present if there is an adduction deformity True shortening may develop as a late feature due to pathologic subluxation or dislocation of the hip	Apparent lengthening may be present due to fixed abduction deformity	True supratrochanteric shortening	True supratrochanteric shortening
Hip movements: Grossly painfully limited in all directions	Hip movements: Abduction, flexion, and external rotation not painful Adduction, extension, and internal rotation painfully restricted	Hip movements: Grossly painfully limited in all directions	Hip movements: Grossly painfully limited in all directions
Investigations			
Plain radiograph of the pelvis: May show: Increase in the medial joint space Distended capsular shadow Erosion of the metaphysis of the femur	Plain radiograph of the pelvis: May show: Increase in the medial joint space Distended capsular shadow	Plain radiograph of the pelvis: Will show: Fracture of the neck of the femur No abnormality of bone texture in the region of the fracture or Posterior dislocation of the hip	Plain radiograph of the pelvis: Will show: Fracture of the neck of the femur Pathologic changes in the femoral neck (cyst/erosive lesion)

(continued)

Table 32.3 (continued)

Ultrasound: Will show effusion in the joint	Ultrasound: Will show effusion in the joint	Ultrasound not indicated	Ultrasound not indicated
MRI: Effusion Signal change may be seen in the femoral head Metaphyseal osteomyelitis if present will be delineated	MRI: Effusion	MRI not indicated	MRI may show the nature of the pathologic lesion in the femoral neck
Laboratory investigations: White cell count elevated (neutrophilia) ESR raised CRP raised Synovial aspirate Cell count over 50,000/ml with predominance of neutrophils Bacteria present	Laboratory investigations: White cell count not elevated ESR normal CRP normal Synovial aspirate Cell count below 50,000/ml no neutrophilic predominance No bacteria	Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis
Diagnosis			
Septic arthritis of the hip	Transient synovitis	Traumatic fracture neck of femur or traumatic hip dislocation	Pathologic fracture neck of femur

Table 32.4 Outline of diagnosis of the cause of *unilateral hip pain with an insidious onset* in a child

History			
Prodromal symptoms often present	Often prodromal symptoms (upper respiratory) present a few days prior to onset of hip pain	No prodromal symptoms	No prodromal symptoms
Usually no history of trauma (but occasionally may be present)	No history of trauma	History of severe trauma	History of trivial trauma
No fever	May have a low-grade fever	No fever	No fever
Can usually bear weight on the limb, with or without an antalgic gait and occasionally a Trendelenburg limp	Usually able to bear weight on the limb with or without an antalgic gait	Able to bear weight	Can usually bear weight on the limb, with or without an antalgic gait and occasionally a Trendelenburg limp
Physical examination			
Not febrile	Intermittent low-grade fever	Not febrile	Not febrile
No increased local warmth	Increased warmth over the joint	No increased local warmth	No increased local warmth
No tenderness over the joint line	Tenderness over the joint line ±	No tenderness over the joint line	No tenderness over the joint line
Fixed deformities often present Flexion, abduction, external rotation initially	Fixed deformities may be present Flexion, abduction, external rotation	Fixed deformity not present	Varying degrees of fixed deformity present Flexion and external rotation deformity

Table 32.4 (continued)

Apparent shortening will be present if there is an adduction deformity True shortening may develop as a late feature due to collapse of the epiphysis	Apparent shortening will be present if there is an adduction deformity True shortening may develop as a late feature due to destruction of the joint	Shortening not present	Varying degrees of true femoral shortening
Hip movements: Varying degrees of irritability with loss of abduction, internal rotation, and extension	Hip movements: Varying degrees of irritability with loss of abduction, internal rotation, and extension	Hip movements: Varying degrees of irritability with loss of internal rotation primarily	Hip movements: Varying degrees of loss of internal rotation, adduction, and extension; obligatory external rotation with increasing passive flexion (Drehmann sign)
Investigations			
Plain radiograph of the pelvis: Early on may show: Increase in the medial joint space Distended capsular shadow Subchondral crescent fracture Later on will show Varying degrees of epiphyseal fragmentation and collapse, along with extrusion	Plain radiograph of the pelvis: Early on may show: Increase in the medial joint space Distended capsular shadow Subchondral crescent fracture Later on will show Varying degrees of epiphyseal fragmentation and collapse, along with extrusion ± Acetabular erosions	Plain radiograph of the pelvis: Is often normal May see increased cortical thickening in the proximal femoral cortex	Plain radiograph of the pelvis: Will show: Physeal widening Varying degrees of epiphyseal slip relative to the metaphysis, usually in an inferior and medial direction In the long-standing case may also see remodeling of the superior and anterior metaphysis
Ultrasound: May show an effusion in the joint	Ultrasound: May show an effusion in the joint	Ultrasound not indicated	Ultrasound not indicated
MRI: Effusion, loss of epiphyseal vascularity on gadolinium enhanced sequences Bone scan: will demonstrate epiphyseal photopenia in the early avascular phase	MRI: Effusion, varying degrees of epiphyseal/metaphyseal and acetabular destruction Bone scan: will demonstrate varying amounts of decreased and increased uptake	MRI: Will demonstrate edema and inflammatory signals in the surrounding marrow Bone scan: Often demonstrates intense local uptake CT: Will demonstrate the cortical thickening; thin cuts will demonstrate the nidus and surrounding lucency	MRI not usually indicated Bone scan not indicated CT scan not indicated
Laboratory investigations: Normal white cell count ESR normal CRP normal Synovial aspirate not needed	Laboratory investigations: White cell mildly elevated ESR raised CRP raised Synovial aspirate Gram stain will demonstrate AFBs and cultures will grow mycobacterium	Laboratory investigations not indicated to establish diagnosis Synovial aspirate not needed	Laboratory investigations often needed to rule out hypothyroidism or endocrine disorder Synovial aspirate not needed
Diagnosis			
Legg-Calvé-Perthes' disease	Tuberculosis of the hip	Osteoid osteoma	Juvenile SCFE

Table 32.5 Outline of diagnosing cause of *bilateral hip pain* in a child

History			
No positive family history	No positive family history	Family history may be present	No positive family history
Hip symptoms: Usually metachronous (symptoms in one hip precedes the other)	Hip symptoms: Synchronous or metachronous	Hip symptoms: Frequently synchronous	Hip symptoms: Frequently synchronous
No other joint involved	No other joint involved	Other joints may be deformed or painful	Other joints painful/swollen/deformed
No steroid intake	History of medication with steroids for any other disease may be present	No steroid intake	No steroid intake (unless administered for the joint symptoms)
Physical examination			
Stature: Normal or very slightly short	Stature: May be short	Stature: Often short with altered body proportions	Stature: Usually normal at onset of symptoms
No increased warmth over the hips	No increased warmth over the hips	No increased warmth over the hips	Warmth may be noted over the hips, knees, and ankles
Fixed deformities: Infrequently seen Symmetric deformities very rare	Fixed deformities: Infrequently seen	Fixed deformities: Flexion deformity frequent Symmetric deformities common	Fixed deformities: Flexion deformity frequent Symmetric deformities common
Range of motion: Selective reduction in abduction and internal rotation The extent of reduction would depend on the stage of evolution of the disease	Range of motion: Reduction in the range of motion variable; often rotations are reduced	Range of motion: Reduction in the range of motion variable; progressive reduction in the range of motion may be seen as the child grows	Range of motion: Initially adduction, internal rotation, and extension are reduced; later global restriction of motion will be present
Investigations			
Plain radiograph: Features of Perthes' disease in different stages of evolution in each hip	Plain radiograph: Sclerosis suggestive of avascular necrosis of the hips	Plain radiograph: Epiphyseal and/or metaphyseal changes with narrowing of joint space	Plain radiograph: Peri-articular osteopenia Varying degrees of joint space reduction and irregularity of the articular margin
Hemoglobin electrophoresis to rule out hemoglobinopathy will be negative	Hemoglobin electrophoresis to rule out hemoglobinopathy will demonstrate the abnormal hemoglobin in sickle cell disease and thalassemia	Not indicated	Not indicated
Thyroid function tests to rule out hypothyroidism are normal	Thyroid function tests to rule out hypothyroidism are normal	Not indicated	Not indicated
Diagnosis			
Bilateral Perthes' disease	Avascular necrosis of the femoral head secondary to a hemoglobinopathy or secondary to steroid administration	Skeletal dysplasia with hip involvement	Juvenile idiopathic arthritis

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Short Stature and Altered Body Proportions

33

Benjamin Joseph

33.1 Introduction

A child considered to be "short" may be brought to an orthopedic surgeon or pediatrician to find out if his or her stature can be increased. Before embarking on any investigations, it is necessary to determine if the child does, in reality, have short stature (Vogiatzi and Copeland 1998). The most widely accepted definition for short stature is height for age that is less than two standard deviations below the average for that gender. This is shown on standard growth charts as falling below the 3rd centile (Fig. 33.1). Once it is established that the stature of the child is indeed short, then the distinction needs to be made between non-pathologic familial short stature and pathologic short stature (Vogiatzi and Copeland 1998; Seaver and Irons 2009).

33.2 Questions to Establish a Diagnosis

- **What is the absolute height of the child?**
- **What is the growth velocity?**
- **Is the weight of the child normal and what is the weight-to-height relationship?**
- **Are the parents of the child also short?**
- **When was the short stature noted? Was the child small from birth or was the short stature appreciated later?**
- **Are the body proportions normal?**
- **Are there obvious physical defects apart from the short stature?**

What is the absolute height of the child?

The standing height of the child needs to be measured at the outset and the height plotted on a gender-specific growth chart for the same ethnic population. While a height below the 3rd centile would be deemed as short stature, it does not necessarily indicate that the growth is pathologic.

What is the growth velocity?

Growth observed over a period of 6 months will give more valuable information than a single measurement of absolute height. A significant reduction in growth velocity in children between 3 and 12 years should be considered pathologic.

Is the weight of the child normal and what is the weight-to-height relationship?

Endocrine disorders that result in short stature are usually associated with either normal weight or obesity. On the other hand, chronic gastrointestinal, renal, cardiac, and pulmonary diseases are associated with low weight, and

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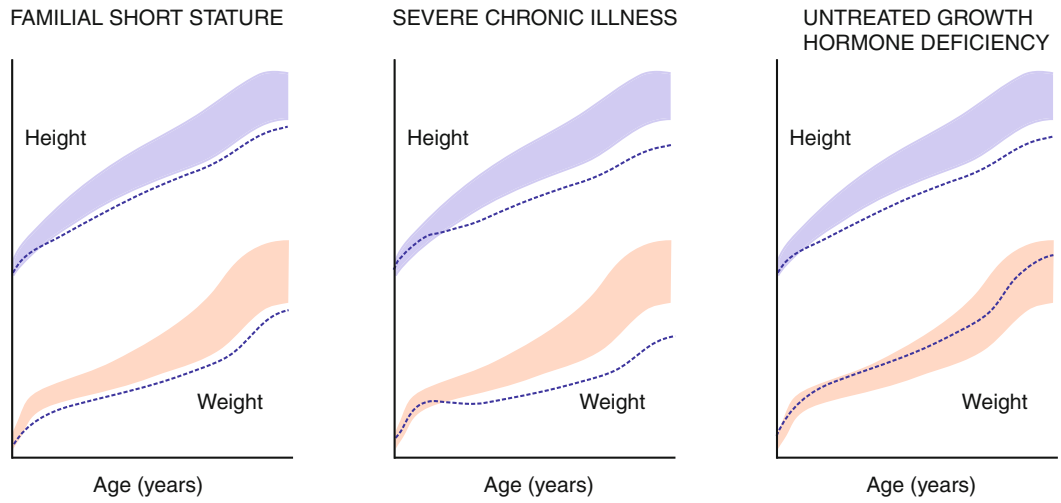

Fig. 33.1 The height and weight of the child are plotted on growth charts. The pattern of growth differs for each type of dwarfism

these children are, in fact, thin even for their short stature (Fig. 33.1).

Are the parents short and what is the predicted target height at maturity?

If the parents are short, the child may have familial short stature. It is important to measure the height of the parents. This will enable a rough estimate of the target adult height of the child using these formulae:

$$\left(\text{Father's height} + \text{Mother's height in cm} + 13\right)/2 \text{ for boys}$$

$$\left(\text{Father's height} + \text{Mother's height in cm} - 13\right)/2 \text{ for girls}$$

When was the short stature noted?

Short stature evident at birth may be on account of being small for gestational age (SGA) or due to intrauterine growth retardation (IUGR). Short stature also manifests at birth in certain skeletal dysplasias (e.g., achondroplasia). In the majority of instances, the short stature may manifest later on during childhood.

Are the body proportions normal?

The sitting height provides a measure of the upper segment of the body (trunk and head). If appropriate equipment for measurement of sitting height is not available, the pubis-to-heel length can be measured and subtracted from the standing height to give a measure of the upper segment. The upper segment/lower segment ratio alters with age as shown in Table 33.1. In general, if symmetric disproportion of the body segments is demonstrated, it may be assumed that the child has a skeletal dysplasia.

Table 33.1 Anthropometric body proportions in different ages

Age	Upper segment/lower segment ratio
Newborn	1.7
2–8 years	~1.0
Adult	0.95

If the child has short limbs relative to the trunk, the U/L ratio will be high, while a child with a short trunk and normal limbs will have a low U/L ratio. Normal body proportions are seen in many conditions with short stature, while abnormal body proportions are typically seen in some forms of skeletal dysplasia. Disproportion may be very evident on clinical examination (e.g., achondroplasia), but in some situations the disproportion is more subtle and will only be evident when anthropometric measurements are made and the upper and lower segment ratio calculated. Some skeletal dysplasias with disproportionate growth may manifest only later on in childhood, and hence a single set of measurements in early childhood may be misleading.

Are there obvious physical defects apart from short stature?

Facial dysmorphism and associated congenital anomalies are seen in children with chromosomal abnormalities and other syndromes. Intrauterine growth retardation is often a feature of some of these syndromes with short stature.

33.3 Physical Examination

33.3.1 Look

Look for facial dysmorphism, altered body proportions, and deformities of the knees, ankles, elbows, wrists, and hands. Note if there are deformities of the spine.

Look at:

- The hair (sparse in some conditions – Fig. 33.2a)
- Eye (refractory errors)
- Palate (cleft palate, high-arched palate – Fig. 33.2b)
- Dentition (abnormal dentition – Fig. 33.2c)
- Spine (kyphosis, scoliosis)
- Upper and lower limbs (deformities of bone or joints, polydactyly, growth abnormalities)

33.3.2 Feel

Palpate the abdomen for hepatosplenomegaly.

Fig. 33.2 Sparse hair in a child with a form of skeletal dysplasia (**a**), high-arched palate in a child with a syndrome associated with short stature (**b**) and abnormal dentition in a child with a skeletal dysplasia (**c**)

33.3.3 Measure and Plot

Anthropometric measurements taken when the child presents may give useful insight into the possible cause of short stature in some instances, while the cause may not be evident in others. Plotting the same measurements repeated over a span of 6 months, however, can provide invaluable information regarding the linear growth velocity. The following measurements are essential:

- Height
- Weight
- Sitting height
- Upper segment/lower segment ratio
- Arm span

33.4 Investigations to Confirm the Diagnosis

33.4.1 Radiographs

A plain radiograph of the left hand and wrist is needed to determine the skeletal age of the child. The status of carpal ossification is compared against the normal from the Greulich and Pyle atlas (Greulich and Pyle 1959) or the Tanner and Whitehouse atlas (Tanner et al. 1975) to establish the bone age.

If a skeletal dysplasia is suspected, a skeletal survey is performed; this includes:

- Anteroposterior and lateral views of the skull.
- Anteroposterior and lateral views of the entire spine.
- Posteroanterior view of the chest and ribs.
- An anteroposterior view of the pelvis and hips, at least one upper limb and one lower limb (if asymmetric deformities are present, both sides must be imaged).
- Lateral view of the knee may be added if needed.
- Lateral view of the ankle may be added if needed.

If any surgical intervention is contemplated in a child with a suspected skeletal dysplasia, flexion and extension views of the cervical spine are essential to exclude occult upper cervical spine instability (Redl 1998).

33.4.2 Endocrine Tests

If an endocrine cause for short stature is suspected, estimation of serum T4, TSH, IFG-1, and IGFB-3 concentrations is done.

33.4.3 Other Tests

Renal function tests and tests for malabsorption and other chronic disease are indicated as appropriate if a chronic illness is suspected.

33.5 Differential Diagnosis of Short Stature

33.5.1 Hormone Deficiency

Growth hormone deficiency is characterized by delayed physical growth and delayed puberty. The slow growth may not be evident till the age of 2 or 3 years. Body proportions are normal and the child is of normal intelligence.

33.5.2 Primary Systemic Disease

Renal, gastrointestinal, hepatic, or other chronic systemic disease can lead to stunting of growth and reduced stature. In many instances the symptoms of the underlying disease would be evident. Occasionally, the cause of the retardation of growth may be unclear; these children need to be investigated for conditions such as inflammatory bowel disease, renal tubular dysfunction, and nephrogenic diabetes insipidus.

33.5.3 Skeletal Dysplasia

Skeletal dysplasias are a heterogeneous group of more than 400 genetic disorders characterized by abnormalities of cartilage and bone growth. Though cartilage and bone growth affects the whole skeleton, the growth aberration may be most evident in one region of the skeleton in a particular dysplasia. This results in characteristic alterations in body proportions often facilitating the diagnosis of the underlying condition (Fig. 33.3). The femur and humerus are most severely affected and demonstrate the most shortening in rhizomelic dwarfism as in achondroplasia and rhizomelic chondrodysplasia punctata (Fig. 33.4a, b). The forearm or leg (radius, ulna, tibia, and fibula) is most affected in mesomelic dysplasias as in Langer mesomelic dysplasia (Fig. 33.5), and more rarely, growth of the hand and foot is predominantly retarded in acromelic dysplasias.

Scrutiny of the radiographs will clarify which part of the bones is primarily affected; the epiphysis is frequently affected, while the metaphysis or the diaphysis may be abnormal in some dysplasias. Accordingly the dysplasia is termed as epiphyseal dysplasia, metaphyseal dysplasia, or diaphyseal dysplasia. If the spine is involved with reduction in the height of the vertebrae (platyspondyly) in addition to changes in the epiphysis or metaphysis, the terms spondyloepiphyseal or spondylometaphyseal dysplasia are used to specify the pattern of involvement (Fig. 33.6). Rarely the spine alone is affected with no demonstrable changes in the epiphysis or metaphysis of long bones; this form of short-trunk dwarfism is referred to as brachyolmia (Gardner and Beighton 2003).

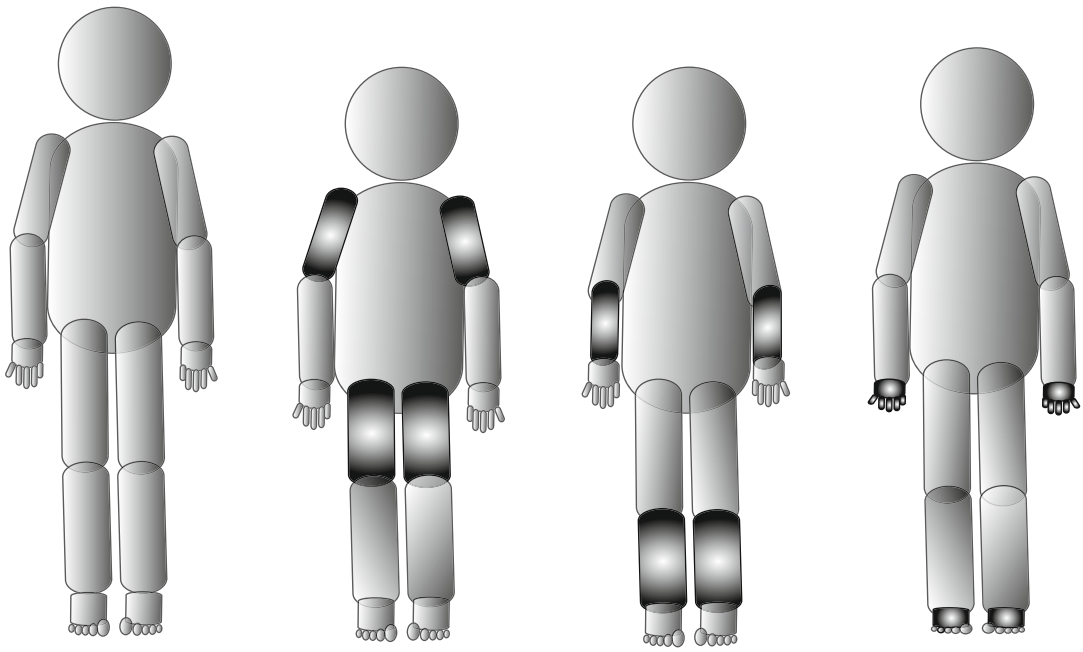

Fig. 33.3 Patterns of growth retardation in different forms of skeletal dysplasia

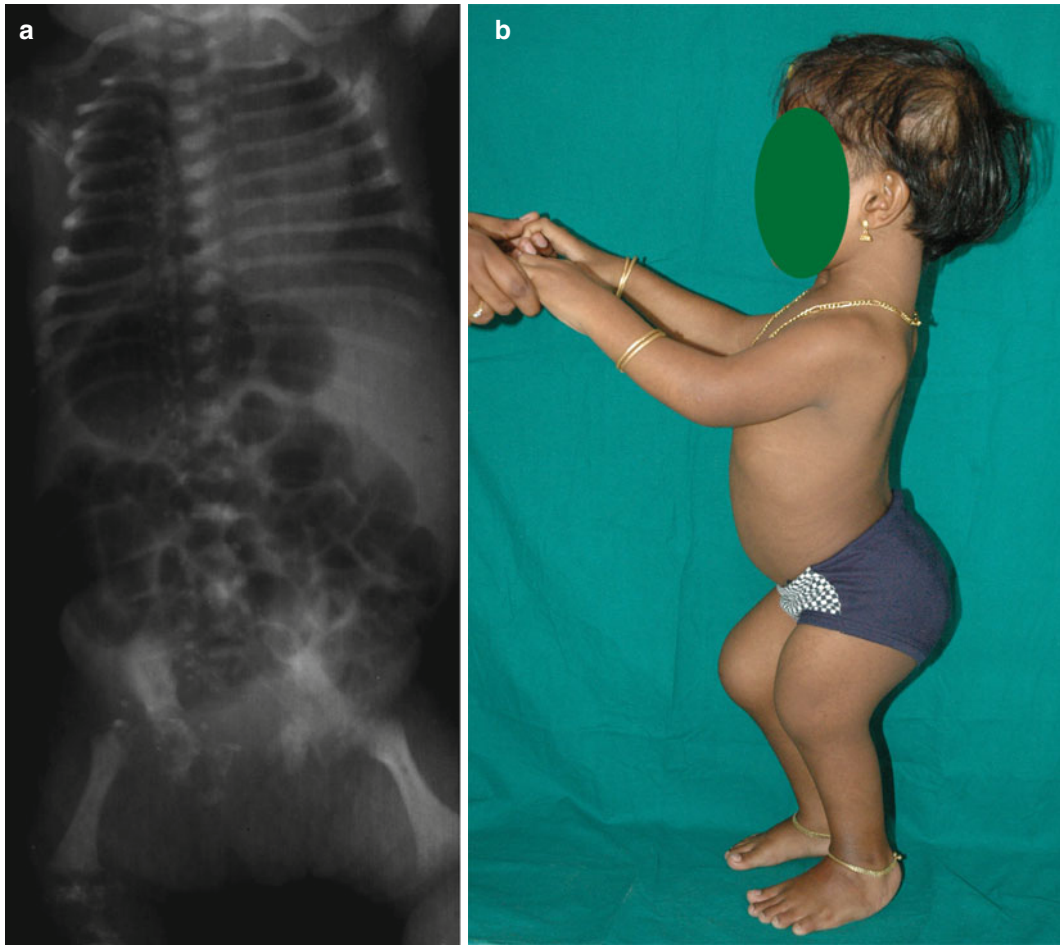

Fig. 33.4 Punctate calcification seen in the epiphysis (**a**) of a child with rhizomelic chondrodysplasia punctata (**b**)

The radiological and clinical characteristics of the spectrum of skeletal dysplasias are summarized in some excellent reviews (Krakow and Remoin 2010; Unger 2002; Offiah and Hall 2003; Rimoin et al. 2007; Alanay and Lachman 2011).

33.5.4 Syndromes with Short Stature

Short stature is one of the features of several dysmorphic syndromes; the relatively more common of these syndromes include Turner syndrome,

Fig. 33.5 Mesomelic dwarfism

Noonan syndrome, Prader-Willi syndrome, Silver-Russell syndrome, and Aarskog-Scott syndrome (Siklar and Berberoglu 2014). Marked intrauterine growth retardation is a characteristic feature of Silver-Russell syndrome, and consequently dwarfism is evident at birth. In many other syndromes, the growth retardation is postnatal.

33.5.5 Familial Short Stature

There is a family history of short stature. The rate of growth is below normal but proceeds parallel to the 5th percentile, and the bone age corresponds with the chronological age. The adult height is below normal but is comparable with the height of the short parent.

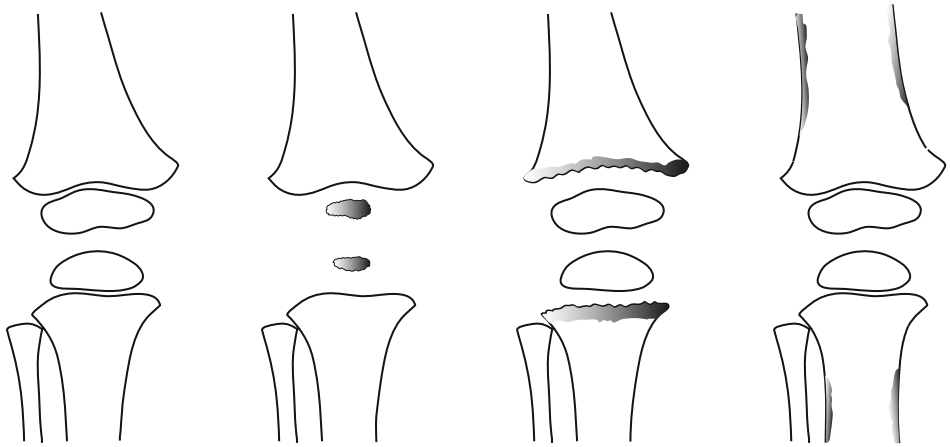

Fig. 33.6 Diagrammatic representation of changes in the bones in different forms of skeletal dysplasia. (**a**) Normal. (**b**) Epiphyseal dysplasia. (**c**) Metaphyseal dysplasia. (**d**) Diaphyseal dysplasia. (**e**) Normal. (**f**) Platyspondyly

33.6 Establishing the Diagnosis

The outline of establishing the cause of short stature is shown in Table 33.2.

