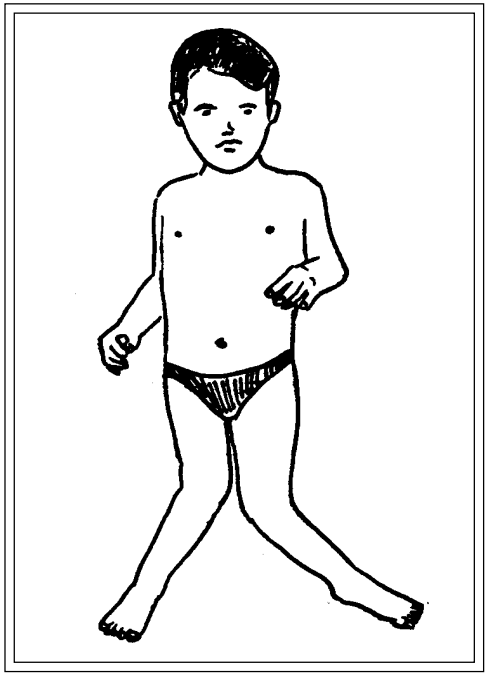
Chapter 7

Congenital and Paediatric Conditions

Generalised abnormalities
Upper limb
Spine
Lower limb



Classification

Generalised abnormalities

- Achondroplasia
- Osteogenesis imperfecta
- Arthrogryposis
- Still's disease
- Hurler's syndrome
- Diaphyseal aclasis
- Hypothyroidism

Upper limb

- Limb deficiences
- Macromelia and macrodactyly
- Trigger fingers
- Syndactyly
- Lobster claw hand
- Absent
- Extra digits

Trunk

- Spina bifida and meningomyelocele
- Scoliosis
- Kyphos and kyphosis

Lower limb

Whole Limb
- Phocomelia
- Macrodactyly

Hip
- Congenital dislocation (CDH)
- Slipped epiphysis
- Perthes' disease
- Transient synovitis
- Septic arthritis
- Coxa vara and valga
- Protrusio acetabuli

Knee
- Genu varum and valgum
- Genu recurvatum
- Osteochondritis
- Congenital webbing

Ankle and foot
- Talipes equino varus
- Talipes calcaneo valgus
- Pes cavus
- Pes planus
- Metatarsus adductus
- Osteochondritis or avascular necrosis
- Exostoses
- Accessory bones
- Syndactyly

Introduction

Diagnosis

A clinical history is essential and can reveal a genetic abnormality which may be autosomal dominant or recessive or sex-linked. Such deformities include diaphyseal aclasis or multiple osteochondromata and achondroplasia with dwarfism. There may be a history of maternal use of drugs such as thalidomide, irradiation of the foetus in the critical first three months of development or a history of maternal rubella infection. Finally there is a large group with no known cause.

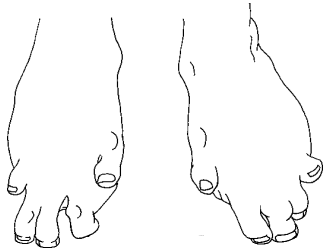
Diagnosis may be easy with evidence of a classical anomaly at birth such as differences in the size or shape of limbs or multiple abnormalities which may be symmetrical, such as microdactyly or talipes equino varus. The opposite limb must always be examined. It may show a similar deformity, which is usually painless, with no evidence that the abnormality is acquired such as operation scars. All gradations of spina bifida including a meningomyelocele must also be looked for.

Besides skeletal abnormalities, there may be other congenital abnormalities and thus a full examination should always be carried out. Other family members may show similar abnormalities or give a classic history which in most cases simplifies the diagnosis.

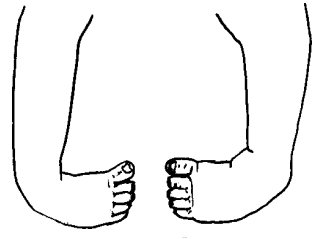
Relatively common non-orthopaedic abnormalities include: cleft lip and palate, Down's syndrome, cardiac abnormalities such as tetralogy of Fallot (which produces a boot shaped heart) and abdominal visceral abnormalities such as pyloric stenosis.

Diagnosis

Orthopaedic anomalies

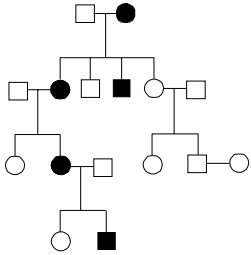


Microdactyly

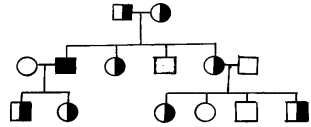


Talipes equino varus

Family history

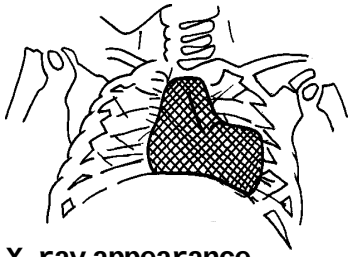


Autosomal dominant disorder

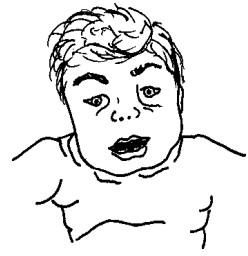


Autosomal recessive disorder

Non-orthopaedic anomalies



X-ray appearance of a 'boot heart'



Facial features — Hurler's syndrome

Generalised Abnormalities

Achondroplasia

Achondroplasia is a congenital condition with an autosomal dominant pattern of inheritance, but approximately 80% of cases arise from a new gene mutation. The main disability is due to a failure of normal ossification of the long bones which are consequently much shorter than normal. The trunk, however, is little affected although spinal stenosis, thoracic kyphosis and an excessive lumbar lordosis are commonly found. This may produce severe neurological sequelae such as spinal cord compression and even quadriplegia.

The hands are broad, quite divergent, and the middle three fingers of equal length.

The head is slightly larger than normal with a depressed nasal bridge and bulging forehead. There is, however, no mental impairment and many achondroplastic dwarfs find gainful employment. Achondroplastic dwarfs are seldom taller than 125 cm.

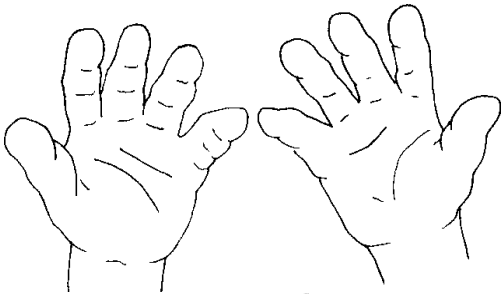
Treatment

The complications of achondroplasia include degenerative joint disease, especially osteoarthritis of the hips, which may require joint replacement. Nerve compression and

Generalised Abnormalities

Achondroplasia

Achondroplastic dwarf – may occasionally require crutches as paresis or paralysis may result from spinal stenosis



Hands of an achrondroplastic dwarf

paralysis due to the spinal stenosis may require laminectomy and decompression occasionally extending for the entire length of the spine.

Osteogenesis imperfecta

(Fragilitas ossium)

Osteogenesis imperfecta is usually inherited as an autosomal dominant trait. As a result of defective collagen synthesis, the bones are abnormally brittle and multiple fractures are common.

Other collagen-containing tissues such as the tendons, ligaments, skin, teeth and sclerae of the eyes may also be affected.