Table 33.2 Establishing the cause of short stature in a child

History				
No family history of short stature	No family history of short stature	Family history of short stature may be present	Family history of short stature may be present	Family history of short stature often present
Short stature usually not evident at birth	Short stature not evident at birth	Short stature may be evident at birth	Short stature may be evident at birth	Short stature may be evident at birth
Physical examination				
Body habitus: Well nourished or obese	Body habitus: Thin child	Body habitus: Normal	Body habitus: Normal	Body habitus: Normal
Growth pattern: Deceleration of linear growth	Growth pattern: Deceleration of linear growth	Growth pattern: Normal	Growth pattern: Normal	Growth pattern: Normal (parallel to lower centiles of growth curve)
No facial dysmorphism	No facial dysmorphism	Facial dysmorphism	No facial dysmorphism	No facial dysmorphism
Normal body proportions	Normal body proportions	Normal body proportions	Altered body proportions	Normal body proportions
Working diagnosis: Endocrine cause for short stature	Working diagnosis: Chronic systemic disease leading to short stature	Working diagnosis: Syndrome associated with short stature	Working diagnosis: Skeletal dysplasia	Working diagnosis: Constitutional dwarfism or familial short stature
Investigations				
T4, TSH estimations	–	–	–	T3, T4, TSH estimations
IGF-1 and IGFB3 estimation	–	–	–	IGF-1 and IGFB3 estimation
–	Renal function tests Liver function tests Tests for GI malabsorption	–	–	–
–	–	–	Skeletal survey	–
Diagnosis				
Endocrine disorder Hypothyroidism (Raised TSH, low T4) Growth hormone deficiency (Normal thyroid function, low IGF-1 and IGFB3)	Chronic renal/gastrointestinal/hepatic disease	Syndrome associated with short stature	Skeletal dysplasia	Familial short stature

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Localized Gigantism of the Limbs

34

Benjamin Joseph

34.1 Introduction

Localized gigantism frequently manifests as macrodactyly (Fig. 34.1) though the overgrowth is seldom limited to the affected digits of the hand or foot (Fig. 34.2). Macrodactyly has been classified in three ways (De Laurenzi 1962; Temtamy and McKusick 1978; Flatt 1994); each classification has clinical relevance (Table 34.1).

There are several conditions associated with macrodactyly and more extensive localized gigantism; the questions that may help to establish the underlying condition are listed here.

34.2 Questions to Establish a Diagnosis

- **Was the gigantism noted at birth or in very early infancy?**
- **Is the gigantism static or progressive?**
- **Is the gigantism localized to the digit or digits?**
- **Does the gigantism involve one half of the body?**
- **Are there features of neurofibromatosis, hemangiomas, arteriovenous malformation, and constriction bands?**

Was the gigantism noted at birth or in very early infancy?

Some forms of gigantism are noted at birth or soon after birth, while others become evident in the young child. This may help in diagnosing the underlying pathology.

Is the gigantism static or progressive?

Static forms of gigantism are relatively easy to treat, while progressive forms can be exceedingly difficult to treat (Hardwicke et al. 2013).

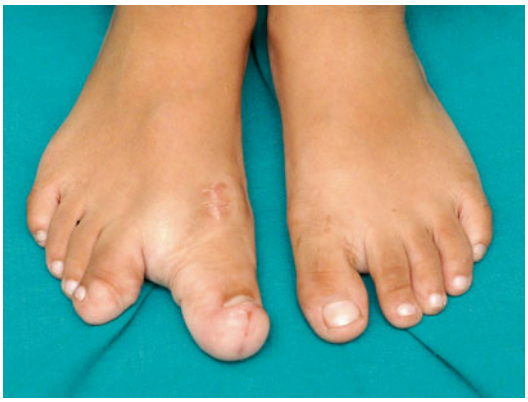

Fig. 34.1 Macrodactyly affecting the great toe and the second toe of the right foot. A debulking procedure and epiphysiodesis have already been performed. The overgrowth is restricted to the first and second ray

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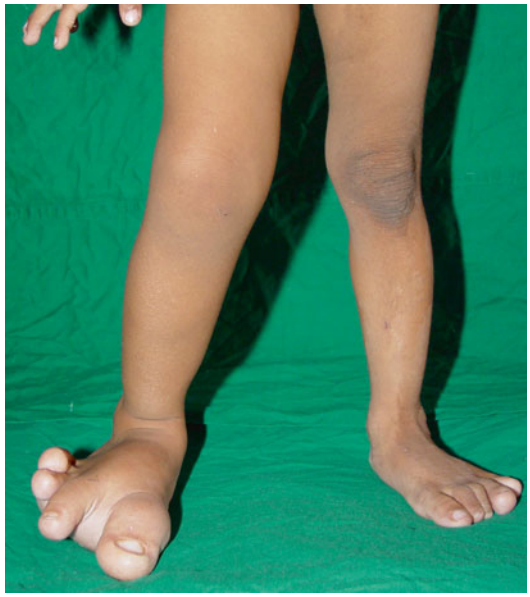

Fig. 34.2 Macrodactyly of the right foot in a young boy; the overgrowth involves the calf and thigh. There is a minor degree of overgrowth of the fourth toe on the left foot

Is the gigantism localized to the digit or digits?

Gigantism may involve one or two digits or may involve the entire limb; gigantism that predominantly involves the digits is relatively easy to treat.

Does the gigantism involve one half of the body?

Gigantism involving one half of the body is referred to as hemihypertrophy. It is a feature of Beckwith-Wiedemann syndrome. These children have a propensity to develop malignant tumors of the kidney and liver and must be screened for them.

Are there features of neurofibromatosis, hemangiomas, arteriovenous malformation, and constriction bands?

Treatment of a child with gigantism and an underlying condition such as a vascular abnormality may have to be directed to both the gigantism and the condition that resulted in overgrowth. The natural history and prognosis may vary with the underlying disorder.

Table 34.1 Classifications of macrodactyly

Classification	Basis for classification	Types
De Laurenzi (1962)	Growth rate of affected digit	1. Static macrodactyly Growth of the enlarged digit remains proportionate to the growth of the rest of the limb Presents at birth or early infancy Less common 2. Progressive macrodactyly Excessive and disproportionate to the growth of the rest of the limb Presents at about 2 years of age More common
Temtamy and McKusick (1978)	Syndromal association	1. Isolated anomaly (a) True macrodactyly Identified at birth or soon after May be static or progressive Involves soft tissue and bones (b) Pseudomacrodactyly Soft tissues primarily affected Seen in association with hemangiomas, arteriovenous malformations, edema in amniotic constriction bands 2. Macrodactyly as part of a syndrome
Flatt (1994)	Underlying disease process	1. Type I: gigantism in lipofibromatosis 2. Type II: gigantism in neurofibromatosis 3. Type III: gigantism with digital hyperostosis 4. Type IV: gigantism with hemihypertrophy

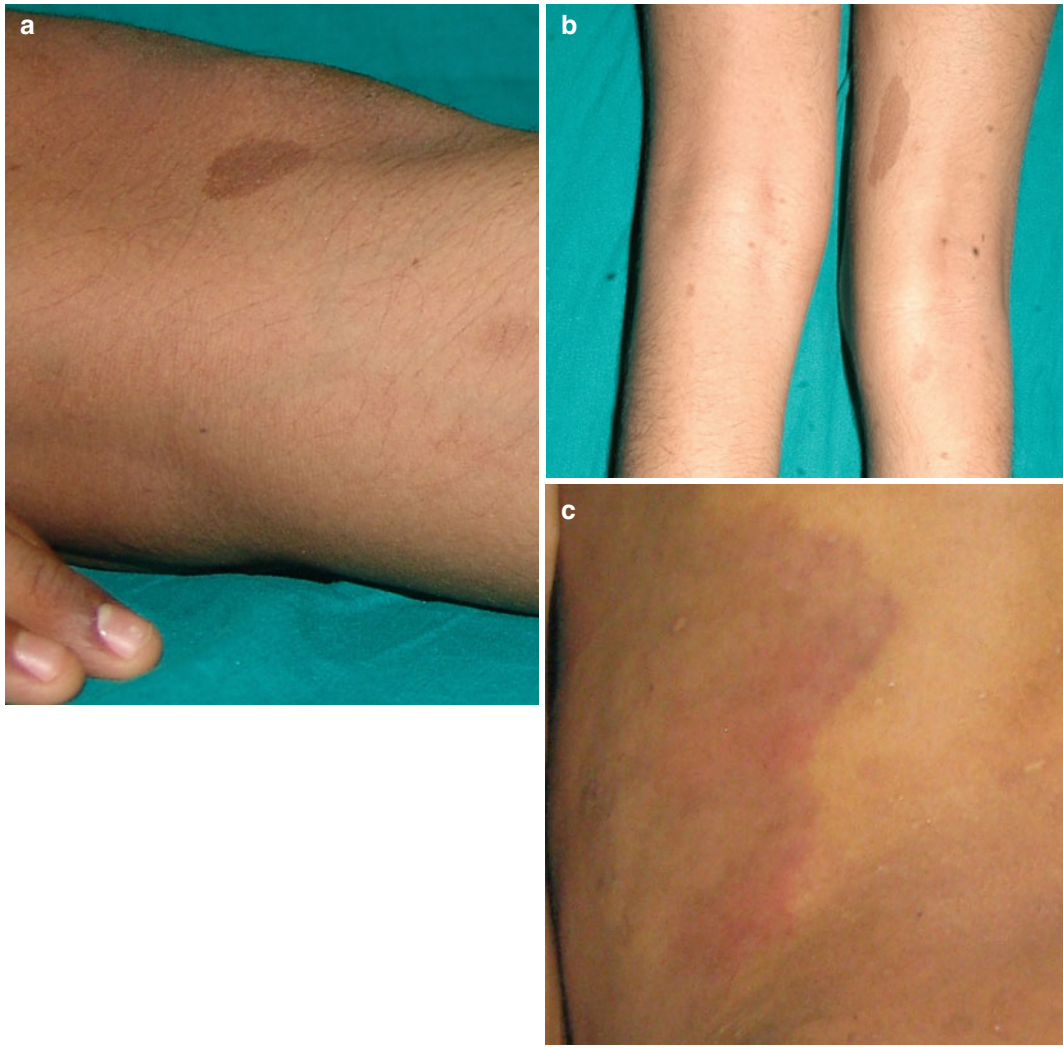

Fig. 34.3 Café-au-lait spots (**a**) and freckling (**b**) in children with neurofibromatosis and a port-wine stain (**c**) in a child with gigantism associated with vascular malformations

34.3 Physical Examination

34.3.1 Look

Observe the site of gigantism and note the proximal extent of overgrowth; note if the overgrowth affects both sides of the body and if the overgrowth is symmetric or asymmetric.

Examine the skin over the limbs and the torso for café-au-lait spots (Fig. 34.3a, b), pigmentation, capillary hemangiomas (Fig. 34.3c), epidermal verrucae, and cerebriform connective tissue nevi (CCTN).

Note if there are deformities of the spine (scoliosis), tibia (anterolateral bowing), and forearm (bowing); all of these may be suggestive of neurofibromatosis.

Observe if there are swellings in the limbs or trunk (hemangioma or plexiform neurofibroma) and note if there are varicosities of the superficial veins of the calf and thigh or forearm and arm (Fig. 34.4a, b).

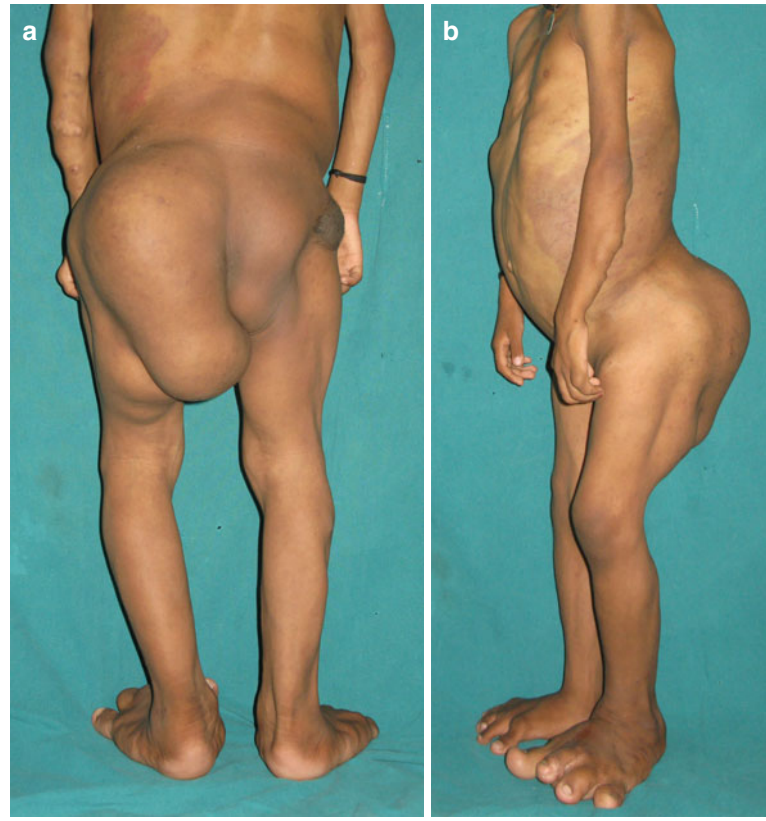

Fig. 34.4 (**a**, **b**) Enormous hemangiomas in a young boy with macrodactyly of the feet and local gigantism; varicose veins are present in the left upper limb

34.3.2 Feel

Palpate swellings on the limbs and trunk and note if they are compressible, soft swellings that decrease in size when the limb is elevated and increase when dependent (hemangioma). Note if there are palpable nodular swellings within the larger soft swelling (phleboliths). Note if there are bony swellings (enchondromas).

34.4 Investigations to Confirm the Diagnosis

Radiography
Plain radiographs are useful to determine the extent of skeletal hypertrophy and to exclude enchondromas. Radiographs of a bowed tibia are necessary to exclude a pseudarthrosis in patients with suspected neurofibromatosis.

Ultrasound
Abdominal ultrasound is essential as a screening method to exclude visceral malignancies.

MRI
MRI scans are useful in delineating the nature and extent of soft tissue abnormalities in some of these overgrowth syndromes (Jamis-Dow et al. 2004).

34.5 Differential Diagnosis

34.5.1 Gigantism and Lipofibromatosis

This is the commonest form of macrodactyly that affects multiple digits more frequently than a single digit (Fig. 34.1). The distribution of affected digits may be in the distribution of a peripheral nerve; involvement of digits supplied by the

median nerve is more frequent than the digits supplied by the ulnar nerve in the hand. The index finger is the most frequently affected followed by the middle finger and then the thumb. The medial toes tend to be affected more frequently than the lateral toes when the foot is affected (Bhat et al. 2005). The nerves to the overgrown digits may be enlarged, but nerve function is preserved and no cutaneous sensory abnormality is present.

34.5.2 Gigantism and Neurofibromatosis

This form of gigantism is associated with features of neurofibromatosis type I or II (NF-1 or NF-2). Macrodactyly is often associated with plexiform neurofibromas (Bendon and Giele 2013). The criteria for the diagnosis of NF-1 include: a first-degree relative with NF-1, café-au-lait spots, neurofibromas, axillary or inguinal freckling, optic gliomas, iris hamartomas (Lisch nodules), and bone lesions (scoliosis, kyphosis, pseudarthrosis of the tibia, and growth disorders).

34.5.3 Gigantism and Vascular Malformations

Capillary hemangiomas, cavernous hemangiomas, varicose veins, and arteriovenous fistulae may all be seen in association with local gigantism (Fig. 34.4a, b). The most well-known association is that of capillary hemangiomas with gigantism – the Klippel-Trenaunay syndrome (see Sect. 34.5.5).

34.5.4 Gigantism Associated with Multiple Enchondromatosis

Multiple enchondromatosis (Ollier's disease) is due to failure of ossification of cartilage in the diaphysis of long bones and bones of the hands and feet. Growth abnormalities, angular deformities, and localized gigantism of the upper or lower limbs often occur (Fig. 34.5).

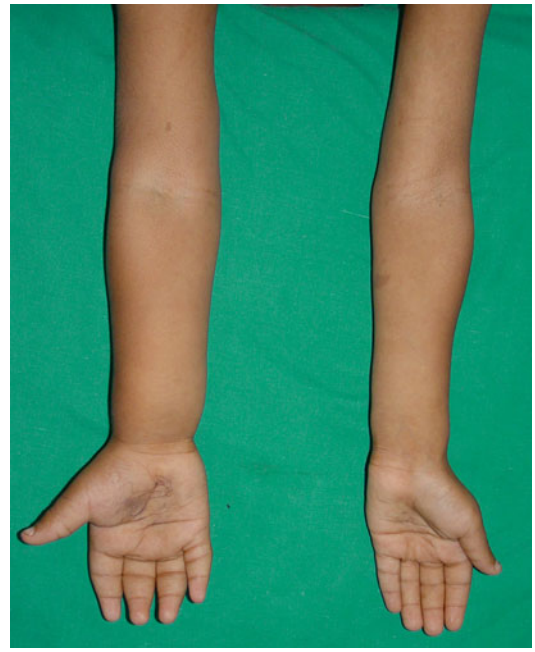

Fig. 34.5 Early features of overgrowth in the right upper limb in a child with Ollier's disease

34.5.5 Syndromes Associated with Local Gigantism

There are several overgrowth syndromes but not all are associated with localized overgrowth involving the extremities. Syndromes with overgrowth of the extremities have been summarized in Table 34.2.

34.5.6 Rarer Causes of Local Gigantism

Localized gigantism has been reported in association with tuberous sclerosis (Tung and Shih 2009) and dysplasia epiphysealis hemimelica (Stockley and Smith 1985).

34.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Table 34.3.

Table 34.2 Cardinal features of syndromes associated with localized gigantism involving the extremity, macrodactyly, or hemihypertrophy

Syndrome	Cardinal features	Comment
Beckwith-Wiedemann syndrome	Hemihypertrophy Macroglossia, exophthalmos Predisposition for embryonal malignancies like Wilms' tumor and hepatoblastoma	Increased rate of growth in latter half of pregnancy and first few years of life Hypoglycemia reported in 30–50 % of babies Visceromegaly frequent
Proteus syndrome (Cohen 2013)	Cerebriform connective tissue nevus Asymmetric disproportionate overgrowth of limbs, skull, viscera Propensity for tumors before second decade (ovarian cystadenoma/parotid adenoma) Lipomas or lipohypoplasia Vascular malformation (capillary/venous/lymphatic) Facial dysmorphism	Three essential criteria for diagnosis have been laid down (Biesecker 2006): The distribution of lesions is characteristically asymmetric and patchy The occurrence of syndrome is sporadic The course of the syndrome is progressive Deep vein thrombosis and pulmonary embolism can occur even in young children
Klippel-Trenaunay-Weber syndrome	Capillary hemangiomas Varicose veins and venous malformation Soft tissue and bone hypertrophy	67 % have all three of these features 98 % have capillary hemangiomas or a port-wine stain Often present at birth Often unilateral – stopping abruptly in the midline A rare but potentially life-threatening complication can be Kasabach-Merritt syndrome (a form of consumption coagulopathy that can lead on to disseminated intravascular coagulation)
Maffucci syndrome	Multiple enchondromatosis Multiple hemangiomas Potential for malignant change in enchondromas	Phleboliths occur in almost half the cases and deformities develop in two-thirds of cases Usually not evident at birth

Table 34.3 Establishing the diagnosis of cause of localized gigantism

History					
Overgrowth of affected part may be evident at birth	Overgrowth seldom evident at birth	Overgrowth of the affected part may be evident at birth	Overgrowth often evident at birth	Overgrowth seldom evident at birth	Overgrowth seldom evident at birth
Physical examination					
Macrodactyly Involvement of digits in median nerve distribution in hand or of medial plantar nerve distribution in foot	Macrodactyly or localized gigantism affecting a limb	Macrodactyly or localized gigantism affecting a limb	Hemihypertrophy	Localized gigantism affecting a limb	Macrodactyly or localized gigantism affecting a limb, skull, vertebrae
No café-au-lait spots No axillary freckling	Café-au-lait spots or axillary freckling present	No café-au-lait spots No axillary freckling	No café-au-lait spots No axillary freckling	No café-au-lait spots No axillary freckling	No café-au-lait spots No axillary freckling
No hemangiomas	No hemangiomas	Capillary hemangioma (port-wine stain) Cavernous hemangiomas	No hemangiomas	Cavernous hemangiomas may be present	Cavernous hemangiomas may be present
No varicose veins	No varicose veins	Varicose veins present	No varicose veins	No varicose veins	No varicose veins
No neurofibromas	Neurofibromas may be present (especially plexiform neurofibroma)	No neurofibromas	No neurofibromas	No neurofibromas	No neurofibromas
Bony deformity may be present in enlarged digit				Bony deformity frequently seen and may affect multiple bones	Bony deformity frequently develops over time and may affect multiple bones
No bony swellings	No bony swellings	No bony swellings	No bony swellings	Bony swellings in hand, feet, long bones	No bony swellings
No scoliosis No pseudarthrosis of tibia	Scoliosis/anterolateral bowing of tibia with or without pseudarthrosis	No scoliosis No pseudarthrosis of tibia	No scoliosis No pseudarthrosis of tibia	No scoliosis No pseudarthrosis of tibia	No scoliosis No pseudarthrosis of tibia
Working diagnosis: Macrodactyly with lipofibromatosis	Working diagnosis: Local gigantism with neurofibromatosis	Working diagnosis: Local gigantism in Klippel-Trenaunay syndrome	Working diagnosis: Hemihypertrophy	Working diagnosis: Local gigantism in Ollier's disease/Maffucci syndrome	Working diagnosis: Local gigantism in Proteus syndrome

(continued)

Table 34.3 (continued)

Investigations					
Plain radiographs: Will show proximal extent of overgrowth	Plain radiographs: Will show the extent of bony overgrowth and confirm tibial pseudarthrosis and scoliosis	Plain radiographs: Will show extent of bony overgrowth May demonstrate phleboliths in hemangiomas	Plain radiographs: Will show extent of bony overgrowth	Plain radiographs: Will show extent of bony overgrowth Will demonstrate enchondromas in affected bone May demonstrate phleboliths in hemangiomas	Plain radiographs: Will show extent of bony overgrowth
Diagnosis					
Macrodactyly in lipofibromatosis	Local gigantism in neurofibromatosis	Local gigantism in Klippel-Trenaunay syndrome	Hemihypertrophy or hemihypertrophy in Beckwith-Wiedemann syndrome	Local gigantism in Ollier's disease or in Maffucci syndrome	Local gigantism in Proteus syndrome

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Acquired Limb Length Inequality

35

Randall T. Loder

35.1 Introduction

Inequality in lower extremity length may be congenital or acquired. The congenital causes of limb length inequality have been covered in Chap. 15 and are not included in this chapter. The causes of acquired limb length inequality are numerous and the natural history varies; an accurate diagnosis is essential to planning proper treatment.

Before proceeding to investigate the cause of shortening, it needs to be confirmed that the shortening is true and not apparent. Deformities of the hip or knee are responsible for apparent limb length inequality. A hip adduction contracture will give an appearance of a shorter limb on the affected side; likewise, a hip abduction contracture will give an appearance of a shorter limb on the opposite side (see Fig. 32.4). Hip and knee flexion contractures can also give the appearance of a shorter limb even if the actual bony length is equal to that of the opposite side. Management of apparent shortening involves identifying the pathology at the affected joint and correcting the joint deformity.

True limb length inequality occurs when one lower limb is abnormally short or abnormally long. The causes of abnormal lengthening due to overgrowth syndromes have been discussed in Chap. 34 and are not included in this chapter; the ensuing discussion is limited to the causes of true shortening of a limb.

35.2 Questions to Establish a Diagnosis

- **Is the discrepancy in limb lengths increasing?**
- **Was the onset abrupt or gradual?**
- **Is there a history of prior trauma or infection?**
- **Is there a history of neurologic disease?**
- **Where is the shortening?**
- **Is there an associated angular deformity in the short limb?**
- **Is there associated atrophy of the muscle groups?**
- **Is there an antalgic or Trendelenburg gait in addition to the short-limb gait?**

Is the discrepancy in limb lengths increasing?

Progressive increase in the limb length discrepancy suggests that the growth of the shorter limb has been affected and this implies that one or more growth plates are damaged. Being aware that the discrepancy is progressive and recognition of the growth plate damage is of vital importance for treatment planning.

Was the onset abrupt or gradual?

An abrupt onset cannot be attributed to growth impairment but is likely to be due to a sudden structural change such as paralytic dislocation

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of the hip, progression of a SCFE, or collapse of the epiphysis in Perthes disease.

Is there a history of prior trauma or infection?

Physeal injury due to trauma or infection or malunion of a fracture with overriding of the fragments can result in limb shortening.

Is there a history of neurologic disease?

Children with cerebral palsy, myelomeningocele, poliomyelitis, and other neurologic disorders can have progressive limb shortening.

Where is the shortening?

Does it involve all portions (e.g., femur, tibia, and foot), or is it localized? Isolated limb segment shortening is typically seen in posttraumatic or infectious conditions. Isolated femoral shortening can also point toward hip dislocation.

Is there an associated angular deformity in the short limb?

Angular deformities at the knee or ankle may be seen following physeal arrests (e.g., traumatic or postinfective physeal damage, Blount's disease); there may be a history of recurrence of the deformity following surgical correction. Joint deformities may be seen in paralytic conditions. Deformity in the diaphysis of the femur or tibia may be seen following malunion of a fracture.

Is there associated atrophy of the muscle groups?

Significant muscle atrophy is seen in lower motor neuron paralysis (e.g., polio, spina bifida); muscle wasting is less obvious in upper motor neuron paralysis (e.g., cerebral palsy). Muscle atrophy may also be seen in children with painful joint disease (e.g., tuberculosis of the hip, Perthes disease).

Is there an antalgic or Trendelenburg gait in addition to the short-limb gait?

All children with shortening will have a short-limb gait with or without compensatory mechanisms for the shortening (see Table 24.2). In addition, there may be an antalgic component or a Trendelenburg gait. An antalgic gait may be seen in Perthes disease and SCFE. A Trendelenburg gait will be present if the hip is dislocated, if the femoral head is destroyed, if there is coxa vara, or if there is abductor weakness (see Table 24.2).

35.3 Physical Examination

35.3.1 Look

Observe the limb and answer these questions: Where is the shortening and what is the magnitude of shortening? Are there associated angular deformities either at the joints or in the thigh or leg? Is there muscle atrophy?

Observe the gait of the child and answer these questions: Is the limp purely a short-limb gait?

Does the short-limb gait have an antalgic component? In addition to the short-limb gait, is there a Trendelenburg gait pattern?

35.3.2 Feel

If there is a deformity, palpate the limb to localize the site of deformity; identify whether there is a contracture of the muscles or scarring of the overlying skin (e.g., as in melorheostosis).

35.3.3 Move

Test the passive ranges of movements of the hip, knee, and ankle. Determine if there are any contractures at these joints

35.3.4 Special Tests

Estimating the Limb Length Discrepancy

The total limb length inequality is determined by the block method which is regarded as the most reliable clinical estimate of leg length discrepancy (Woerman and Binder-Macleod 1984). Tape measurements from the anterior superior iliac spine to the medial malleolus are notoriously inaccurate (Stanitski 1999).

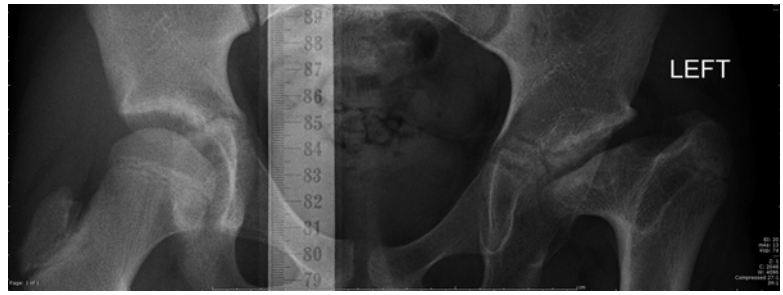

Fig. 35.1 Radiograph demonstrating a moderate leg length discrepancy due to septic arthritis of the hip at 8 months of age

35.4 Investigations to Confirm the Diagnosis

35.4.1 Plain Radiographs

If the child can stand, a standing AP alignment film from the pelvis to the ankles is first obtained ensuring that both patellae are facing forward. This allows an evaluation of the mechanical axis; a line drawn from the center of the femoral head to the center of the tibial ankle plafond should fall through the middle of the knee. It also allows an overall assessment of limb length if rulers are placed on the film. If the child cannot stand, then the study can be performed supine.

If there appears to be pathology in the hip, then AP and frog lateral pelvis radiographs are obtained; if in the femur, then bilateral AP and lateral femoral radiographs are needed.

If there is significant limb shortening, a scanogram is obtained. It provides an accurate measurement of femoral and tibial length as long as there are no joint contractures.

An AP radiograph of the hand and wrist is obtained to estimate the skeletal age of the child in order to plan treatment.

35.4.2 CT Scanogram

If joint contractures are present, then a CT scanogram should be obtained, since the actual bony length can be measured regardless of contracture with the CT method (Stanitski 1999).

35.4.3 Additional Imaging

Further imaging is performed to more accurately define the underlying pathologic process if a lesion that is causing the shortening is identified.

If there is a concern for a physeal arrest, then either CT or MRI scans are performed to determine the anatomic location of the physeal bar as well as its overall cross-sectional area of the respective physis (Khoshhal and Kiefer 2005).

If there is a concern that the underlying pathology is neoplastic, then CT and MRI scans are performed as indicated based upon the initial plain radiographic appearance.

35.5 Differential Diagnosis

35.5.1 Shortening Secondary to Physeal Arrest from Septic Processes

Destruction of the physis from septic arthritis and osteomyelitis is not infrequent. The shortening and deformities that ensue are progressive. The younger the child at the time of the infective episode, the more severe the leg length discrepancy will be at maturity. Several factors that predispose to septic physeal arrest and growth deformity have been identified (Frank et al. 2005; Samora and Klingele 2013). Septic arthritis of the hip and acute osteomyelitis of the proximal femur in the young child can lead to rapid destruction of the hip joint, resulting in physeal arrest and hip deformity (Fig. 35.1) to more

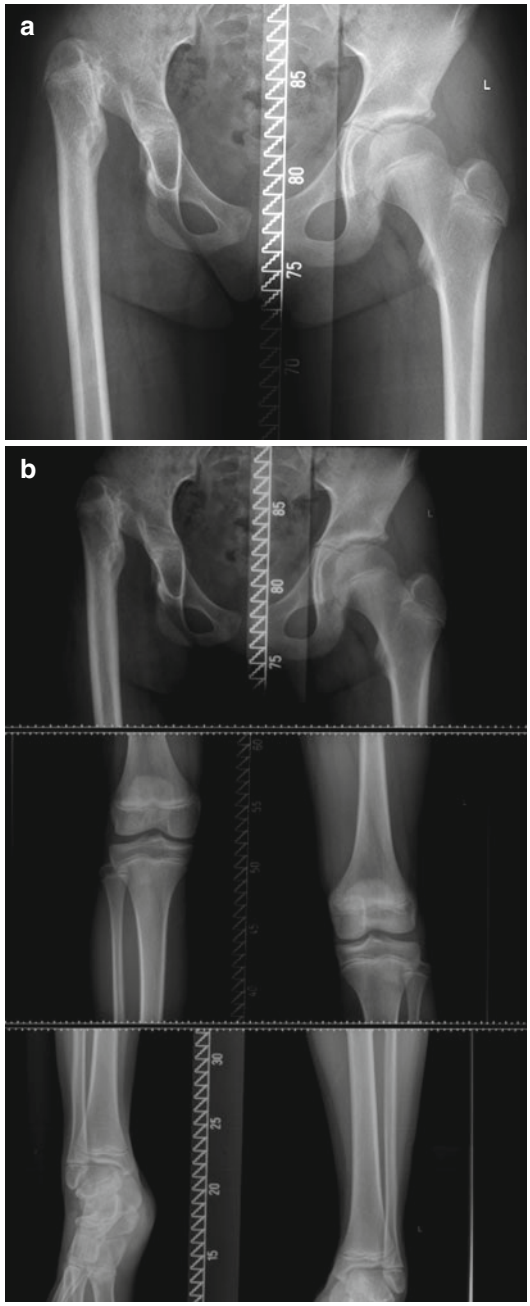

Fig. 35.2 Limb length discrepancy from septic arthritis or osteomyelitis of the hip in infancy. Radiographs demonstrating complete destruction of the femoral head and neck with secondary dislocation from untreated infection in infancy. The AP pelvis radiograph (**a**) demonstrates the proximal dislocation along with the destruction of the proximal femoral epiphysis and metaphysis. The scanogram (**b**) demonstrates the leg length discrepancy

aggressive destruction of the entire proximal femur with secondary hip dislocation (Fig. 35.2). Purpura fulminans, either due to meningococcemia or other pyogenic bacteria, often leads to multiple physeal arrests of the lower extremities which results in leg length discrepancies and angular deformities (Belthur et al. 2005; Canavese et al. 2010; Park and Bradish 2011). The radiographic hallmark is central tenting of the physis.

35.5.2 Shortening Due to Trauma

Shortening of the limb following trauma can either be due to physeal injury or malunion from the fracture healing with overriding of the fragments. Shortening due to a malunion occurs most often in the femur and is typically noted immediately after fracture union.

Shortening resulting from a physeal injury may be recognized only after a few years. Shortening is usually due to direct injury to the physis associated with an epiphyseal fracture (Fig. 35.3) but can rarely result just from significant soft tissue injury in a limb having sustained major trauma (Bowler et al. 1990; Hresko and Kasser 1989). When the peripheral part of the physis is injured, not only limb length discrepancy but angular deformities also develop. The distal femoral (Arkader et al. 2007; Eid and Hafez 2002; Kasser 1990) and distal tibial physes (Blackburn et al. 2012) are extremely prone to this problem. Fractures involving the proximal tibial physis are less common; thus, fewer deformities are seen from such fractures (Pappas et al. 1984; Vrettakos et al. 2012). Paraphyseal fractures involving the femoral neck may result in a deformity; however, those are typically due to malunion (e.g., posttraumatic coxa vara) or avascular events with femoral head collapse and limb shortening.

35.5.3 Shortening from Acquired Lower Extremity Disorders

Developmental Hip Dysplasia

In the child with a late diagnosis of DDH, there will be shortening due to proximal migration of the dislocated hip. In a child with treated DDH,

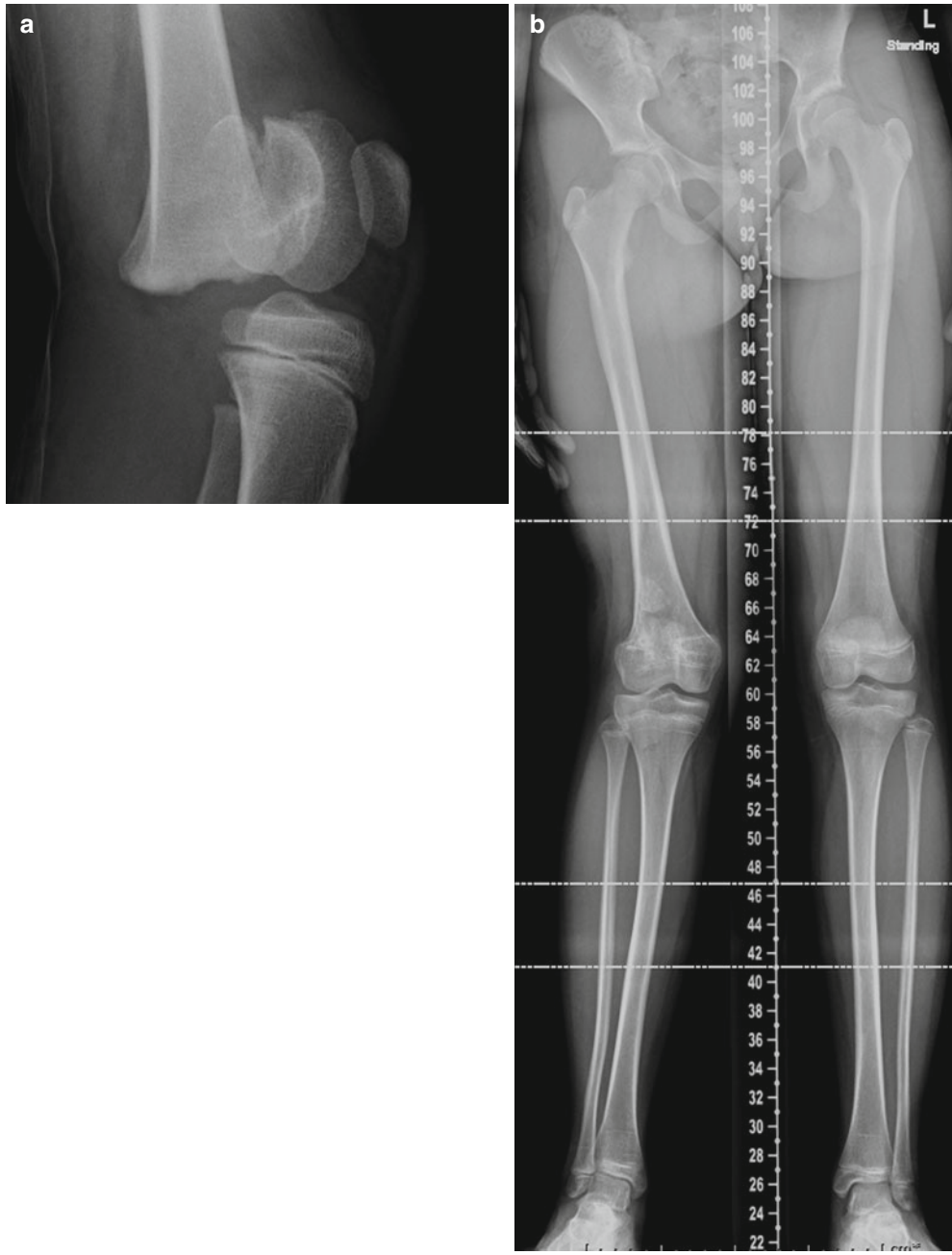

Fig. 35.3 Leg length discrepancy due to physeal trauma with growth arrest. (**a**) A markedly displaced distal femoral Salter I epiphyseal fracture at age 9 years. (**b**) At age 12 years, there has been nearly complete physeal arrest resulting in significant leg length discrepancy and slight valgus deformity

shortening may occur from the treatment itself (e.g., femoral osteotomy) or avascular necrosis with coxa vara. The history and radiographs will lead to the correct diagnosis.

Slipped Capital Femoral Epiphysis (SCFE)

Children with SCFE can develop shortening due to two different causes: the actual slip itself and

sequelae of treatment. The SCFE itself, with increasing inferior epiphyseal slip, results in proximal migration of the femoral metaphysis and subsequent shortening (Kim et al. 2013). After treatment with screw fixation, especially in the younger patient with a SCFE (Segal et al. 1991), the premature physeal closure contributes an additional cause to limb shortening.

Legg-Calvé-Perthes Disease

These children can develop shortening due to both the epiphyseal collapse, as well as the coxa vara that often results after healing of the epiphyseal necrosis (Grzegorzewski et al. 2005). Shortening can also occur due to premature closure of the capital femoral physis.

Tibia Vara

Children with tibia vara (Blount's disease), especially the infantile type, readily develop shortening along with a varus deformity (Birch 2013). This is due to significant growth retardation from the involved proximal medial tibial physis.

35.5.4 Shortening Secondary to Neurologic Conditions

Children with hemiplegic cerebral palsy (Riad et al. 2010), poliomyelitis (Emara and Khames 2008), and other central nervous system and/or spinal cord problems (transverse myelitis) can easily develop a limb length difference due to the asymmetry of the neurologic insult/involvement (Gourineni et al. 2009).

35.5.5 Neoplastic and Dysplastic Conditions

Children with many benign neoplastic processes, including enchondromatosis (Shapiro 1982), multiple hereditary exostoses (Clement et al. 2012; Schmale et al. 1994), focal fibrocartilaginous dysplasia, melorheostosis (Campbell et al. 1968; Younge et al. 1979), dysplasia epiphysealis hemimelica (Trevor's disease) (Smith et al. 2007), and bone cysts (Glowacki et al. 2011) (both simple and aneurysmal) can develop a shortened lower extremity (Figs. 35.4 and 35.5).

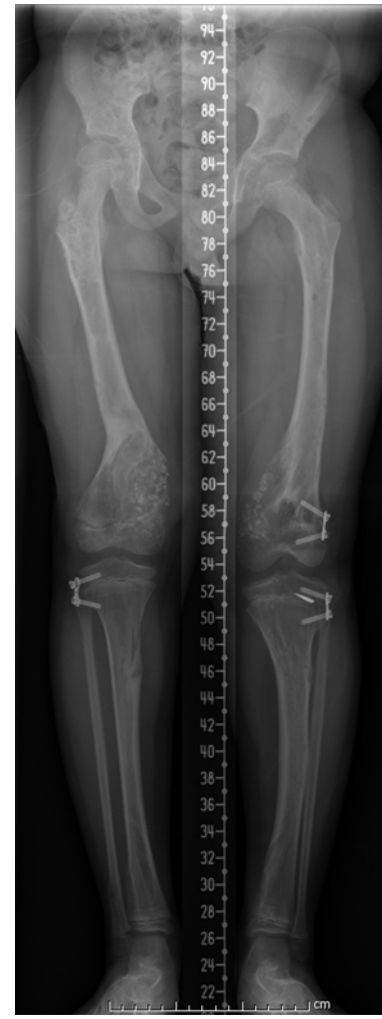

Fig. 35.4 Leg length discrepancy associated with Ollier's disease in an 8-year-old boy

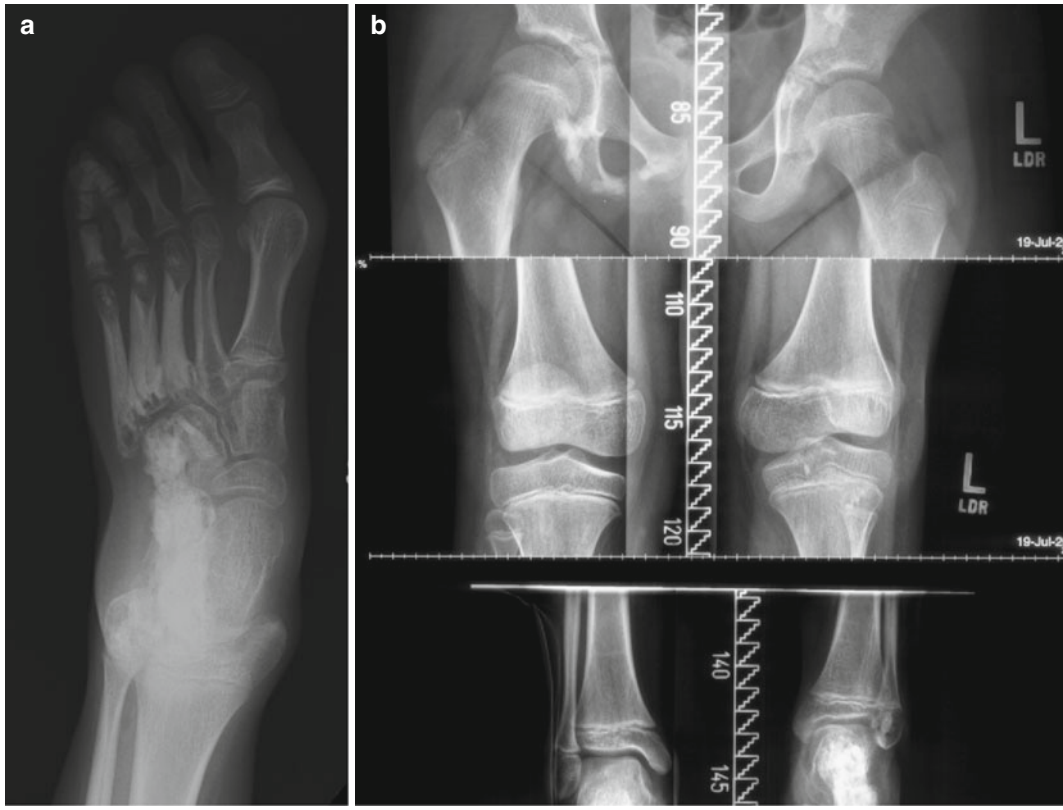

Fig. 35.5 Leg length discrepancy associated with melorheostosis in an 8-year-old girl. (**a**) Note the classical findings of candle wax drippings on the bones. (**b**) Scanogram demonstrating leg length discrepancy, being shorter on the side with melorheostosis (*left side*)

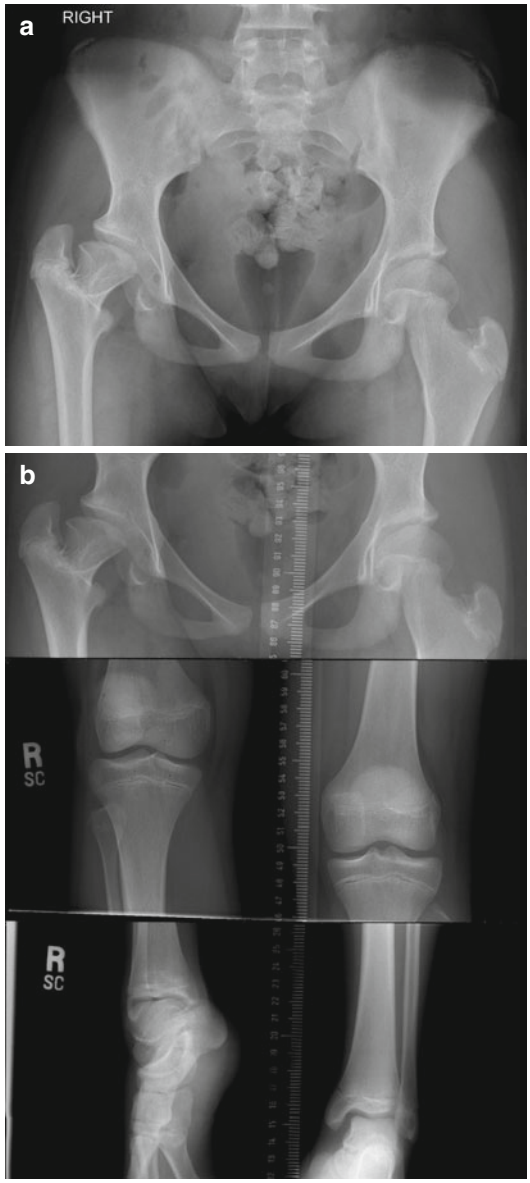

Fig. 35.6 Leg length discrepancy in a child with multiple femoral arterial cannulations in infancy. (**a**) The AP pelvis radiograph at age 13 years demonstrates complete arrest of the right proximal femoral epiphysis resulting in coxa vara with a small femoral head and a reduced articulotrochanteric distance. The left proximal femur demonstrates lateral physeal tethering with a subsequent coxa valga deformity. (**b**) The scanogram demonstrates a marked leg length discrepancy, the right shorter than the left

35.5.6 Miscellaneous Causes of Shortening

Arterial cannulation in the newborn or infant can result in lower extremity shortening (Macnicol and Anagnostopoulos 2000) (Fig. 35.6). Extravasation of intravenous fluids in neonates can also produce a growth arrest (Sanpera et al. 1994).

35.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Tables 35.1 and 35.2.

Table 35.1 An outline of diagnosis of shortening of one lower limb *with a history of trauma or infection*

History				
History of trauma present	History of trauma present	History of infection present	History of infection present	History of infection present
Shortening not progressive (may be some reduction of the discrepancy)	Progressive shortening	Progressive shortening	Progressive shortening	Progressive shortening
Shortening noted soon after fracture union	Shortening noted late	Shortening noted late	Shortening noted late	Shortening noted late
The fracture did not involve the physis	The fracture did involve the physis	The infection involved the metaphysis	The infection involved the metaphysis	The infection involved the metaphysis
Physical examination				
One segment of the lower extremity is short (+) Galeazzi sign indicates femoral shortening Knees at same level with medial malleolus at different levels indicates tibial shortening	One segment of the lower extremity is short (+) Galeazzi sign indicates femoral shortening Knees at same level with medial malleolus at different levels indicates tibial shortening	One segment of the lower extremity is short (+) Galeazzi sign indicates femoral shortening Knees at same level with medial malleolus at different levels indicates tibial shortening	Multiple segments of the lower extremity are short	One segment of the lower extremity is short (+) Galeazzi sign No tibial discrepancy
Angular deformity may or may not be present	Angular deformity often present	Angular deformity often present	Angular deformity often present	May have a mild angular deformity
Hip rotation may be asymmetric when the femur is the short segment	Flexion or extension deformities of the knee or ankle may be present	Flexion or extension deformities of the knee or ankle may be present	Flexion or extension deformities of the knee or ankle may be present	May have decreased abduction of the hip
(±) Trendelenburg gait	(±) Trendelenburg gait	(−) Trendelenburg gait	(±) Trendelenburg gait	(+++) Trendelenburg gait

(continued)

Table 35.1 (continued)

Investigations				
Standing long leg alignment film will show Shortened bone that is involved May show angular deformity (valgus/varus) of involved bone	Standing long leg alignment film will show Shortened bone that is involved Often shows an angular deformity (valgus/varus) of involved bone Radiographs of the involved joint often suspicious for a physeal bar	Standing long leg alignment film will show Shortened bone that is involved Often shows an angular deformity (valgus/varus) of involved bone Radiographs of the involved joint often suspicious for a physeal bar No joint dislocation	Standing long leg alignment film will show Shortened bone that is involved Often shows an angular deformity (valgus/varus) of involved bone Radiographs of the involved joint often suspicious for a physeal bar, often in the center of the physis and tented	Standing long leg alignment film will show Hip joint is dislocated Often the entire femoral head is absent
MRI/CT scans not needed to make the diagnosis	MRI/CT scans: Will demonstrate a physeal bar	MRI/CT scans: Will demonstrate the physeal bar	MRI/CT scans: Will demonstrate the physeal bar	MRI/CT scans not needed to make the diagnosis
Diagnosis				
Femoral or tibial fracture malunion	Posttraumatic physeal arrest	Physeal arrest due to osteomyelitis and/or septic arthritis	Physeal arrest due to purpura fulminans	Septic hip joint dislocation due to metaphyseal osteomyelitis and septic arthritis of the hip

Table 35.2 An outline of diagnosis of shortening of one lower limb *with a history of developmental and neurologic conditions*

History					
Noted in infancy or early childhood	Noted in mid childhood	Noted in late childhood/ adolescence	Noted in childhood and/ or adolescence	Noted in childhood and/ or adolescence	Noted in childhood and/ or adolescence
No history of significant childhood viral illness with subsequent muscle weakness	No history of significant childhood viral illness with subsequent muscle weakness	No history of significant childhood viral illness with subsequent muscle weakness	No history of significant childhood viral illness with subsequent muscle weakness	No history of significant childhood viral illness with subsequent muscle weakness	History of significant childhood viral illness with subsequent muscle weakness
No history of prenatal/ perinatal difficulty	No history of prenatal/ perinatal difficulty	No history of prenatal/perinatal difficulty	No history of prenatal/ perinatal difficulty	May have a history of prenatal/perinatal difficulty	No history of prenatal/ perinatal difficulty
Physical examination					
One segment of the lower extremity is short (+) Galeazzi sign No tibial discrepancy	One segment of the lower extremity is short (+) Galeazzi sign No tibial discrepancy	One segment of the lower extremity is short (+) Galeazzi sign No tibial discrepancy	One segment of the lower extremity is short (−) Galeazzi sign Tibial shortening seen	Both segments of the lower extremity are short	Both segments of the lower extremity are short Tibia often more involved than the femur
Decreased hip abduction Increased hip internal rotation Flexion contracture ±	Decreased hip abduction, and internal rotation Hip flexion contracture +	Varying degrees of decreased hip abduction, internal rotation, and extension	Knee exam demonstrates varus deformity with lateral ligamentous laxity	Varying degrees of decreased hip, knee, and ankle motion	Often with increased passive motion
(+) Trendelenburg gait	(±) Trendelenburg gait	(±) Trendelenburg gait	(−) Trendelenburg gait	Often with mild hip/ knee/ankle flexion contractures	With or without hip/ knee/ankle contractures
Aside from the Trendelenburg findings, general lower extremity motor strength normal	Aside from the Trendelenburg findings, general lower extremity motor strength normal	Aside from the Trendelenburg findings, general lower extremity motor strength normal	Aside from the Trendelenburg findings, general lower extremity motor strength normal	Varying degrees of spastic muscle weakness	Varying degrees of flaccid muscle weakness

(continued)

Table 35.2 (continued)

Investigations					
Standing long leg alignment film will show Developmental hip dislocation AP and frog pelvis radiographs will demonstrate a hip dislocation with varying degrees of acetabular dysplasia	Standing long leg alignment film will show Varying degrees of femoral shortening and coxa vara AP and frog pelvis radiographs will show Perthes disease	Standing long leg alignment film will show Mild leg length discrepancy AP and frog pelvis radiographs will show a SCFE	Standing long leg alignment film will show Shortened bone that is involved Often shows an angular deformity (valgus/varus) of involved bone Radiographs of the involved joint often suspicious for a physeal bar, often in the center of the physis and tented	Standing long leg alignment film will show Both femoral and tibial shortening May show an angular deformity (valgus/varus) of involved bones and joints Lateral radiographs of the knee may show patella alta	Standing long leg alignment film will show Both femoral and tibial shortening Tibial shortening often greater than femoral May show an angular deformity (valgus/varus) of involved bones and joints
MRI/CT scans not needed to make the diagnosis	MRI/CT scans not needed to make the diagnosis	MRI/CT scans not needed to make the diagnosis	MRI/CT scans not needed to make the diagnosis May demonstrate a proximal tibial physeal bar	MRI/CT scans not needed to make the diagnosis	MRI/CT scans not needed to make the diagnosis
Diagnosis					
Developmental hip dislocation	Legg-Calvé-Perthes disease	Slipped capital femoral epiphysis	Infantile tibia vara (Blount's disease)	Leg length discrepancy due to hemiplegic cerebral palsy	Leg length discrepancy due to residuals of poliomyelitis

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Part V

The Adolescent

The Painful Hip in Adolescence

36

Randall T. Loder

36.1 Introduction

Hip pain in an adolescent is a common complaint, with a myriad of possible diagnoses, and can range from straightforward conditions with a good prognosis to severe conditions with a poor prognosis, either for the future of the hip or the life of the patient. Many of the conditions overlap with the school-age child. This chapter will address only those not discussed in that chapter.

36.2 Questions to Establish a Diagnosis

- **Was trauma involved?**
- **Was the onset gradual or abrupt?**
- **Can the child still walk or not?**
- **Are there associated constitutional symptoms (fever, chills, weight loss)?**
- **Does the pain occur at night or all the time?**
- **Are the symptoms unilateral or bilateral?**
- **Is the pain relieved by NSAIDs?**
- **Are there any other associated medical conditions?**

Was trauma involved?

If so, then this typically narrows down the diagnoses to those of a fracture (either normal or pathologic bone) or possible slipped capital femoral epiphysis (SCFE).

Was the onset gradual or abrupt?

If abrupt, fractures, infections, and unstable SCFE are more likely. If gradual, then inflammatory conditions, neoplasms, stable SCFE, acetabular dysplasia, and femoroacetabular impingement are more likely.

Can the child walk or not?

The ability to ambulate is lost with fractures and unstable SCFE and often with severe infections.

Are there associated constitutional symptoms?

Fevers and chills are frequently associated with infections, and weight loss with malignant neoplasms.

Does the pain occur at night?

The classic history for an osteoid osteoma is night pain. Pain associated with SCFE, dysplasias, femoroacetabular impingement, and stress fracture is frequently relieved by rest and aggravated by activity.

Are the symptoms unilateral or bilateral?

Bilateral symptoms are quite rare with fractures, infections, and neoplasms. Patients with SCFE, AVN, and dysplasias may have either unilateral or bilateral symptoms/involvement.

Is the pain relieved by NSAIDs?

NSAIDs often markedly relieve pain of osteoid osteomas and to some extent pain associated with other conditions (e.g., AVN, dysplasias).

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_36

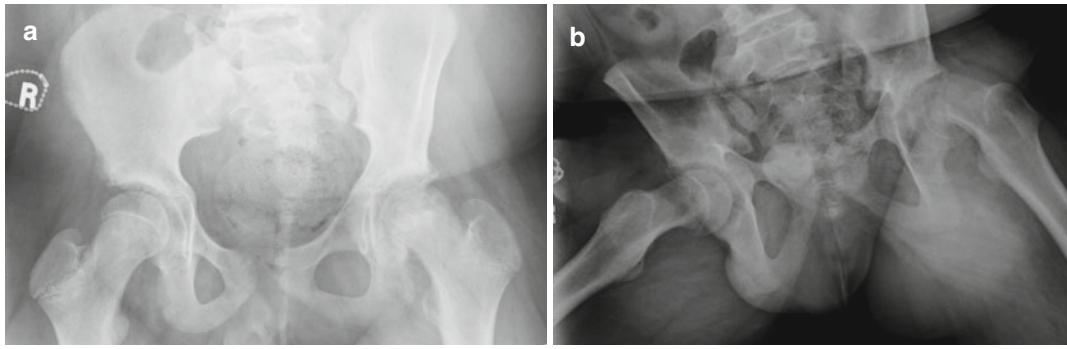

Fig. 36.1 AP (**a**) and frog-lateral radiograph (**b**) of a typical patient with a stable SCFE, presenting with several months of intermittent hip/groin/thigh knee pain; note the mild SCFE on the right and the severe SCFE on the left

Are there any other associated medical conditions?

Children with the various epiphyseal and/or skeletal dysplasias frequently have hip involvement. Those with hemoglobinopathies also can have hip involvement. Those who have been treated for a previous malignancy or other conditions with high-dose steroids may have AVN.

36.3 Physical Examination

This is essentially the same approach as in Chap. 32.

36.3.1 Look

Does the patient appear ill or cachectic, pointing toward infection or malignancy?

36.3.2 Feel

Is there point tenderness, and if so, where? Tenderness over the iliac spines and ischial tuberosity points to avulsion injuries. Point tenderness over the greater trochanter may indicate a greater trochanteric bursitis. Is there warmth over the pelvis or hip? This will point toward an inflammatory process (e.g., infection).

36.3.3 Move

The most important component of the physical examination is the range of motion of the hip and pattern of gait. The same tests are used as discussed in Chap. 32; see that chapter for details. The questions we ask in this age group are: If the child can walk, is the gait antalgic? Is there a Trendelenburg component indicating hip abductor weakness? Is there pain with provocative tests, e.g., flexion/adduction/internal rotation for a labral tear or femoroacetabular cam impingement?

36.4 Investigations to Confirm the Diagnosis

Plain Radiography

This is the first step and an AP and frog leg pelvis radiograph should be obtained. This is extremely important as early hip diseases, especially SCFE, can be missed on a single AP view (Figs. 36.1 and 36.2).

Laboratory Studies

Appropriate laboratory studies are also obtained where indicated (e.g., CBC with differential, ESR, CRP if concerned about sepsis) and hemoglobin electrophoresis/sickle cell prep if concerned about a hemoglobinopathy.

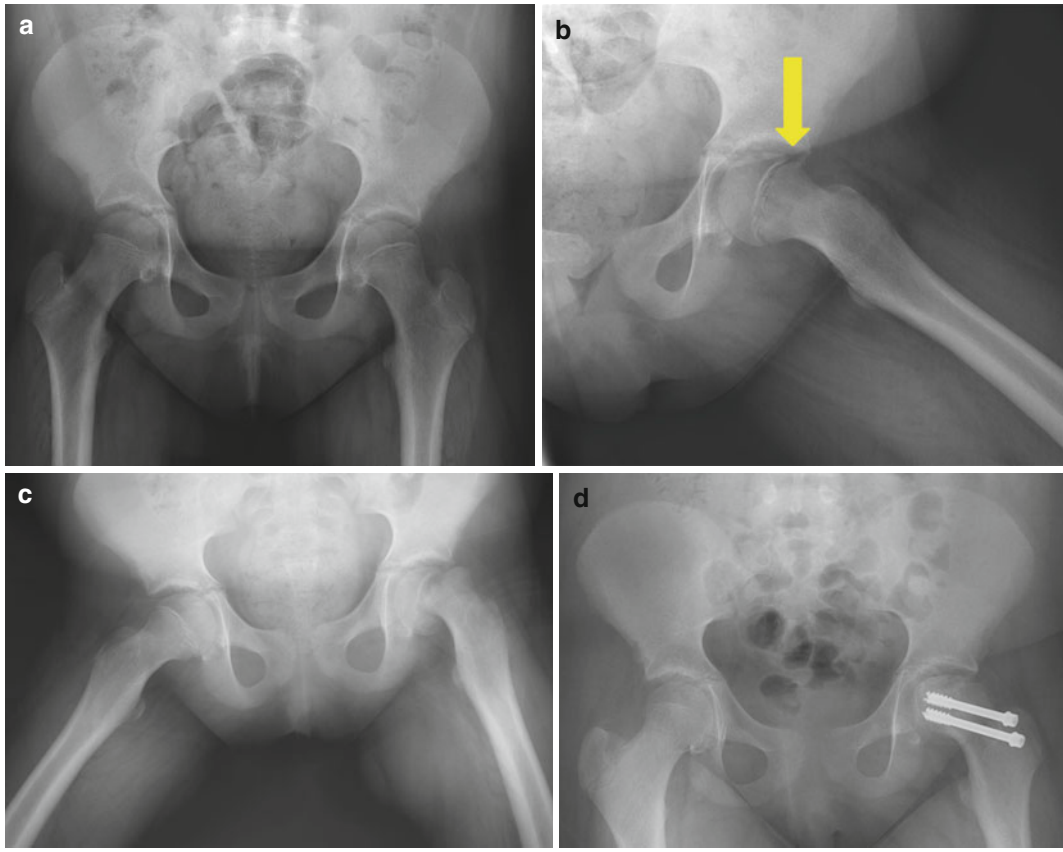

Fig. 36.2 The need to diagnose SCFE is evident in this series of radiographs. An AP pelvis (**a**) and lateral left hip (**b**) radiograph in a 12-year-old girl with thigh and knee pain. Note the very subtle left SCFE (*arrow*). This was not observed. A month later the patient fell resulting in an abrupt, unstable SCFE (**c**). In spite of urgent treatment, the patient went on to develop avascular necrosis, a horrific complication (**d**)

Other Imaging

Special imaging studies such as a bone scan and/or MRI are needed if the above are negative and there is a concern for an early osteomyelitis/pyomyositis, osteoid osteoma, or stress fracture. If there is a concern about femoroacetabular impingement and labral tear, an MR gadolinium injection arthrogram is often necessary.

36.5 Differential Diagnosis

36.5.1 Slipped Capital Femoral Epiphysis

This is a common cause of pain in the hip, groin, thigh, or knee in the adolescent, especially those who are obese (Loder et al. 2000, 2008; Loder and Skopelja 2011). The typical history for the stable SCFE is that of several weeks to months of intermittent pain which comes and goes. In the unstable SCFE, the same prodromal symptoms are often seen, after which an abrupt event occurs that disallows the child from being able to walk, with or without crutches (Loder et al. 1993). Physical examination in the stable SCFE documents varying degrees of loss of internal rotation of the hip, leg length discrepancy, and obligatory external rotation of the hip with increasing flexion. The diagnosis is easily confirmed with plain AP and lateral radiographs (Fig. 36.1); both hips must be imaged with both views. However, the slip can be very subtle in the early stages and can be missed, which can lead to disastrous results if the SCFE becomes unstable (Fig. 36.2).

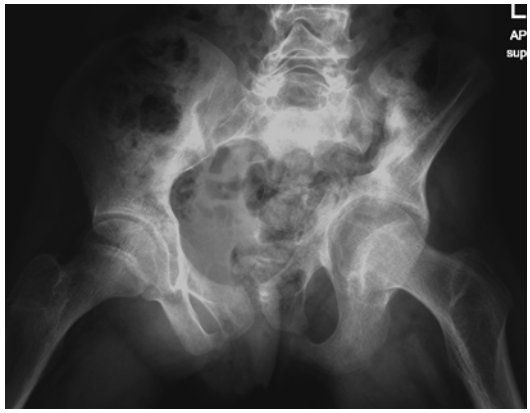

Fig. 36.3 Chondrolysis of the left hip in a 12-year-old syndromic girl. Note both the decreased joint space compared to the right hip as well as the marked acetabular protrusion

36.5.2 Avascular Necrosis

The causes of avascular necrosis of the femoral head in the teenager include trauma, SCFE, steroid use, and adolescent Perthes disease. The course of the Perthes disease in the adolescent is atypical, and the prognosis is poor once collapse of the epiphysis occurs; early diagnosis is essential. The adolescent with idiopathic AVN typically presents with vague groin and thigh pain without any preceding event (e.g., trauma). Weight bearing will be painful. Physical examination will demonstrate a mild loss of internal rotation in the early stages and later even loss of abduction and extension. Plain radiographs are often normal, and MRI is the easiest way to establish the diagnosis.

36.5.3 Idiopathic Chondrolysis

This is extremely rare but may also be associated with SCFE and acetabular protrusion (Sherlock 1995). It may also be confused with pauciarticular juvenile arthritis (van der Hoeven et al. 1989). It is often seen in girls (Daluga and Millar 1989) or those of African descent (Roy and Crawford 1988). The hallmark presentation is increasing hip/groin/thigh pain with no precipitating event. Physical examination demonstrates significant loss of hip motion, especially internal rotation, abduction, and extension. The gait is often antalgic and in a flexion posture (Rowe and Ho 1996). Plain radiographs demonstrate decreased joint space compared to the opposite side (Fig. 36.3). As such, the concern for a subtle inflammatory process and/or infection is raised. However, laboratory studies are nearly always normal. Bone scans may demonstrate increased uptake; MRI is the most sensitive study after plain radiographs and will demonstrate decreased thickness of the articular cartilage with no effusion (Johnson et al. 2003; Laor and Crawford 2009).

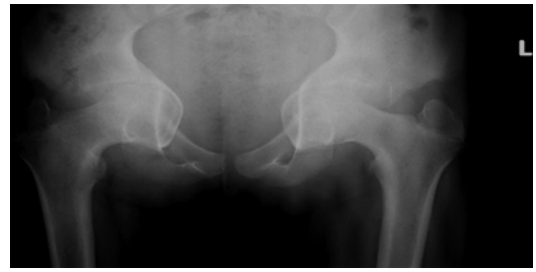

Fig. 36.4 Bilateral acetabular protrusio in a 15-year-old syndromic girl. The diagnosis is confirmed by the center-edge angle of Wiberg > 50°

36.5.4 Acetabular Protrusio

This is a relatively rare condition. It can be idiopathic or associated with other syndromes (Bennett and McMurray 1990; Gusis et al. 1993; Sponseller et al. 2006) (Fig. 36.4). The patients present with hip/groin pain and demonstrate loss of hip motion in most planes. It can often be associated with chondrolysis, and the differentiation between idiopathic chondrolysis and acetabular protrusion can be difficult. The gait is often in a flexed position. Certain authors consider both conditions to be different spectrums of the same pathologic process (Sherlock 1995).

36.5.5 Femoroacetabular Impingement (FAI)

FAI is a recently described phenomenon and is characterized by abnormal mechanical contact between the acetabular rim and the proximal femur (Sankar et al. 2013). In adolescent patients it is

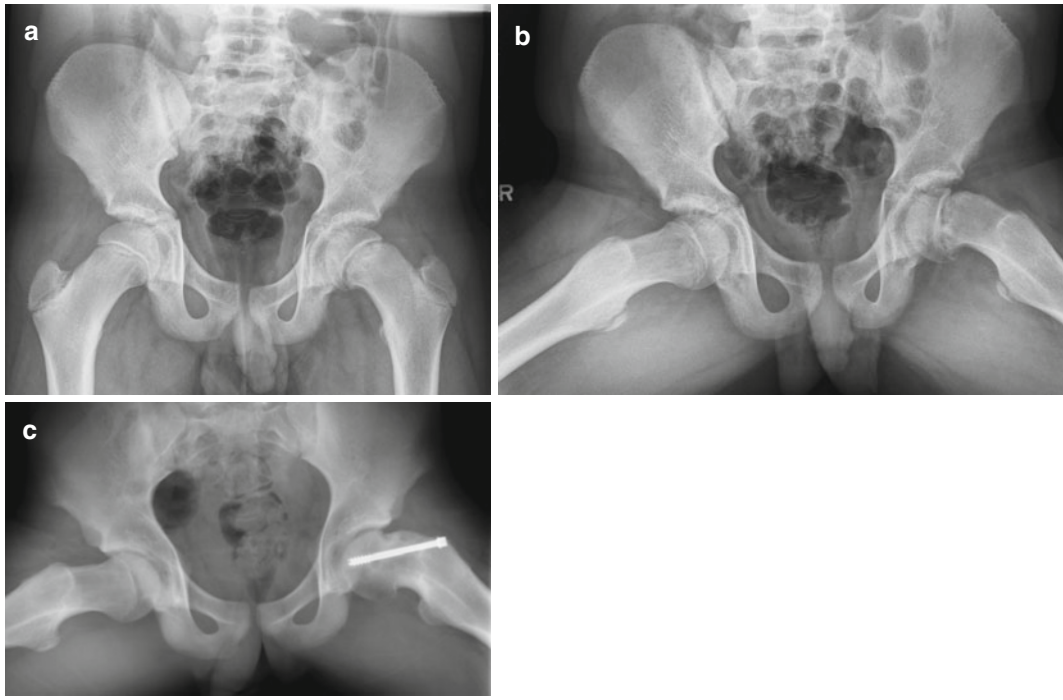

Fig. 36.5 Femoroacetabular impingement secondary to SCFE. The initial presenting radiographs demonstrating a mild SCFE on the AP (**a**) and frog-lateral (**b**) radiographs. Three years after single screw fixation, there is persistent impingement of the anterior metaphyseal bump on the lateral acetabular edge (**c**), causing pain

usually secondary to SCFE (Millis and Novais 2011; Sankar et al. 2013) (Fig. 36.5) or Perthes disease. There are two major types of FAI: pincer and cam impingement. Pincer impingement is due to a very deep acetabulum (even acetabular protrusio) which pinches the femoral metaphysis in certain positions. Cam impingement is due to the proximal femoral metaphysis abutting upon the acetabular rim and labral structures, usually at the extremes of flexion and internal rotation (Leunig et al. 2000; Rab 1999). A positive impingement test occurs when there is groin pain with passive flexion, internal rotation, and adduction of the hip (Sink and Kim 2012). The diagnosis can be further suggested by plain radiographs. MRI scans will demonstrate an increased α angle along with varying degrees of labral tears/detachment and even loss of femoral head articular cartilage (Bedi and Kelly 2013; Frank et al. 2013; Leunig et al. 2000; Millis and Novais 2011; Sankar et al. 2013). The diagnosis and results of treatment are controversial (Rubin 2013).

36.5.6 Developmental Hip Dysplasia (DDH)

DDH can present late in childhood but have different demographics of typical infantile/childhood hip dysplasia (Lee et al. 2013). These children are those without classic hip dislocation, but rather represent those with mild acetabular dysplasia/subluxation that had no findings or complaints on childhood examinations, but later in the teen years began to complain of hip/groin pain. The demographics of patients with late presentation of DDH are different than those with the typical infantile/childhood DDH. The patients will present with groin/hip pain and fatigue after activities (Fig. 36.6).

36.5.7 Musculoskeletal Infection

Septic arthritis and osteomyelitis of the hip and proximal femur also occur in the adolescent, similar to the school-age child. The workup is the same.

One particularly unique process in the teenager is septic involvement of the sacroiliac joint, typically associated with iliac osteomyelitis (Figs. 36.7 and 36.8). It can often be difficult to diagnose (Ford et al. 2004). These patients have multiple different presenting complaints, which can be low back pain, pelvic pain, and thigh/groin pain (Wada et al. 2008). As the process evolves, the patient will lose the ability to bear weight and then eventually lose the ability to walk. They may have associated abdominal pain. The patient will also become febrile and toxic in appearance. Physical examination will demonstrate tenderness to palpation over the sacroiliac joint as well as pain/limitation with the flexion/abduction/external rotation maneuver (FABER test). Laboratory studies usually demonstrate an increased white blood cell count with elevated inflammatory markers (ESR, CRP). Plain radiographs are typically normal; the diagnosis is confirmed with an MRI scan, confirmatory laboratory tests, and hopefully positive blood cultures (Grippi et al. 2006; Karmazyn et al. 2006, 2007).

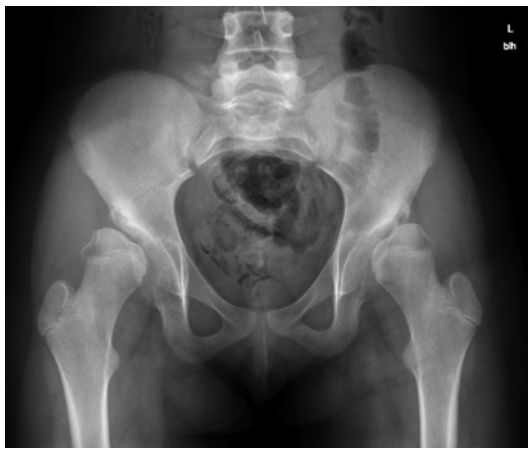

Fig. 36.6 Late presentation of developmental hip/acetabular dysplasia in a 14-year-old girl who had always had a slight limp but just recently began complaining of pain

36.5.8 Neoplastic Processes

These can be either benign or malignant. Benign processes that can result in hip pain in the adolescent are osteoid osteoma and osteochondroma. Malignant processes are typically either Ewing sarcoma (especially the pelvis) or occasionally osteosarcoma.

36.5.9 Trauma

Femoral neck and pelvic ring fractures can result in hip pain in this population age. Plain radiographs are usually sufficient to make the diagnosis (Fig. 36.9).

36.5.10 Overuse Syndromes

These are becoming more common as the intensity and time of children involved in athletics is increasing. Most of the overuse syndromes around the hip are an apophysitis of the pelvis (Chang et al. 2013; Frank et al. 2007, 2013; Hoang and Mortazavi 2012). The most common apophyses

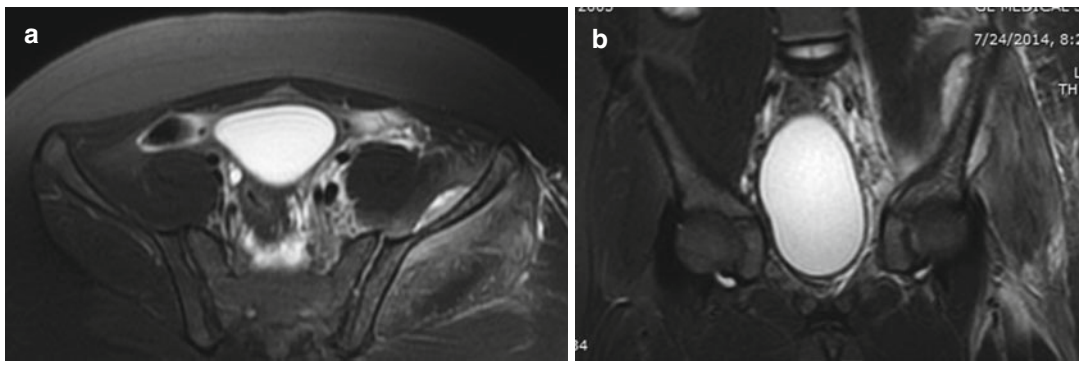

Fig. 36.7 Osteomyelitis of the ilium with subperiosteal abscess and muscle myositis due to MRSA in a 13-year-old girl with a 5-day history of left hip pain, low-grade fever, and inability to bear weight. A plain pelvis radiograph was normal. MRI scans demonstrated the massive subperiosteal abscess of the left ilium and signal change within the ilium indicative of acute osteomyelitis. Edema and myositis in the gluteal muscles were also noted. The axial T2 fat-suppressed image (**a**) and coronal STIR (**b**) images are shown

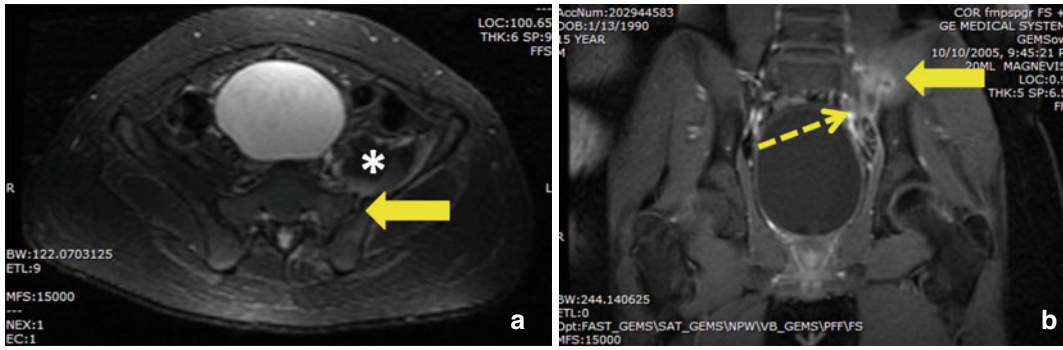

Fig. 36.8 Sacroiliac joint infection with concomitant iliac osteomyelitis and anterior soft tissue abscess in a 14-year-old boy with a 1-week history of progressive buttock/hip pain, increasing fevers, and eventual inability to walk. The axial STIR MR image (**a**) demonstrates a large anterior pelvic abscess (*white asterisk*) originating from the SI joint (*yellow arrow*). The coronal fat-suppressed MR image after gadolinium enhancement (**b**) indicates the significant edema and vascularity (*block yellow arrow*) around the abscess at the level of the SI joint (*dashed yellow arrow*)

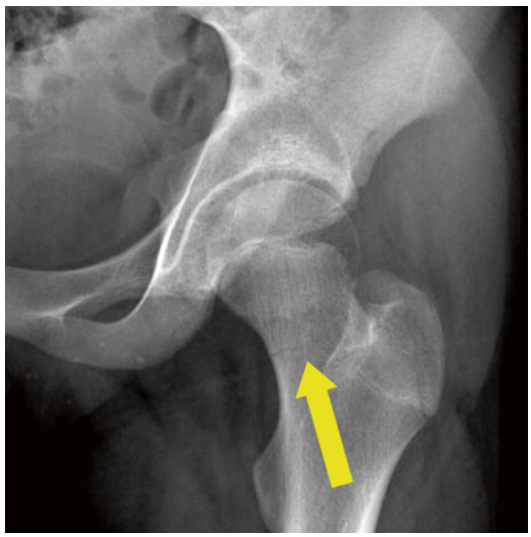

Fig. 36.9 Femoral neck fracture in a 13-year-old girl after an injury on a trampoline. She was unable to bear weight but otherwise was very comfortable. Note the subtle, non-displaced fracture of the femoral neck (*yellow arrow*)

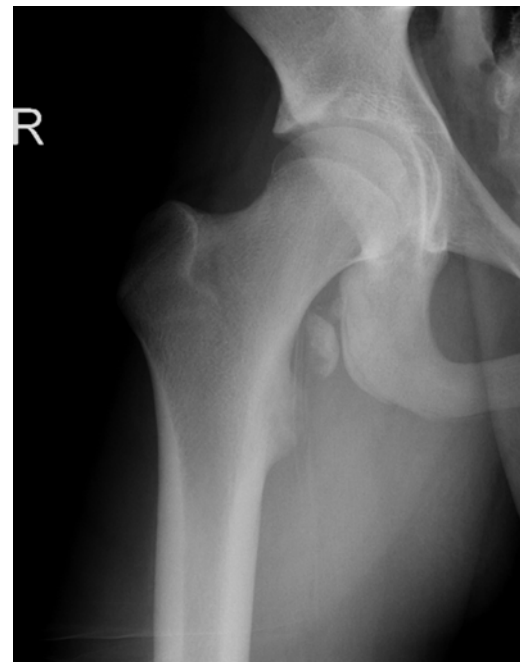

Fig. 36.10 An acute avulsion of the lesser trochanter in a 14-year-old football player

involved are the anterior superior and inferior iliac spines, the ischial tuberosity, and the lesser trochanter. The syndrome initially starts out with stress on the apophysis from prolonged sporting activity and leads to dull, activity-related pain and discomfort at the involved area. This stress weakens the apophyseal attachment, and with continuing rapid contractions of the attached muscles, the apophysis can avulse resulting in severe and sudden pain with the patient often unable to walk. The diagnosis in the pre-avulsed state can usually be made by noting tenderness to palpation at the involved site. Radiographs may show some mild callus at the involved area. In the acute avulsion, radiographs will demonstrate the avulsion, often with significant displacement (Fig. 36.10).

36.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis is shown in Tables 36.1 and 36.2.

Table 36.1 An outline of diagnosis of *acute-onset hip pain* in an adolescent

History			
Unilateral symptoms	Unilateral symptoms	Unilateral symptoms	Unilateral symptoms
Prodromal symptoms may or may not have been present	Prodromal symptoms may or may not have been present	Prodromal symptoms may or may not have been present	No prodromal symptoms
Usually no history of trauma	Mild trauma (e.g., stepping off a curb and falling) may be present	Overuse trauma	Distinct history of trauma
Fever (begins low grade but then increases in magnitude and frequency)	No fever	No fever	No fever
Initially able to bear weight on the limb usually, but then often progresses to inability to weight bear	Complete inability to ambulate with or without crutches	Usually complete inability to ambulate with or without crutches	Usually complete inability to ambulate with or without crutches
Physical examination			
Febrile	Not febrile	Not febrile	Not febrile
+ increased warmth over the SI joint and/or pelvic soft tissues	No warmth noted	No warmth noted	No warmth noted
Tenderness over the sacroiliac joint or iliac crest/gluteal musculatures	Mild tenderness over the hip	Exquisite tenderness over the affected apophysis	Mild to severe tenderness over the hip
Fixed deformities may be present, but difficult to ascertain due to pain with passive range of motion May see flexion, abduction, external rotation contractures and/or positioning	Fixed deformities cannot be assessed – there is complete resistance to any passive range of motion	Fixed deformities cannot be assessed – there is complete resistance to any passive range of motion	Fixed deformities cannot be assessed – there is complete resistance to any passive range of motion Usually lies in a position of mild to severe external rotation depending upon the amount of fracture displacement
Hip movements: Grossly painfully limited in all directions	Hip movements: Grossly painfully limited in all directions	Hip movements: Grossly painfully limited in all directions	Hip movements: Grossly painfully limited in all directions
Investigations			
Plain radiograph of the pelvis: May show: Increase in the medial joint space Distended capsular shadow Erosion of the metaphysis of the femur	Plain radiograph of the pelvis: Will show: Marked displacement of the proximal femoral epiphysis from the metaphysis	Plain radiograph of the pelvis: Will show: Avulsion of the affected apophysis	Plain radiograph of the pelvis: Will show: Femoral neck fracture

Ultrasound: Minimal (reactive) hip joint effusion	Ultrasound: Will demonstrate an effusion due to hemarthrosis	Ultrasound not indicated	Ultrasound not indicated
MRI: An abscess anterior and/or posterior to the sacroiliac joint or signal changes in the ilium/ sacrum In later cases abscesses either anterior to the sacroiliac joint or subperiosteal abscesses (inner or outer tables or both) of the ilium	MRI not indicated	MRI not indicated	MRI not indicated
Laboratory investigations: White cell count elevated (neutrophilia) ESR raised CRP raised Synovial aspirate Normal	Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis
Diagnosis			
Septic sacroiliac joint with or without breakthrough abscess or iliac wing osteomyelitis with or without subperiosteal abscesses	Unstable SCFE	Pelvic apophyseal avulsion injury	Traumatic femoral neck fracture

Table 36.2 An outline of diagnosis of *insidious-onset hip pain* in an adolescent

History				
Unilateral or bilateral	Unilateral or bilateral	Unilateral or bilateral	Usually unilateral	Unilateral or bilateral symptoms
Prodromal symptoms may or may not have been present	Prodromal symptoms may or may not have been present	Prodromal symptoms for several months	Prodromal symptoms for several months	Prodromal symptoms for months to years
Usually no history of trauma	No history of trauma	No history of trauma	No history of trauma	No history of trauma
Can usually bear weight	Can bear weight	Can bear weight	Can bear weight	Can bear weight
Physical examination				
Not febrile	Not febrile	Not febrile	Not febrile	Not febrile
Mild tenderness over the hip may occasionally be present	Minimal tenderness	Minimal tenderness	Minimal tenderness	Minimal tenderness
Fixed deformities may be present and become more noticeable with increasing severity of the SCFE Will see flexion, abduction, external rotation contractures and/or positioning	No fixed deformities	Fixed deformities will be seen late, such as flexion and internal rotation contractures, often with marked decrease in both adduction and abduction	Fixed deformities such as flexion and internal rotation contractures may be seen, often with decrease in both adduction and abduction	Minimal deformity
Hip movements: Mild pain at limits of passive range of motion	Hip movements: Will demonstrate pain with passive hip flexion, adduction, and internal rotation	Hip movements: Pain with passive extension, internal and external rotation, and adduction/abduction	Hip movements: Pain with passive extension, internal and external rotation, and adduction/abduction	Hip movements: Mild pain at limits of passive range of motion
Investigations				
Plain radiograph of the pelvis: Will show: Inferior and posterior displacement of the epiphysis relative to the metaphysis Anterior and superior metaphyseal remodeling seen in the long standing situation	Plain radiograph of the pelvis: May show: Cam type of femoral lesion	Plain radiograph of the pelvis: Will show: Protrusion of the acetabulum into the pelvis, as noted by a crossover of the tear drop iliac and ischial lines	Plain radiograph of the pelvis: May show: Decreased joint space compared to the opposite side	Plain radiograph of the pelvis: Will show: Acetabular dysplasia with varying degrees of subluxation and arthritic changes

Ultrasound not indicated	Ultrasound not indicated	Ultrasound not indicated	Ultrasound not indicated	Ultrasound not indicated
MRI not indicated	MRI may show Increased α angle Acetabular labral tears Lateral acetabular cartilage injury	MRI not indicated	MRI will demonstrate decreased articular cartilage thickness, both femoral and acetabular	MRI will determine if there is any articular cartilage attenuation, labral tears, and obstacles to reduction e.g. (pulvinar)
Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis	Laboratory investigations not indicated to establish diagnosis
Diagnosis				
Stable SCFE	Femoroacetabular impingement	Acetabular protrusio	Idiopathic chondrolysis	Developmental hip dysplasia

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The Painful Knee in Adolescence

37

James Robb

37.1 Introduction

Knee problems are common in adolescence, and as the child approaches skeletal maturity, adult patterns of injury, particularly of the ACL, begin to emerge. It is essential not to overlook referred pain from the hip as the source of the problem, and unfortunately delays in diagnosing a slipped capital femoral epiphysis still occur. Adolescents are often active and minor trauma to the knee is commonplace, but again, it is important not to overlook a more serious diagnosis. For example, delays in diagnosing a malignant primary bone tumor of the distal femur or proximal tibia may occur because pain has been ascribed to recent minor trauma and red flag signs and symptoms have been overlooked.

37.2 Questions to Establish a Diagnosis

37.2.1 Questions in the History

- **Is there a history or trauma and if so, what was the mechanism?**
- **Was the onset of pain sudden or gradual?**
- **What is the site of maximum tenderness?**
- **Is the patient aware of a swelling, and if so, does it fluctuate or does it feel solid?**
- **Does the knee lock?**
- **Does the knee give way?**
- **Does the knee click, and if so, when?**
- **Is there morning stiffness?**
- **Is the pain related or unrelated to activity?**
- **Does pain disturb sleep or not respond to rest and adequate analgesia?**
- **Does the patient have associated groin or anterior thigh pain?**

Is there a history or trauma and if so, what was the mechanism?

Before skeletal maturity, avulsion injuries are more common than in adulthood, so an anterior cruciate ligament (ACL) injury is more likely to comprise an avulsion of the tibial spine in the younger adolescent rather than a mid-substance tear which affects the more skeletally mature adolescent. Although a sleeve fracture of the patella is more likely in juveniles (8–12 years), it may be seen in a skeletally immature adolescent too. The mechanism is of sudden or forced flexion, and there may be patella alta and inferior pole tenderness, and active extension is not possible. The same mechanism can result in a tibial tuberosity avulsion in slightly older patients (12–15 years) who have similar physical findings apart from the tenderness being over the tibial tuberosity instead.

A more distant history of trauma may result in a posttraumatic growth arrest and associated deformity.

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_37

Was the onset of pain sudden or gradual?

Gradual onset of pain may suggest an inflammatory cause or more serious pathology if associated with night pain or a recent swelling.

What is the site of maximum tenderness?

Common sites of localized pain in the knee are the tibial tubercle, distal pole of the patella, patellar tendon, joint line, upper border of the patella, and over the origin of a collateral ligament. More diffuse pain may be seen in an effusion, sepsis, JIA, or tumor.

Is the patient aware of a swelling, and if so, does it fluctuate or does it feel solid?

An effusion is associated with trauma, sepsis, or inflammation. The patient may be aware of a loose body. There are over a dozen bursae which may enlarge.

Does the knee lock?

Locking can be a subjective term, and patients may describe a catching sensation when extending the knee. True locking indicates an inability to extend the knee fully and is associated with a meniscal tear or loose body.

Does the knee give way?

Typically this is a consequence of an ACL injury or may also be seen in conditions resulting in quadriceps weakness. Difficulties with cutting maneuvers suggest a cruciate injury. Some patients may complain of the knee "giving way" when descending the stairs, and this may suggest patellofemoral pathology.

Does the knee click, and if so, when?

It is important to distinguish between "physiologic" clicks which may be audible and are normal for some patients and those that relate to pathology. Lateral patellar subluxation may be seen in the terminal 20° of extension.

Is there morning stiffness?

This suggests an inflammatory cause such as JIA.

Is the pain related or unrelated to activity?

Pain unrelated to activity may suggest an inflammatory cause but also more serious pathology such as infection or tumor. Pain after prolonged sitting may indicate patellofemoral pathology. Pain related to activity suggests a mechanical cause such as osteochondritis dissecans (OCD) or an overuse condition. Pain worse on climbing up and down the stairs or after squatting suggests patellofemoral pathology.

Does pain disturb sleep or not respond to rest and adequate analgesia?

These symptoms suggest serious pathology such as tumor or infection.

Does the patient have associated groin or anterior thigh pain?

Hip pathology must also be considered in patients with "knee pain."

37.2.2 Questions Related to Examination

- **On standing: Is sagittal alignment normal and symmetric?**
- **On standing: Is coronal plane alignment symmetric?**
- **Is there a leg length discrepancy?**
- **Is a swelling visible on the front or back of the knee?**
- **Is there a limp?**

If there is a limp:

Consider the sagittal plane in gait:

- **Is the range of flexion and extension normal?**
- **Is the knee fully extended at initial contact?**
- **Is knee flexion in swing normal?**
- **Does the knee extend fully in gait?**
- **Does the knee hyperextend in stance?**

Consider the coronal plane in gait:

- **Is the knee progression angle normal?**

On standing: Is sagittal alignment normal and symmetrical?

Bilateral recurvatum may suggest joint laxity and bilateral flexed knees in diplegic CP. Unilateral asymmetry suggests pathology. Excessive unilateral recurvatum may be a consequence of a previous growth plate injury or a posterior cruciate insufficiency.

On standing: Is coronal plane alignment symmetrical?

A unilateral abnormality suggests pathology such as a previous growth plate injury.

Is there a leg length discrepancy?

Limb length inequality may be due to growth plate injury. It may also be present in asym-

metric paralysis that involves one limb more than the other.

Is a swelling visible on the front or back of the knee?

An effusion may be posttraumatic or due to a meniscal tear, sepsis, or inflammatory arthritis. If there is a limp:

Is the range of flexion and extension normal?

In normal gait the knee has an arc of about 60° and the hip 45° of motion. Two features that should be considered when observing gait are range of motion (normal, insufficient, or excessive) and timing of events (normal, delayed, or premature).

Is the knee fully extended at initial contact?

Flexion at initial contact can be due to functionally short hamstrings, gastrocnemius, or psoas; knee extensor weakness; a knee, hip, or ankle flexion contracture; or compensation for a contralateral short lower limb.

Is knee flexion in swing normal?

Excessive knee flexion suggests an extensor/flexor imbalance and a relative overaction of the flexors. Insufficient knee flexion may be associated with limited foot clearance and toe drag and may be due to cocontraction of the quadriceps and hamstrings or an extensor/flexor imbalance and a relative overaction of the extensors.

Does the knee extend fully in gait?

Flexed knee gait is seen in neuromuscular disorders, particularly CP and spina bifida. When long standing there is often an associated elongation of the patellar tendon, the patella itself, and patella alta.

Does the knee hyperextend in stance?

This may be a normal variant in a patient who is loose jointed, but it is also seen in conditions where there is weakness of the quadriceps and the patient is using hyperextension to compensate for this and to prevent the knee from giving way in stance. Hyperextension may also occur if there has been damage to the anterior part of the distal femoral or proximal tibial growth plate.

Is the knee progression angle normal?

Usually the patella is about 10–15° external with respect to the plane of progression. If excessively internal, consider hip pathology such as persisting femoral anteversion that has not resolved by adolescence. If excessively external, consider hip pathology such as a retroverted femoral neck or a slipped capital epiphysis.

37.3 Physical Examination

37.3.1 Look

Is thigh muscle bulk symmetrical? Is there an effusion or swelling? Is coronal alignment symmetrical? Are limb lengths equal? Is there a swelling of the tibial tubercle? Have the patient sit on the side of the couch and demonstrate knee extension and flexion: does the patella track normally? Tracking can also be tested by having the patient contract their quadriceps with their knee extended and observe if the patella jumps laterally.

37.3.2 Feel

Is the joint warm? Is there synovial thickening or an effusion? Identify points of maximal tenderness such as the insertion of the quadriceps tendon, the origin and insertion of the patellar tendon, the tibial tuberosity, and the joint lines. There are over a dozen bursae around the knee, and the commonest anterior ones are the supra-, pre-, and infrapatellar bursae.

37.3.3 Move

Check active and passive range of knee motion and note any spasticity. Note any snapping or clicking from the joint during flexion and extension. Palpate the patellar retinacula and compress the patella against the trochlea to evaluate articular pain. Place the patient prone to check rectus femoris length; hold their shin with one hand and place the palm of the other hand over the posterior iliac spine on the same side. Flex the knee gently and if the buttock rises, the rectus femoris is short (Fig. 37.1).

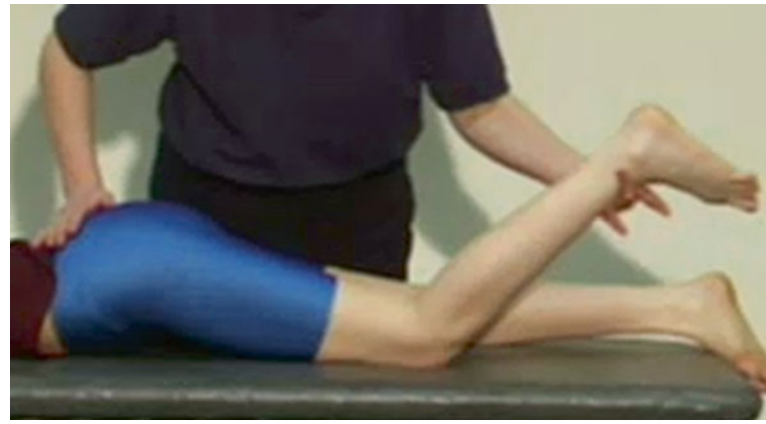

Fig. 37.1 Assessing rectus femoris length. In this example the muscle is short, and by flexing the knee, the ipsilateral hip flexes and the ipsilateral buttock rises off the couch

37.3.4 Special Tests

Perform a standard examination of the knee for ligamentous laxity and a patellar apprehension test if indicated from the patient's history.

37.4 Investigations to Confirm the Diagnosis

37.4.1 Radiography

Standard AP and lateral views may show a loose body and patella alta. An intercondylar view may show pathology such as OCD of the medial condyle, and a skyline view of the patella will help to show the static articular relationship of the patella and the femoral trochlea.

37.4.2 MRI

MRI is helpful to show stability of an OCD fragment, chondral defect, bone bruising, and tendonitis, and specificity for meniscal pathologies such as a tear or a discoid lateral meniscus is improving.

37.4.3 CT Scan

CT is useful to confirm the rotational profile and patellofemoral relationships.

37.5 Differential Diagnosis

37.5.1 Overuse Conditions

The knee is a common site for overuse problems, and it is probably more accurate to avoid eponymous names in their description. The diagnosis can usually be made from the history and physical examination, and further imaging is not usually necessary unless there are concerns about more serious pathology. Areas typically affected are the tibial tubercle, proximal and distal poles of the patella, the patellar tendon, and the medial collateral ligament. Persisting traction on the tibial tuberosity can result in its enlargement and a visible swelling.

37.5.2 Intraarticular Pathology

Bony Pathology

The commonest nontraumatic condition is OCD which usually affects the lateral aspect of the medial femoral condyle (Fig. 37.2) although the patella can also be affected. The patient may complain of joint pain worse after activity and intermittent effusion. A separate fragment can cause typical symptoms of a loose body with intermittent true locking and the sensation of something moving around the knee. The femoral OCD lesion may be seen on plain radiography or an intercondylar view and the patellar lesion on a skyline view, but MRI is key in determining the site, extent, and fragment prognosis. Unlike the

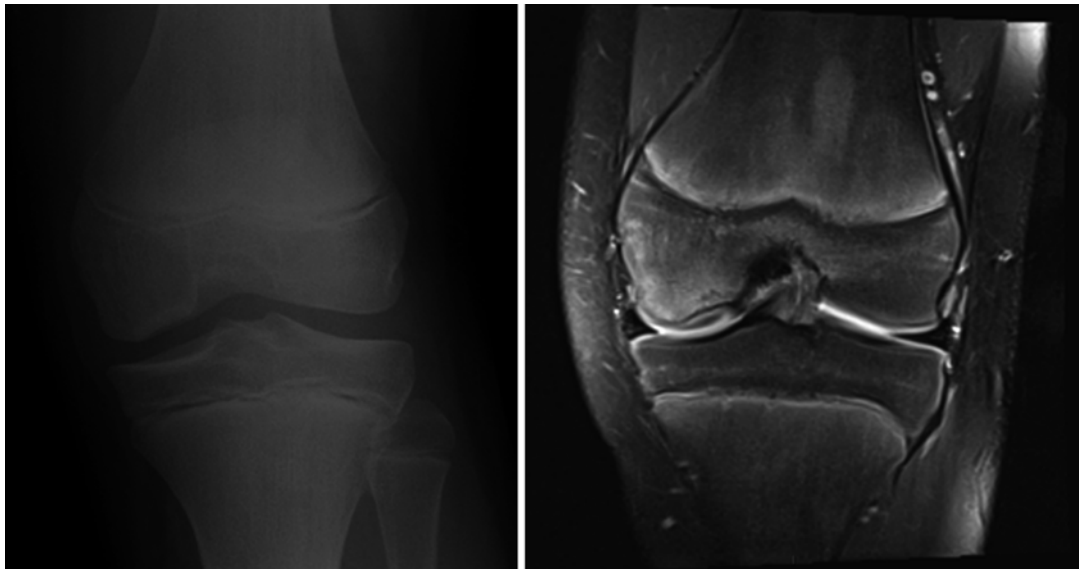

Fig. 37.2 OCD of the medial femoral condyle. The MRI shows that the overlying articular cartilage is intact

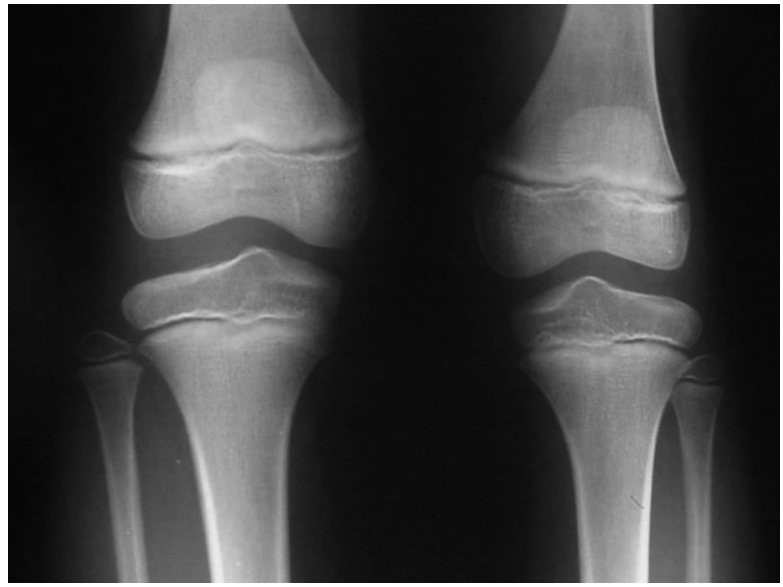

Fig. 37.3 AP radiograph of the knees showing widening of the lateral joint line in patient's right knee due to a discoid lateral meniscus

juvenile type, the prognosis for healing is less certain in adolescents (Detterline et al. 2008).

Soft Tissue Pathology

A discoid lateral meniscus may cause lateral joint line tenderness and a snapping sensation and later mechanical symptoms if a tear develops. The condition is more commonly seen in juveniles than adolescents (Fig. 37.3).

37.5.3 Patellofemoral Disorders

Nonspecific Anterior Knee Pain

This condition is poorly understood but is usually self-limiting, and it is important to exclude an underlying diagnosis, particularly referred pain from hip pathology. The term "chondromalacia patellae" as a descriptor of nonspecific anterior knee pain is inaccurate.

Patellar Subluxation

In patellar mal-tracking, the patella fails to track symmetrically in the trochlear groove during flexion and extension and usually displaces laterally in flexion. There may be an underlying hypoplastic lateral condyle and a contracture of the lateral side of the quadriceps mechanism. Subluxation may also be seen in generalized joint laxity and external tibial torsion.

Bipartite Patella

This is usually an incidental radiological finding and the bipartite fragment is superolateral.

37.5.4 Inflammatory Causes

JIA, pigmented villonodular synovitis (although rare), and hemophilia should be considered as part of the differential diagnosis of a knee effusion in the absence of trauma and in a patient who is well (Fig. 37.4). Septic arthritis and osteomyelitis of the distal femoral metaphysis, which is intraarticular, should be considered in a patient who is unwell and febrile and has an effusion.

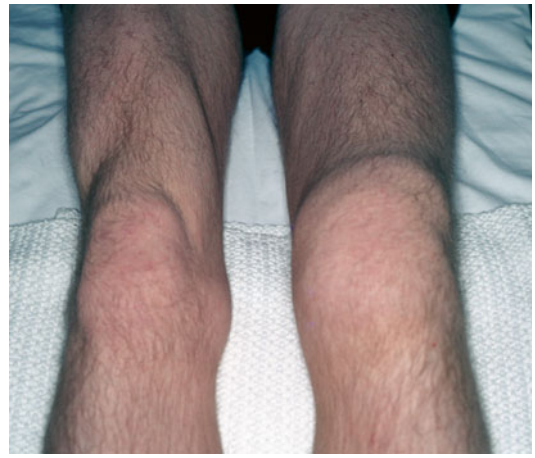

Fig. 37.4 Large effusion of the left knee showing the margins of the suprapatellar bursa

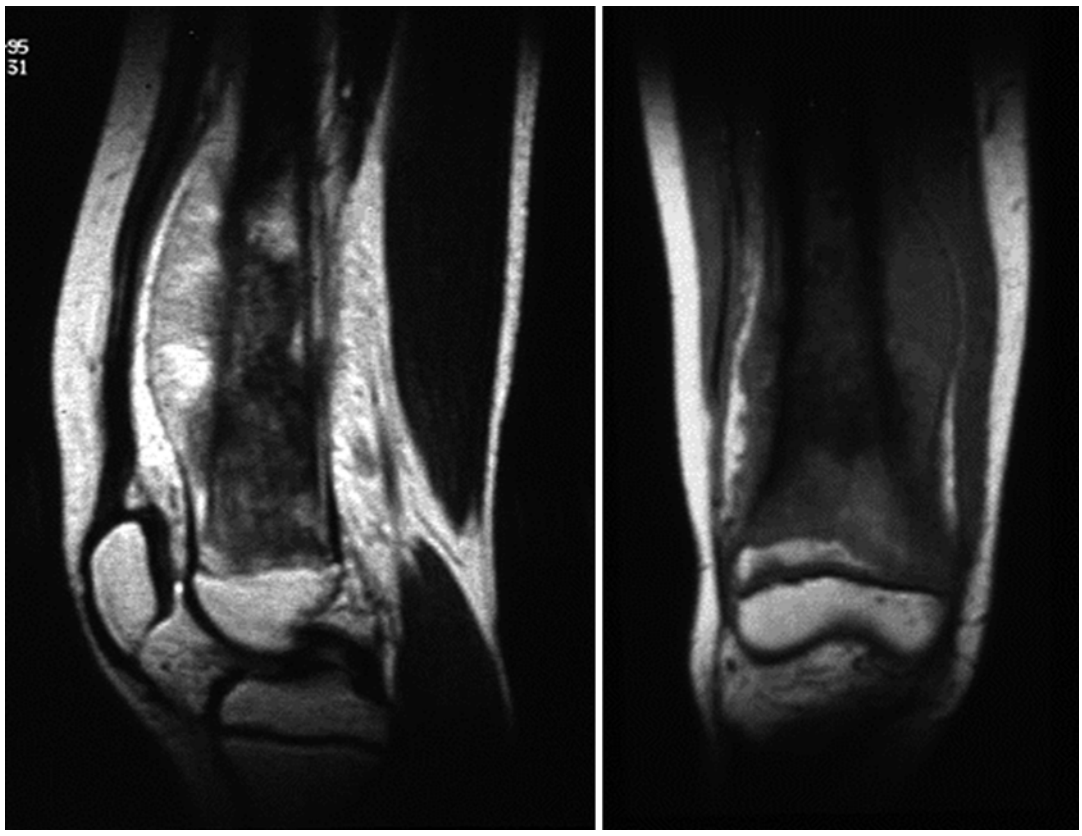

Fig. 37.5 MRI showing the soft tissue and osseous components of a distal femoral osteosarcoma

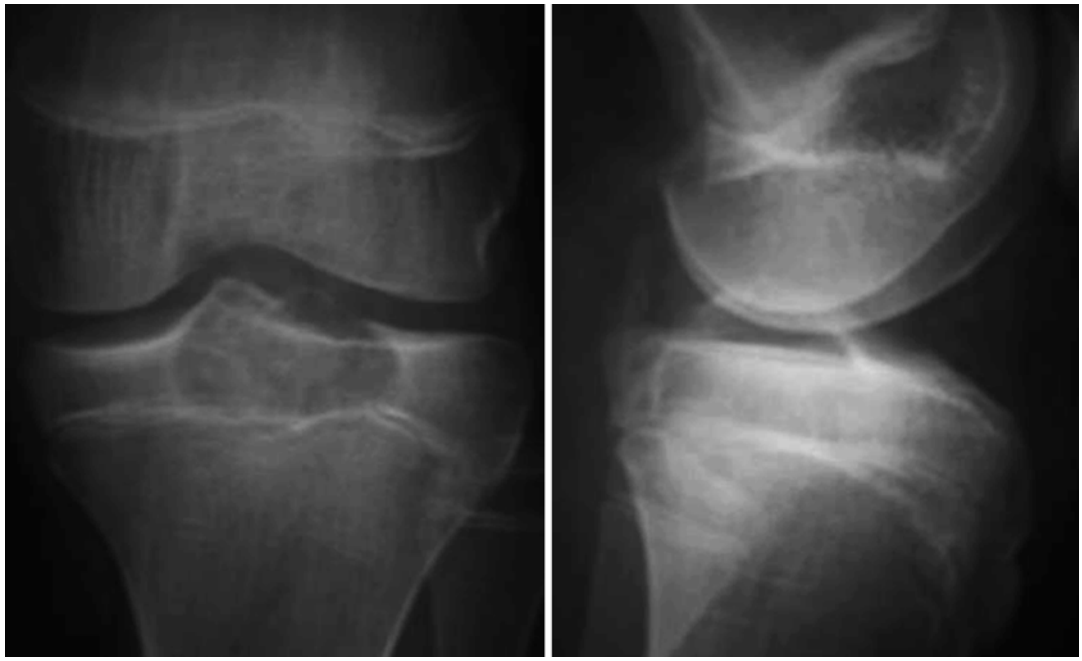

Fig. 37.6 This lytic lesion of the tibial epiphysis was a chondroblastoma

37.5.5 Tumors

The proximal tibial and distal femoral metaphyses are typical sites for osteosarcoma (Fig. 37.5) and Ewing's sarcoma which are the commonest adolescent primary malignant bone tumors.

Benign tumors that may be painful include an osteochondroma (Fig. 37.6) causing rubbing of an overlying muscle group or tendon, osteoid osteoma, and chondroblastoma, an epiphyseal cartilaginous tumor (Fig. 37.7).

37.5.6 Trauma

As children approach skeletal maturity, adult patterns of ACL injury appear rather than an avulsion of the tibial spine. The history is characteristic involving a rotational mechanism, and the patient may recall a "popping" sensation and rapid joint swelling due to a hemarthrosis (Frank and Gambacorta 2013; Pandya et al. 2014). A PCL injury may result from a direct blow to the tibia and knee in hyperextension (Fig. 37.8).

The differential of an acute knee swelling should include ACL tear, osteochondral fracture (Fig. 37.9), meniscal tear, and bone bruising with microfractures. Acute ligament injury in this age group may be difficult to diagnose on ligament laxity tests because of pain. Plain radiography will demonstrate most fractures and growth plate injuries, and MRI will demonstrate bone bruising and is becoming more specific and sensitive for ligament and meniscal injury.

A sleeve fracture of the patella after forced flexion of the knee usually affects juveniles but may be seen in young adolescents who are more skeletally immature than their peers. However, an avulsion of the tibial tuberosity (Fig. 37.10) is seen in adolescence, and the mechanism of injury is similar – forced knee flexion resulting in patella alta and loss of active knee extension.

37.5.7 Knee Bursae

There are at least a dozen bursae associated with the knee, and the largest and most important clinically is the suprapatellar bursa (see Fig. 37.3).

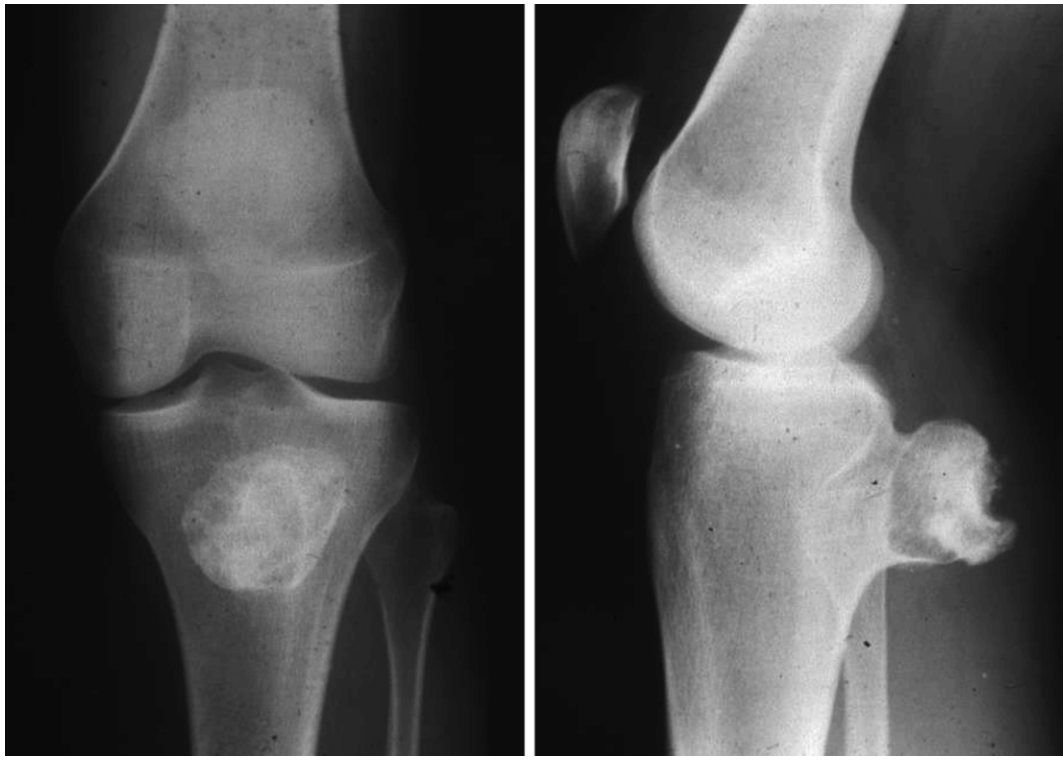

Fig. 37.7 Osteochondroma of the proximal tibia; the patient complained of a swelling in the calf and discomfort when moving his ankle

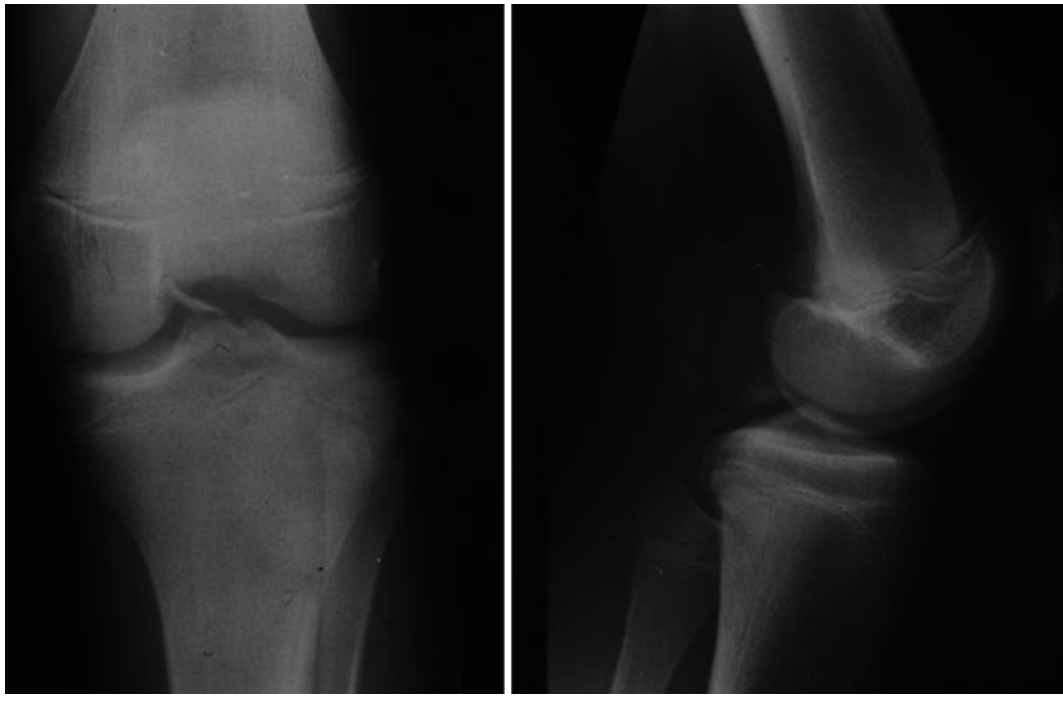

Fig. 37.8 Avulsion of the bony tibial origin of the PCL. The bony fragment is seen in the intercondylar area posteriorly

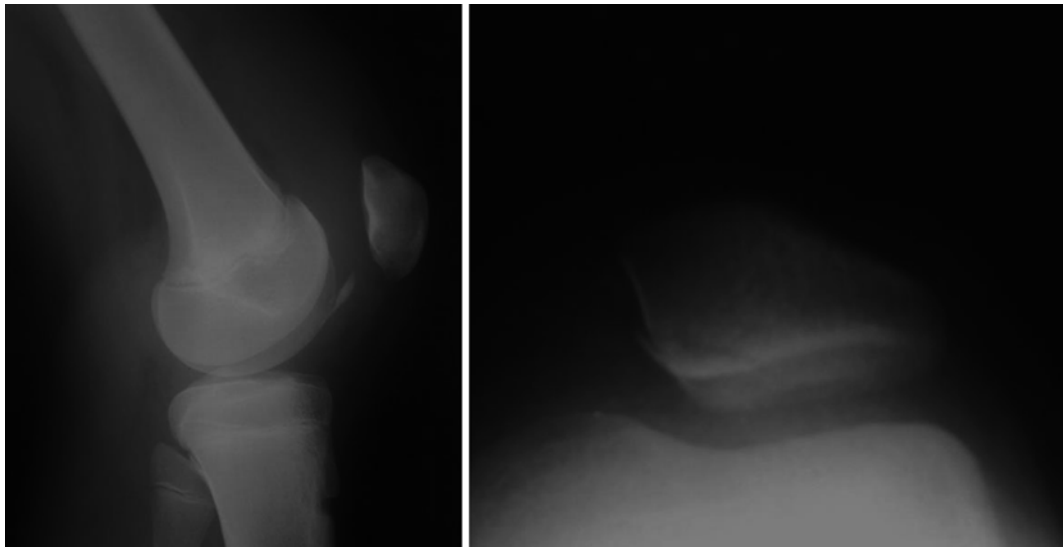

Fig. 37.9 Lateral and skyline radiographs showing an osteochondral fragment originating from the patella lying anteriorly to the femoral condyles (*left*) and a coronal fracture of the patella (*right*)

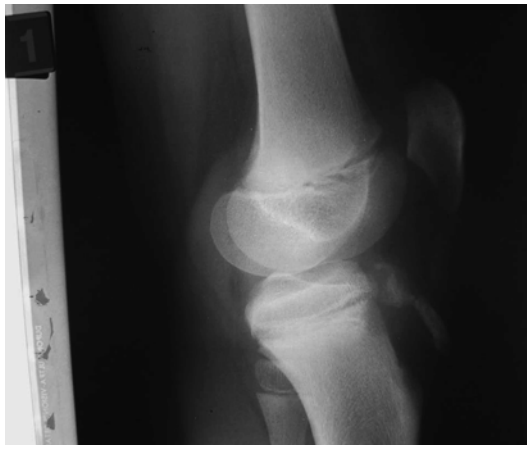

Fig. 37.10 Avulsion of the tibial tuberosity

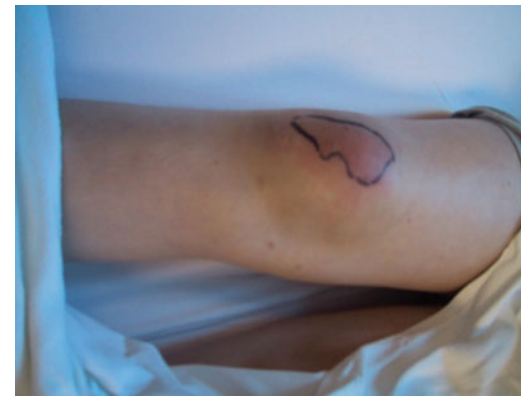

Fig. 37.11 Prepatellar bursitis

Patients may complain of pain and rubbing over the prepatellar (Fig. 37.11), pretibial, and pes anserinus bursae. A semimembranosus bursa should be considered as part of the differential diagnosis of a medial knee swelling.

37.5.8 Knee Pain in Neuromuscular Disorders

This is most commonly seen in flexed knee gait diplegic cerebral palsy. The pain is patellofemoral, and often patients have patella alta and an elongated patellar tendon and patellar bone (Fig. 37.12).

A stress fracture of the inferior pole of the patella may also be seen when symptoms are particularly acute (Fig. 37.13). Similar features are seen in patients who have L4/5 spina bifida as they rely on their quadriceps for walking because the muscles below the knee are paralyzed.

Although knee hyperextension is seen in CP, it is more characteristic of conditions, such as polio, resulting in quadriceps weakness.

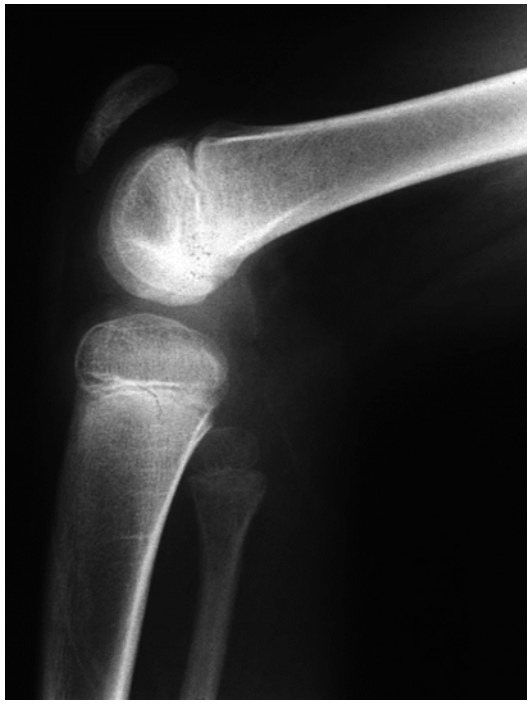

Fig. 37.12 An elongated patella seen in a patient with diplegic CP who walked with a flexed knee (crouch) gait

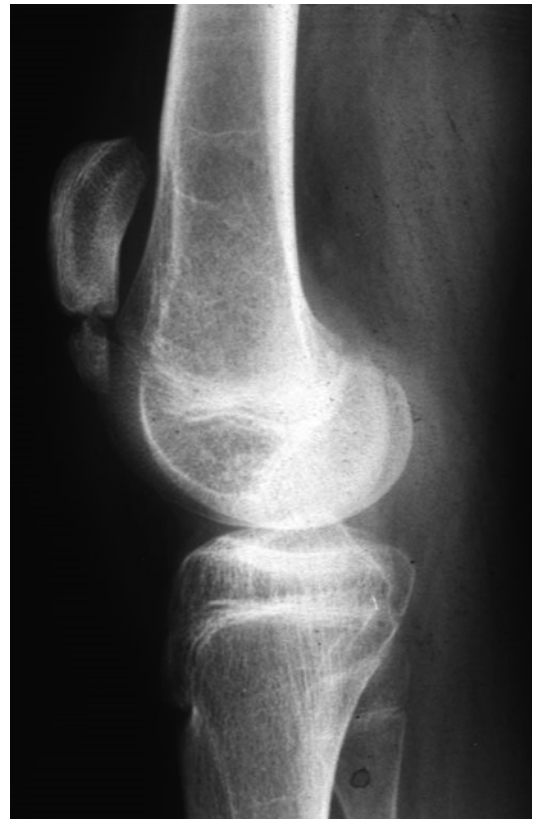

Fig. 37.13 Patella alta and an avulsion of the inferior pole of the patella in a diplegic who walked with a flexed knee (crouch) gait

37.5.9 Less Common Causes of Knee Pain

Plica

These are usually variants of normal anatomy but very occasionally can become entrapped between the patella and a femoral condyle causing pain over the medial femoral condyle particularly when squatting.

Metabolic Bone Disease

Severe vitamin D deficiency can result in bone pain.

Complex Regional Pain Syndrome Type 1

This may result from previous trauma, and the patient complains of exquisite local tenderness and changes in skin color and temperature.

Avascular Necrosis After Chemotherapy

Chemotherapy for leukemia and other malignancies can result in avascular necrosis of bones in various sites of the body. The femoral head is a common site for this complication, and pain may be referred to the knee. The distal femur or the upper tibia may be affected with localized pain in the region of the knee (Fig. 37.14).

37.6 Establishing the Diagnosis

An outline of the approach to establishing a diagnosis of chronic nontraumatic knee pain in the adolescent is shown in Table 37.1.

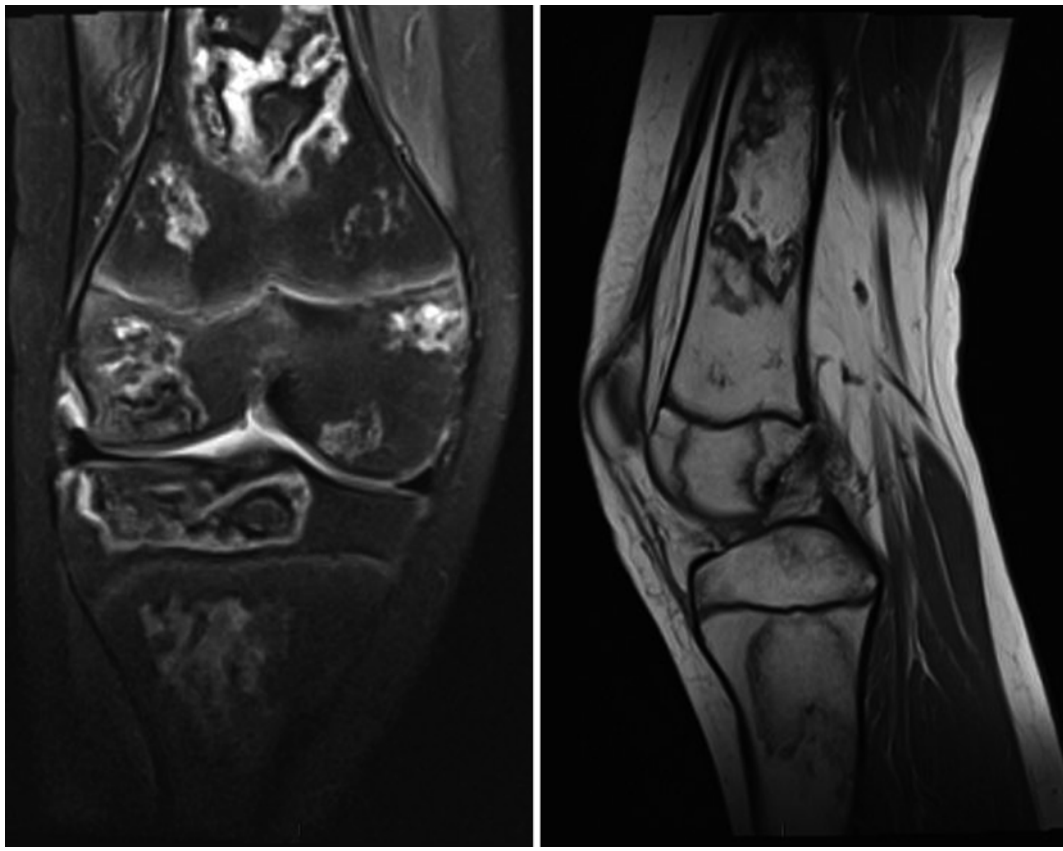

Fig. 37.14 MRI showing bone infarcts in a patient treated with chemotherapy and steroids for leukemia

Table 37.1 Establishing the diagnosis of nontraumatic chronic knee pain in the adolescent

History				
History suggestive of overuse	No history suggestive of overuse	No history suggestive of overuse	No history suggestive of overuse	No history suggestive of overuse
Pain related to activity Seldom present at rest	Pain related to activity	Pain may be related to activity	Pain may be aggravated by activity	Pain not related to activity May be present at rest
Pain localized to site of attachment of quadriceps tendon, patellar tendon, or collateral ligament	Pain localized to joint line	Pain localized to the patella and the front of the knee	Pain in the suprapatellar region and the joint line	Pain may be poorly localized
No history of effusion, giving way	History of recurrent episodes of effusion may be present History of locking may be present	History of patella slipping laterally may be present	History of recurrent or persistent effusion frequently present	History of effusion seldom present
Physical examination				
Tenderness at one of the following sites: Proximal pole of patella Distal pole of patella Tibial tubercle Medial collateral ligament Patellar tendon	Tenderness in anterior joint line	Tenderness either under the patella or on the medial border of the patella	Tenderness in the joint line and over the suprapatellar bursa	Tenderness either in the metaphyseal or epiphyseal region of the femur or tibia
No effusion	Effusion may be present	Effusion not present unless recent episode of patellar dislocation	Effusion present	No effusion
Patellar mal-tracking not present	Patellar mal-tracking not present	Patellar mal-tracking/subluxation may be present	Patellar mal-tracking not present	Patellar mal-tracking not present
Patellar apprehension test negative	Patellar apprehension test negative	Patellar apprehension test may be positive	Patellar apprehension test negative	Patellar apprehension test negative
–	–	In adolescent with CP – crouch gait with patella alta may be present	–	–
Range of passive movement of the knee normal	Range of passive movement of the knee normal	Flexion deformity may be present with limitation of extension in CP Flexion may be limited due to apprehension of patellar dislocation	Range of passive motion restricted due to effusion and pain	Range of passive movement usually normal
Working diagnosis: Overuse syndrome	Working diagnosis: Intraarticular pathology	Working diagnosis: Patellofemoral disorder	Working diagnosis: Inflammatory arthritis	Working diagnosis: Tumor in the vicinity of the knee

Investigations				
Plain radiograph: Usually not needed If taken may show characteristic changes in the tibial tubercle or the distal pole of the patella	Plain AP radiograph: May show a lesion in the lateral aspect of the medial femoral condyle An intercondylar tunnel view of the knee will demonstrate the lesion better	Plain AP radiograph: May show genu valgum Lateral radiograph may show patella alta and fragmentation of patella in adolescent with CP Skyline view may show patellofemoral malalignment	Plain radiograph: May show periarticular osteopenia The joint space may be reduced	Plain radiograph: Lytic or sclerotic lesion may be seen in the epiphyseal or metaphyseal region of the femur or tibia
MRI not indicated	MRI: Will demonstrate OCD lesion and also demonstrate if the fragment is loose	MRI not indicated	MRI not indicated	MRI: Will demonstrate the nature and extent of the lesion
		CT scan may be useful in demonstrating and measuring torsional deformities of the femur and tibia		
Diagnosis				
Overuse syndrome	Osteochondritis dissecans	Patellofemoral pathology such as: Recurrent subluxation or dislocation of the patella Patellar mal-tracking associated with femoral anteversion or genu valgum	Inflammatory arthritis such as: JIA Seronegative arthritis	Benign or malignant bone tumors such as: Chondroblastoma Osteoid osteoma Ewing's tumor Osteosarcoma

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Further Reading

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The Painful Foot in Adolescence

38

James Robb

38.1 Introduction

By this age normal variants are a less common cause for concern, and foot pain is more likely to result from a trauma, a structural cause, an infection, or, rarely, a tumor. An adolescent presenting with persisting foot pain should therefore be investigated to determine a diagnosis to explain their symptoms.

38.2 Questions to Establish a Diagnosis

38.2.1 Questions in the History

- **Was the onset of pain sudden or gradual?**
- **What is the site of pain?**
- **Are there relieving or exacerbating features?**
- **Is the patient aware of a deformity or swelling?**
- **Is there morning stiffness or pain unrelated to activity?**
- **Do symptoms worsen as the day goes on?**
- **Does pain disturb sleep, or does it not respond to rest and adequate analgesia?**
- **Is the pain related to footwear?**

Was the onset of pain sudden or gradual?

If the onset was sudden, consider trauma and infection as the most likely causes. If infection is suspected, enquire about local causes such as puncture wounds, infected skin lesions of the foot, or direct foot trauma. Do not forget to enquire about other potential sources such as throat or respiratory symptoms, earache or discharge, dysuria, and lower abdominal pain. Is there a history of previous musculoskeletal infection or immunodeficiency?

What is the site of pain?

This will help guide physical examination of the ankle, hindfoot, midfoot, forefoot, or toes.

Are there relieving or exacerbating features?

Pain worsening after activity and relieved by rest is suggestive of a structural or overuse problem in the foot. Consider causes such as a stress fracture, osteochondritis (particularly if there is an associated swelling, e.g., of the ankle), tarsal coalition, and accessory navicular.

Is the patient aware of a deformity or swelling?

A unilateral flatfoot usually indicates pathology such as a tarsal coalition. Does the patient have cerebral palsy? Bilateral flatfeet may be physiologic or a consequence of a short gastrocnemius and/or soleus. A heel bump (calcaneal boss) is usually seen on the superolateral corner of the calcaneus. An accessory navicular may first present as a medial prominence of the foot. Toe deformities and

B. Joseph et al., *Paediatric Orthopaedic Diagnosis: Asking the Right Questions*,
DOI 10.1007/978-81-322-2392-4_38

associated rubbing of the overlying skin are common – does the patient have a bunion, hammer, claw, or overriding toes?

Is there morning stiffness or pain unrelated to activity?

The foot is a common site of symptoms in juvenile inflammatory arthritis (JIA). Joint stiffness may also be a symptom of noninflammatory conditions such as tarsal coalition or a hallux rigidus.

Do symptoms worsen as the day goes on?

This may suggest a neuromuscular condition.

Does pain disturb sleep, or does it not respond to rest and adequate analgesia?

This suggests serious pathology such as tumor or infection. If there are progressive neurological symptoms, consider neuromuscular disorders and spinal cord tumor or spinal infection.

Is the pain related to footwear?

Pain in the foot may often be related to pressure of the footwear on a bony prominence such as the medial aspect of the great toe in an adolescent with hallux valgus. The symptoms may be aggravated if the footwear has a high heel or if the toe box is narrow. Similarly pain from shoe wearing can occur in the base of the fifth metatarsal or the calcaneal tuberosity if it is prominent.

38.2.2 Questions Related to Examination

- **Is the medial longitudinal arch normal, accentuated, or flat?**
- **Is there a limp?**
- **If there is a limp: see Chap. 24.**

Consider the sagittal plane in gait:

- **Are the three foot rockers present?**
- **Which part of the foot makes contact with the ground at initial contact?**
- **Does the tibia progress over the foot normally?**
- **Is foot clearance in swing normal or is there toe drag?**
- **Is foot prepositioning at the end of swing normal?**

Consider the coronal plane in gait:

- **Is the foot progression angle within normal limits and symmetrical?**
- **Is the coronal alignment of the thigh and shank normal?**
- **Is there asymmetry of coronal plane alignment?**
- **Does the heel go into physiologic varus in swing and physiologic valgus in stance?**

Is the arch normal, accentuated (cavus), or flat (planovalgus)?

Ask the patient to stand on tiptoe; normally the heel will shift into varus when standing on tiptoe and go into physiologic valgus in stance, when viewed from behind. If not, seek causes of a structural flatfoot. The commonest cause in this age group is a tarsal coalition. Pes valgus is also seen in an overcorrected clubfoot and skewfoot and in true ankle valgus in spina bifida.

Increased height of the medial longitudinal arch may be physiologic but the commonest underlying cause is neuromuscular disease. Unilateral cavus should be assumed to be pathologic and an underlying cause sought.

Is there a limp?

If there is a limp, the gait must be observed in greater detail.

Consider the sagittal plane in gait:

Are the three foot rockers present?

These are three rotational events of the foot – heel contact, tibial progression over the stationary foot, which is equivalent to ankle dorsiflexion, and then a rotation about the metatarsal heads before foot off, which is equivalent to ankle plantar flexion (Fig. 38.1). If ankle motion is painful or if the ankle is stiff, one or more of these rockers will not be present.

Which part of the foot makes contact with the ground at initial contact?

Normally this is the heel, but foot flat or toe contact may suggest an imbalance of the posterior and anterior calf muscles (short/weak/overactive posterior and long/weak anterior), an ipsilateral knee or hip contracture, or a compensation for an ipsilateral short lower limb. A child with a painful heel may prefer to walk on tiptoe to avoid weight bearing on the heel.

Is the timing of heel lift normal?

Normally this should not occur until the opposite (swing) foot has passed the stance heel.

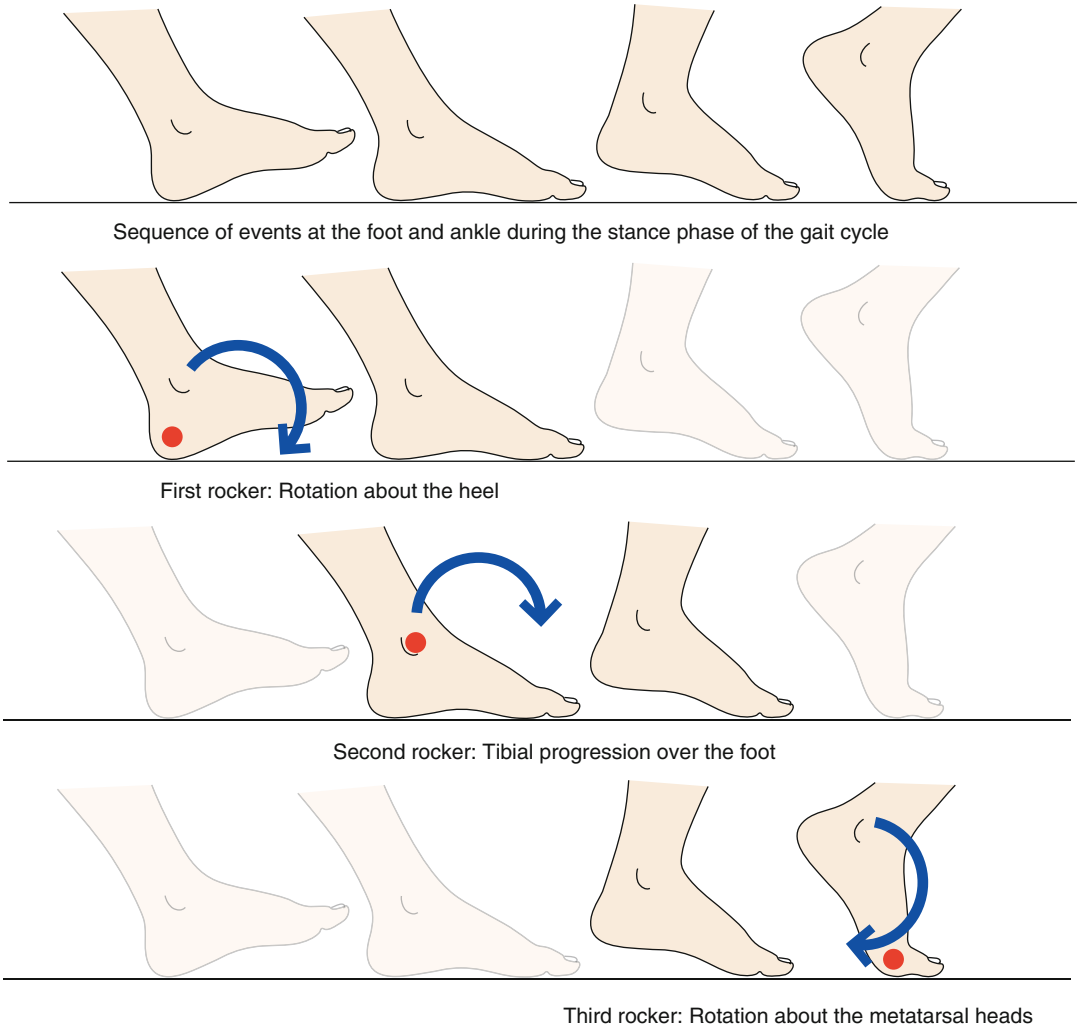


Fig. 38.1 Rocker sequence in the foot

Premature heel lift may occur as a result of a functionally short gastrocnemius/soleus or a mechanism to assist with foot clearance in the opposite limb (hip hitching).

Does the tibia progress over the foot normally?

Excessive tibial progression may suggest weakness of gastrocnemius or soleus permitting the ankle to go into excessive dorsiflexion.

Is foot clearance in swing normal or is there toe drag?

This may suggest weakness of tibialis anterior or shortening of the posterior calf muscles or a combination of both.

Is foot prepositioning at the end of swing normal?

Tibialis anterior should be active at the end of swing to enable initial contact by the heel.

Consider the coronal plane in gait:

Is the foot progression angle within normal limits and symmetrical?

After about the age of 8 years, there is little further potential for resolution of normal variants such as in- or out-toeing. A child with painful limitation of ankle movement may walk with an out-toeing gait to avoid the normal rockers during the stance phase of gait which entail dorsiflexion and plantar flexion of the ankle.

Is the coronal alignment of the thigh and shank normal?

Excessive genu valgum or external tibial torsion without a compensatory internal femoral torsion will give the appearance of a "flatfoot."

Is there is asymmetry of coronal plane alignment?

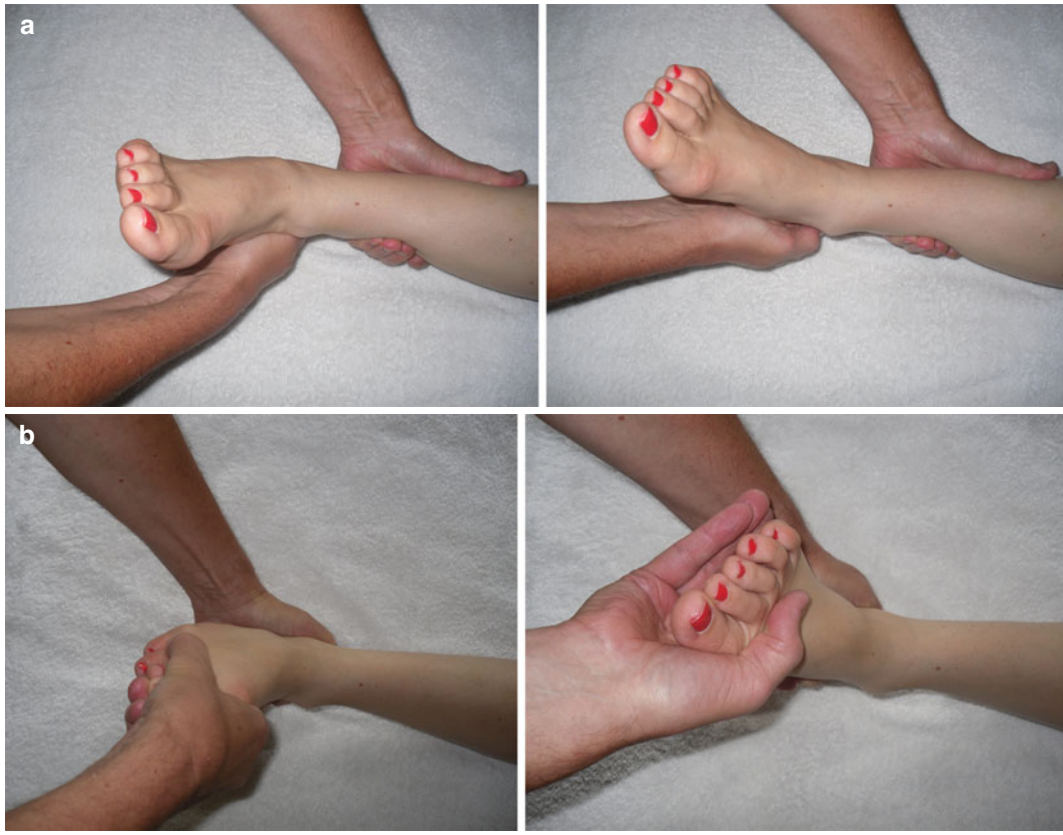


Fig. 38.2 Isolating hindfoot (**a**) and midfoot (**b**) components of inversion and eversion

If so, look for a more proximal cause, e.g., hip dysplasia or hemiplegia.

Does the heel go into physiologic varus in swing and physiologic valgus in stance?

If not, consider hindfoot pathology such as a talocalcaneal coalition or an imbalance of the invertors and evertors.

38.3 Physical Examination

38.3.1 Look

Look for swellings on the medial aspect of the midfoot, the heel, and the toes (e.g., an accessory navicular, heel bump, an adolescent bunion, or toe deformities). While the child is standing, note if the arch is well formed, accentuated, or lost. Observe if the hindfoot is in valgus. Note if there are deformities of the forefoot or the toes. Observe the gait pattern and look for gait deviations listed above.

38.3.2 Feel

Feel if there is localized warmth or tenderness. The calcaneal apophysis may be tender in cases of overuse, an accessory navicular and associated tibialis posterior tendon may be tender, or there may be redness due to rubbing from shoe wear over an accessory navicular or bunion. If there is tenderness and swelling of the dorsal aspect of the second metatarsal, it is suggestive of Freiberg's infraction.

38.3.3 Move

Observe active dorsiflexion and plantar flexion at the ankle and combined inversion and eversion in the hind- and midfoot and toe movement. Inversion and eversion are a combination of movements occurring at the subtalar, calcaneocuboid, and talonavicular joints, and the midfoot contribution can be separated from that of the hindfoot. Check these movements passively and actively (Fig. 38.2).

Check relevant muscle lengths, and if shortening of the gastrocnemius or soleus is suspected, perform Silferskiöld's test (see Chap. 22). Check the movements of the metatarsophalangeal joints of the toes.

Do not omit a general orthopedic examination of the lower limbs and spine, and if a neuromuscular cause is suspected, perform a screening neurological examination and include strength testing of major muscle groups.

38.4 Investigations to Confirm the Diagnosis

Imaging

Radiographs of the foot should generally be taken with the child standing; the weight-bearing lateral radiograph is particularly informative. An oblique radiograph should be taken to exclude a calcaneonavicular bar and an ankle mortise view if osteochondritis dissecans (OCD) of the talus is suspected. Further details of coalitions are usually best seen on a CT scan, and the stability of an OCD fragment is best evaluated with MRI.

Other Investigations

If sepsis (septic arthritis or acute osteomyelitis) is suspected particularly in children with an acute onset of foot pain, full blood count, ESR, CRP, and blood culture should be done straight away.

A rational approach to the choice of imaging and investigations is shown in Table 38.1.

Table 38.1 Recommended investigations based on the clinical situation in an adolescent with a painful foot

Clinical situation	Investigation
Trauma, fracture suspected	Plain radiographs, consider MRI or CT for complex injuries such as a talar fracture, growth plate injury, osteochondral fracture, or tarsometatarsal disruption (Lisfranc injury)
Trauma, soft tissue injury suspected	Clinical examination usually suffices but obtain plain radiographs if an associated fracture cannot be excluded
Overuse condition suspected from history of pain made worse by activity and relieved by rest, and patient can clearly localize the pain	Plain radiography not needed if tenderness is clearly localized to the calcaneal apophysis, Achilles tendon, or origin of the plantar fascia
Overuse condition suspected from history of pain made worse by activity and relieved by rest, but patient cannot clearly localize the pain	AP and lateral radiograph of the foot
Septic arthritis suspected	Full blood count, ESR, CRP, blood culture, and plain radiographs, and obtain an aspirate for culture and sensitivities and microscopy
Osteomyelitis suspected	Full blood count, ESR, CRP, blood culture, and plain radiographs. Consider ultrasound or MRI to demonstrate an early periosteal elevation and a subperiosteal abscess and to localize the site and extent of infection clearly
OCD of the ankle suspected from the history	Plain radiography to include a mortise view to demonstrate OCD of the talus. Consider MRI to evaluate fragment stability and viability
Tarsal coalition suspected	Obtain AP, lateral and oblique views of the foot. CT will give further definition of a coalition
Pes cavus	Obtain a standing AP and lateral of the foot
Toe deformities	Plain radiograph of the foot when standing if there is a bunion. Lesser toe deformities can be evaluated clinically
Inflammatory arthritis suspected	Full blood count, ESR, CRP, rheumatoid factor, antinuclear antibodies, and HLA-B27. Plain radiographs
Benign tumor suspected	Obtain AP, lateral radiographs. Consider a bone scan or CT to localize an osteoid osteoma
Malignant tumor suspected	Plain radiography and obtain an MRI for local imaging. Full blood count and film to exclude leukemia, ESR, CRP, and blood culture to exclude infection which can mimic malignancy radiologically. If malignancy is confirmed, obtain a bone scan to evaluate possible skip lesions and the remainder of the skeleton, a chest x-ray and chest CT for lung metastases. Refer to a regional tumor center for biopsy

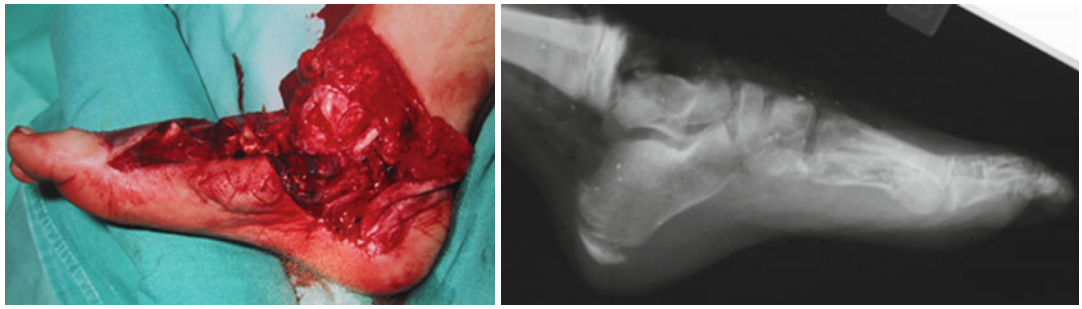


Fig. 38.3 Severe open trauma of the foot

38.5 Differential Diagnosis

38.5.1 Trauma

Acute trauma should be evident from the history (Fig. 38.3), and metatarsals are the commonest site. A stress fracture, particularly of a metatarsal, should be considered in a patient who complains of foot pain, is very active, and can localize the pain, and examination is otherwise unremarkable. A stress fracture may not show up on an initial radiograph and is worth repeating 14 days later to see if there is a periosteal reaction. AVN of the talus may ensue after a previous talar fracture.

38.5.2 Infection

Septic arthritis can affect joints in the foot (Fig. 38.4) and should be considered in a patient who has nonmechanical foot pain, is unwell, and shows signs of sepsis clinically. Osteomyelitis in the foot (Fig. 38.5) is not as common as in other parts of the skeleton but should also be considered in cases of nonmechanical pain. Infections in the hindfoot, particularly the calcaneus, may be indolent and do not have such an acute course as in a long bone (Fig. 38.6).

There may be a history of a stubbed toe and nail bed disruption, and the patient presents with a later infection of the distal phalanx.

A history of a puncture wound should raise the possibility of osteomyelitis and *Pseudomonas* as an infecting organism.

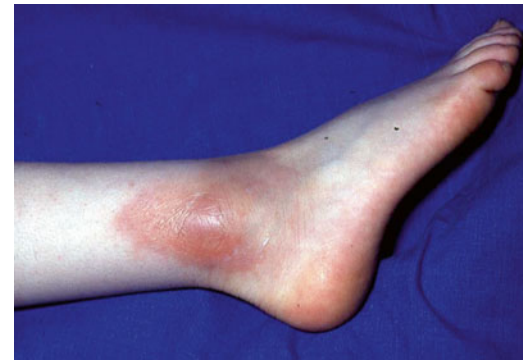


Fig. 38.4 Infection; the differential diagnosis includes cellulitis, septic arthritis of the ankle, or osteomyelitis of the fibula or hindfoot

38.5.3 Osteochondritis Dissecans

The commonest site in the foot is the talus where the anterolateral and posteromedial aspects of the talar dome are typical sites. The patient may complain of mechanical type of pain, intermittent stiffness, and possibly ankle swelling. The fragment may be undisplaced and stable, loose, or displaced and is best evaluated with MRI.

38.5.4 JIA

Juvenile inflammatory arthritis should be considered and excluded in patients who present with nonmechanical foot pain, intermittent fever, and possibly a rash. Morning stiffness may be a feature that tends to improve as the day wears on. Typically the affected joint is warm but tenderness may be diffuse.

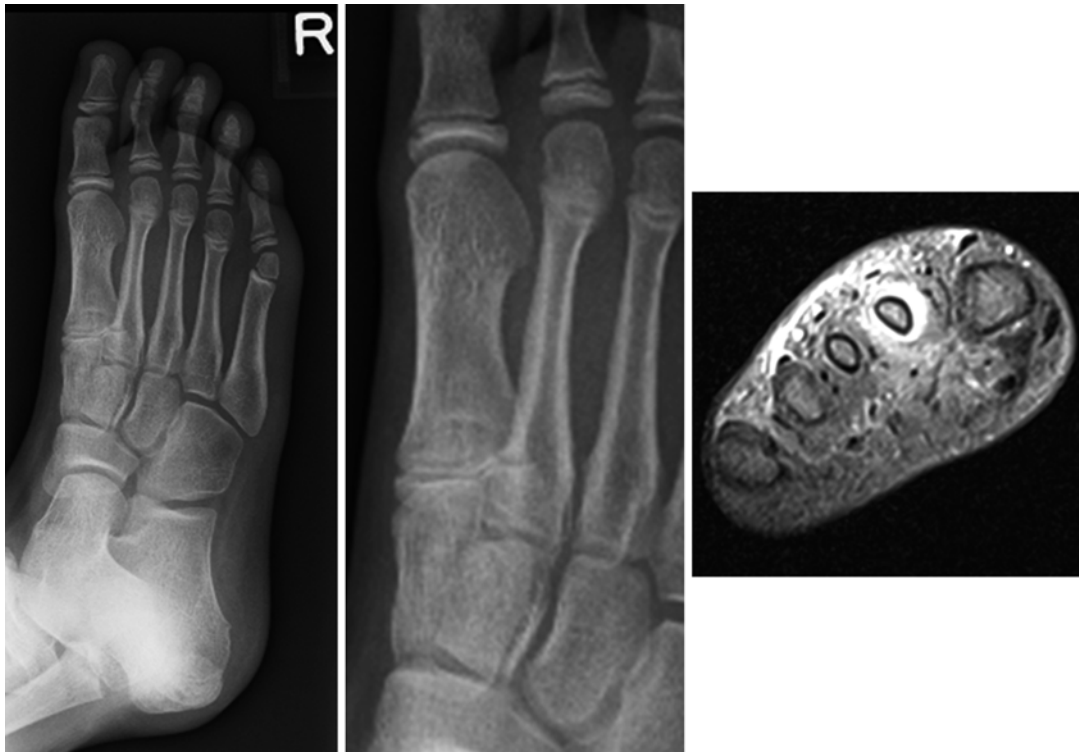


Fig. 38.5 Osteomyelitis of the shaft of the second metatarsal. There is a periosteal reaction over the shaft and increased signal on the MRI

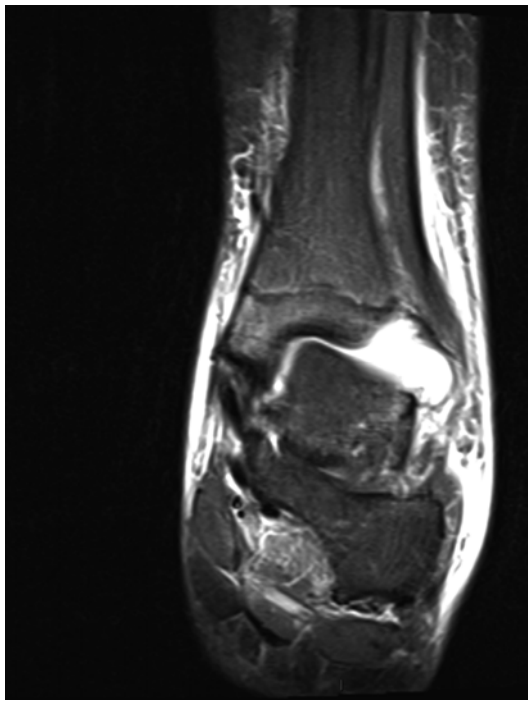


Fig. 38.6 MRI of septic arthritis of the ankle showing an effusion

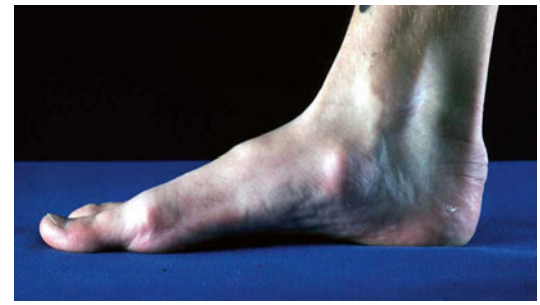


Fig. 38.7 Accessory navicular

38.5.5 Accessory Navicular

Swelling and tenderness may be present over the accessory navicular on the medial side of the foot (Fig. 38.7). There may also be tenderness over the tibialis posterior tendon which has an attachment to the navicular. The ossicle may be sufficiently large to cause rubbing against shoe wear.

38.5.6 Tarsal Coalition

The coalition may be bony, cartilaginous, or fibrous. Typically, when standing, the patient holds the foot flat and in valgus, and the medial arch does not appear when the patient stands on tiptoe. Subtalar movement is restricted and may be painful. The calcaneonavicular coalition is the commonest type and may have an "anteater's nose" appearance on a plain radiograph (Fig. 38.8a). CT is useful (Fig. 38.8b) for demonstrating coalitions which may also occur at the talocalcaneal, talonavicular, or naviculocuneiform joints.

38.5.7 Flatfeet

A nonstructural (physiologic) flatfoot is seen when the medial longitudinal arch appears when the patient stands on tiptoe and is typically bilateral. In the nonstructural type, the patient can be evaluated for joint laxity using the Beighton criteria (Fig. 38.9, Beighton et al. 1973).

Apparent flattening may be seen in patients who have genu valgum or external tibial torsion. Patients may also appear to have a flatfoot when there is shortening or spasticity of the gastrocnemius or gastrocnemius and soleus. The flatfoot is structural if the arch does not appear when the patient stands on tiptoe, and a unilateral flatfoot should be considered pathologic until proved otherwise. Causes include tarsal coalition (vide supra), overcorrected clubfoot, skewfoot, and structural ankle valgus, for example, seen in spina bifida.

38.5.8 Cavus

This can be considered to be an increased height of the medial longitudinal arch and may be physiologic (within a spectrum of normal variation), but the commonest cause is an underlying neurological condition (Fig. 38.10). Clinically it is often associated with claw toes and a varus heel (Fig. 38.11). Unilateral cavus should be assumed pathologic until proved otherwise. Pes cavus requires a neurological evaluation to exclude neuromuscular disease and spinal pathology (Weiner et al. 2013). A history of neuromuscular disease, for example, HMSN, should be sought. Calcaneocavus is seen typically in conditions where there is muscle weakness, e.g., polio, spina bifida, or after over-lengthening of the heel cord (Fig. 38.12). Cavovarus is typically seen in HMSN and hemiplegia (Vander Have et al. 2013).

38.5.9 Toe Pathology

Great Toe

Hallux valgus in the adolescent may be associated with generalized joint laxity where the adolescent has a broad forefoot, and this may be familial. Bunions are associated with metatarsus primus varus but can also be secondary to other pathology, for example, CP. Pes valgus in diplegic CP often results in a secondary bunion (Fig. 38.13). Shortening of the gastrocnemius or soleus results in escape valgus of the heel, and the forefoot is no longer aligned in the normal plane of progression and a bunion results. There is often an over- or underriding of the second toe.

A dorsal bunion is seen in conditions such as polio or spina bifida where there is a relative overaction of tibialis anterior and weakness of peroneus longus which results in an elevation of the first metatarsal and pain over its dorsum.

Interphalangeal valgus of great toe is also seen and gives the appearance of hallux valgus, but the abnormality lies at the interphalangeal joint and not the metatarsophalangeal joint.

Hallux rigidus occurs in adolescents and may be posttraumatic.

Lesser Toes

Rubbing of lesser toes against shoe wear can occur in children with a bunionette, overriding toes, hammer and claw toes, and overriding fifth toe. Flexibility of the deformity should be assessed and whether or not there is an underlying neurological cause.

38.5.10 Less Common Causes of Foot Pain

Benign and Malignant Tumors

These are seen in the foot but none is specific to the foot. Examples of benign tumors include

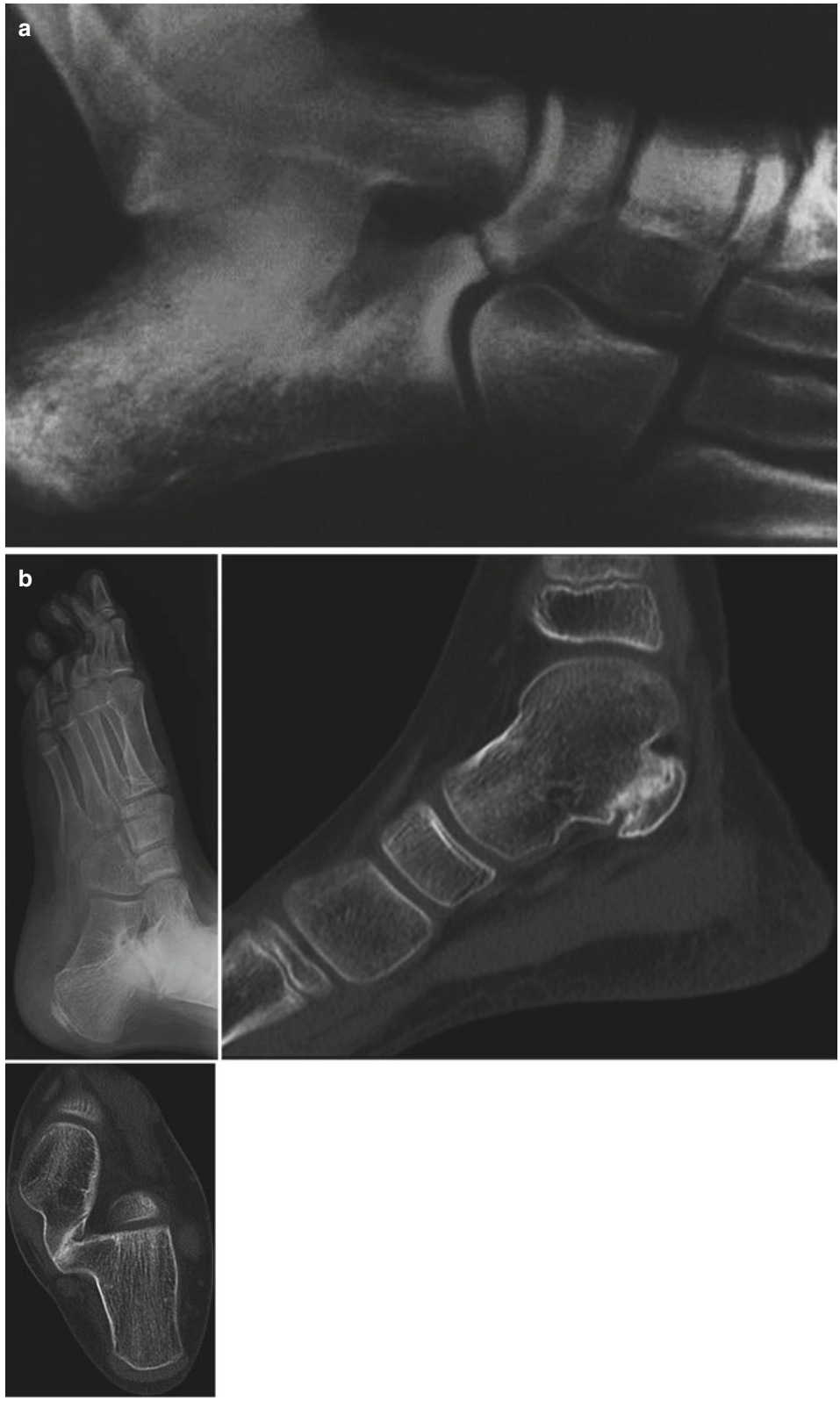
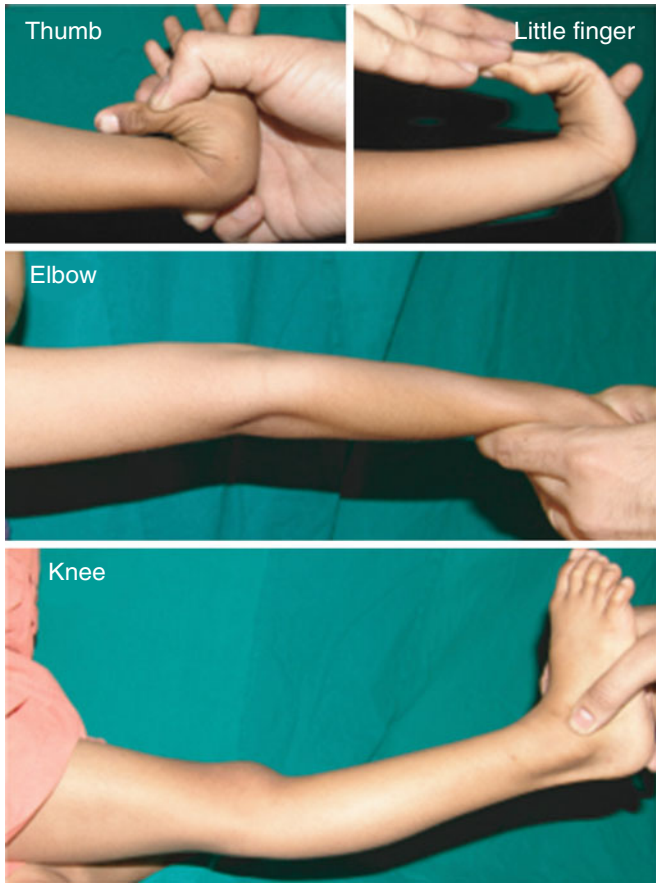


Fig. 38.8 Tarsal coalition. Anteater's nose appearance in a calcaneonavicular coalition (**a**) and CT demonstrating a talocalcaneal coalition (**b**)



BEIGHTON SCORE
Thumb can passively touch volar aspect of the forearm
Score: 1 for each thumb

Little finger passively hyper-extends till it is parallel to the dorsal surface of the forearm
Score: 1 for each little finger

Elbow hyperextends beyond neutral
Score: 1 for each elbow

Knee hyperextends beyond neutral
Score: 1 for each knee

Add 1 to score if child can place both hands on the floor without bending knees

Total Score of 4 or more (out of a maximum of 9) is regarded as generalised hypermobility

Fig. 38.9 The Beighton score

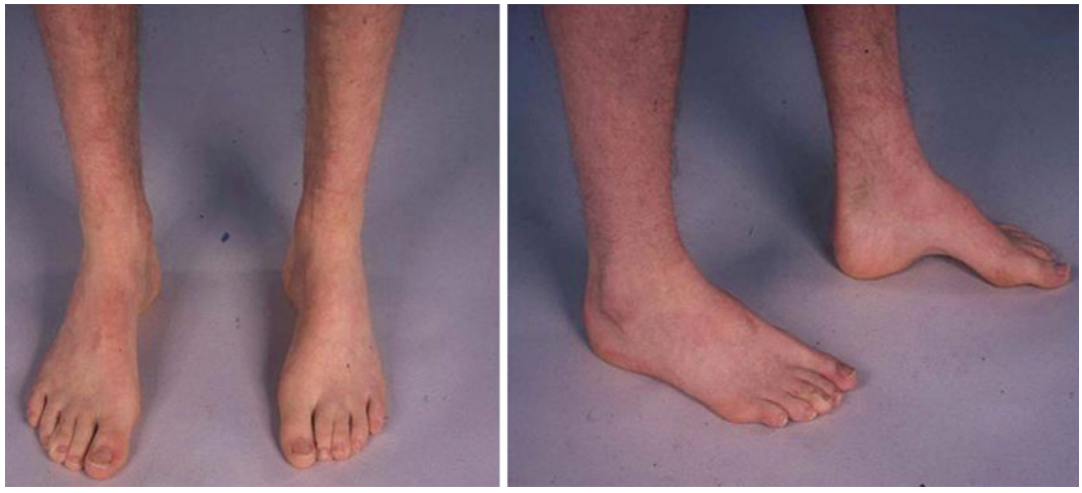


Fig. 38.10 Pes cavus in mild HMSN without clawing of the toes

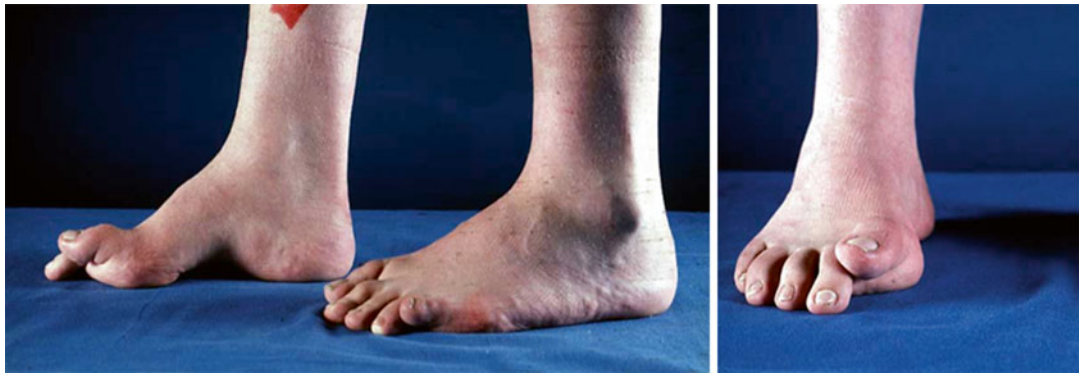


Fig. 38.11 Pes cavus right foot; the great toe deformity was fixed

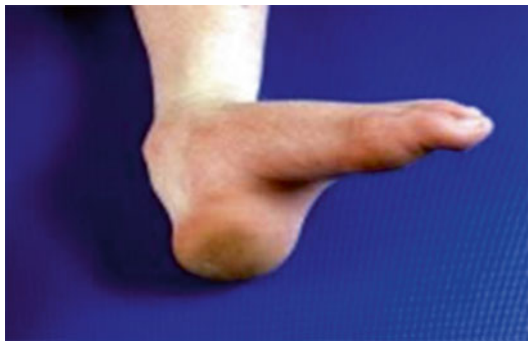


Fig. 38.12 Calcaneocavus in spina bifida

subungual exostosis, osteochondromas, aneurysmal bone cyst (often in the calcaneus), osteoid osteoma, fibrous dysplasia, and neurofibromatosis (Fig. 38.14). Malignant tumors are less common in the foot than in long bones, and in this age group, Ewing's sarcoma and osteosarcoma are the commonest type.

Freiberg's Infraction

Freiberg's infraction is an idiopathic avascular necrosis of the head of a metatarsal, usually the second metatarsal (Fig. 38.15).

Plantar Fasciitis

There may be tenderness over the origin of the plantar fascia, and the condition is seen in young athletes.

Tarsal Tunnel Syndrome

This is rare and the patient may walk holding the foot inverted, and there may be a positive Tinel's sign over the tarsal tunnel and foot pain.

Calcaneal Apophysitis (Sever's Disease)

This is usually seen in the juvenile age group and is an overuse phenomenon typically occurring after activity and relieved by rest. Adolescents may also complain of pain over the calcaneal apophysis because the posterior calcaneus unites with the body of the calcaneus at about 14 years in females and at about 16 years in males.

Complex Regional Pain Syndrome Type 1

This may result from previous trauma, and the patient complains of exquisite local tenderness and changes in skin color and temperature.

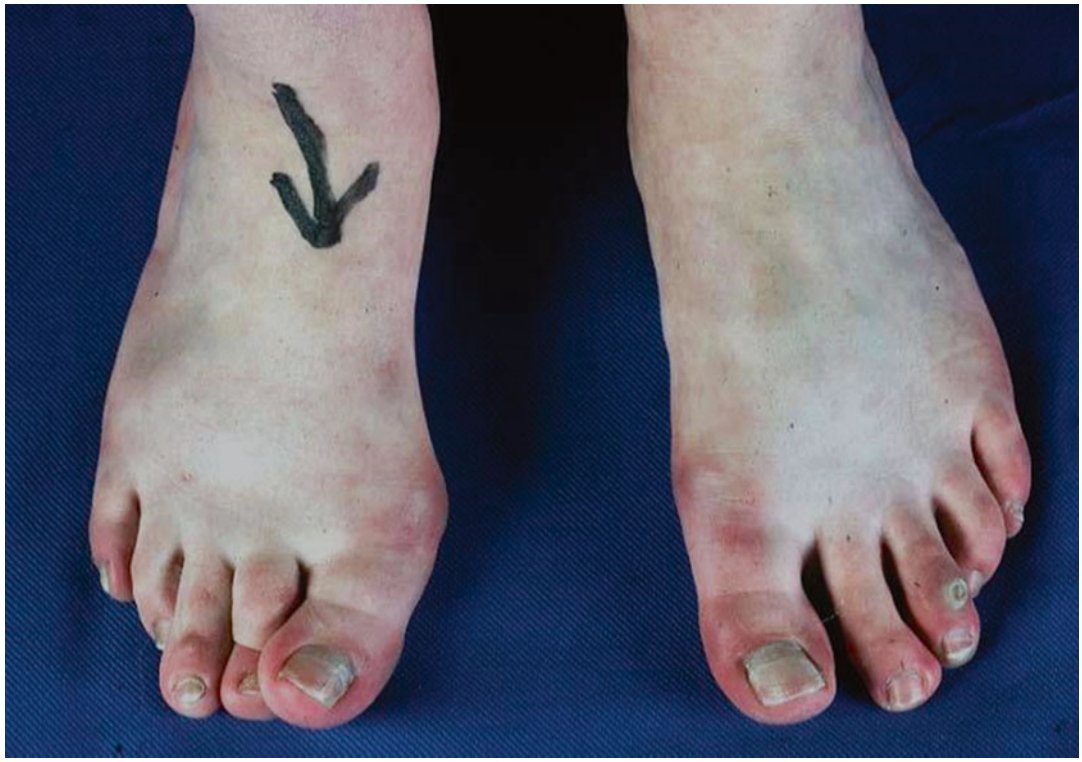


Fig. 38.13 Acquired adolescent bunion in diplegic CP; the patient had a valgus hindfoot

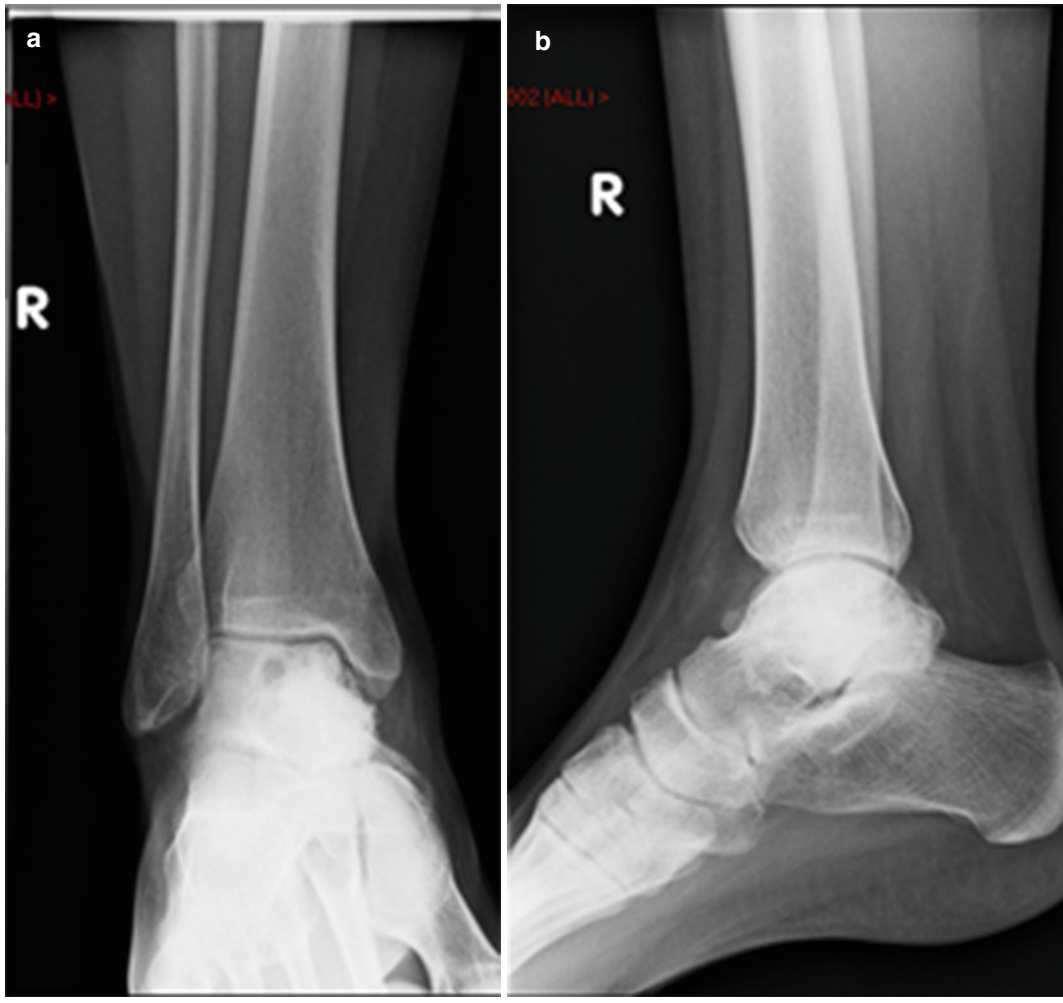


Fig. 38.14 Chondroblastoma of the talus in a 16-year-old girl

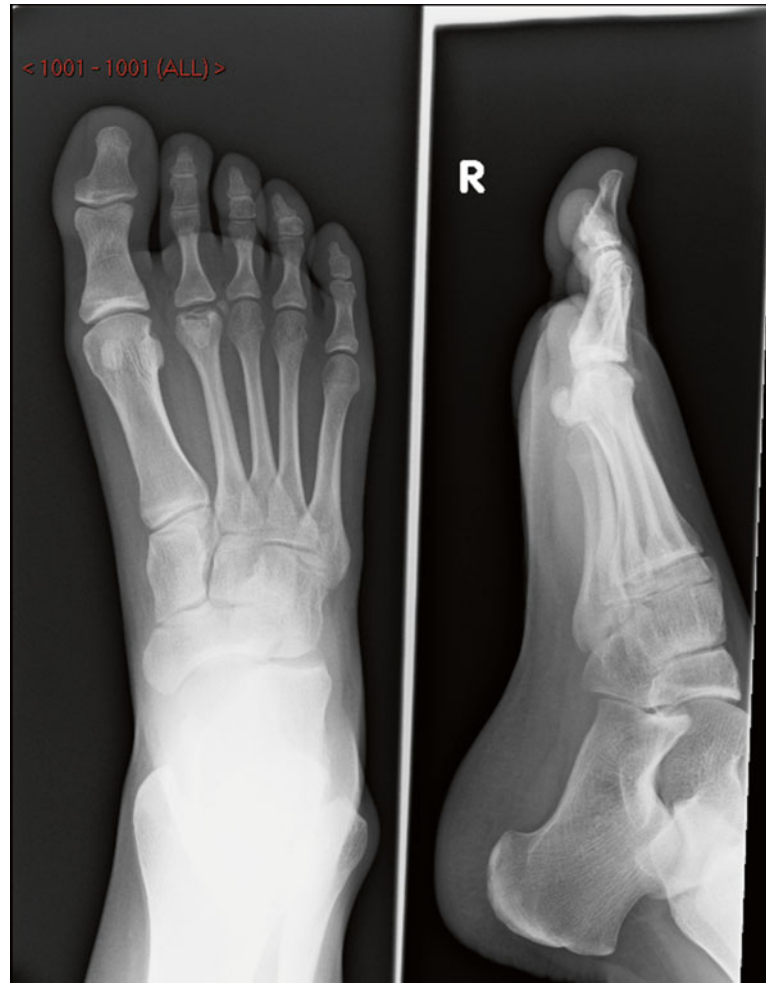


Fig. 38.15 Freiberg's infraction of the head of the second metatarsal in a skeletally mature adolescent

38.6 Establishing the Diagnosis

An outline of an approach to the diagnosis of foot pain in the adolescent is shown in Table 38.2.

Pain of acute onset associated with trauma or fever has been excluded.

Table 38.2 Establishing the diagnosis of the cause of pain in the foot in an adolescent

History				
Pain related to footwear	Pain not related to footwear	Pain not related to footwear	Pain not related to footwear	Pain may be related to footwear (pain may be aggravated with high-heel footwear)
Pain not related to movement of the foot	Pain aggravated on walking up or down a slope	Pain aggravated on walking on uneven ground	Pain may be aggravated by walking on uneven ground	Pain on walking not affected by surface or slope
Site of pain: Over deformed toe or over bony prominence	Site of pain: Ankle and hindfoot	Site of pain: Hindfoot	Site of pain: Midfoot	Site of pain: Forefoot
Physical examination				
Bony prominence present at site of pain	No bony prominence at site of pain	No bony prominence at site of pain	No bony prominence at site of pain	No bony prominence at site of pain
Medial longitudinal arch normal	Medial longitudinal arch normal	Medial longitudinal arch reduced	Medial longitudinal arch may be normal or reduced	Medial longitudinal arch may be normal or accentuated
Foot deformity: Deformities of toes may be present	Foot deformity: No deformity	Foot deformity: Hindfoot valgus	Foot deformity: Hindfoot valgus may be present	Foot deformity: Equinus may be present Cavus may be present Hallux valgus may be present Foot may not be deformed
Normal passive range of motion of the joints of the foot	Range of ankle motion reduced	Range of subtalar motion reduced	Range of midtarsal motion reduced (subtalar motion may also be reduced)	Range of passive dorsiflexion of ankle may be reduced Active dorsiflexion may be weak or absent Movement of the metatarsophalangeal joint of the great toe or second toe may be reduced or painful
No characteristic alteration in gait pattern	Absent rockers during the stance phase of gait	Rockers present during the stance phase	Rockers present during the stance phase	Rockers absent if there is an equinus deformity
Working diagnosis: Localized pressure from footwear	Working diagnosis: Ankle pathology	Working diagnosis: Subtalar joint pathology	Working diagnosis: Midtarsal or midfoot pathology	Working diagnosis: Forefoot pathology

(continued)

Table 38.2 (continued)

Investigations				
Radiograph not indicated	Plain radiograph may show: Avascular necrosis of body of talus OCD of talus Lytic lesion in talus Narrowing of ankle joint space	Plain radiograph may show: Talocalcaneal coalition	Plain radiograph may show: Accessory navicular	Plain radiograph may show: Freiberg's infraction Hallux valgus
Diagnosis				
Pressure of footwear on bony prominence	Ankle pathology: AVN talus OCD talus Lesion in talus Ankle arthritis	Subtalar pathology: Tarsal coalition Subtalar arthritis	Midfoot pathology: Accessory navicular Midtarsal arthritis	Forefoot pathology: Hallux valgus Hallux rigidus Freiberg's infraction Metatarsalgia

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Further Reading

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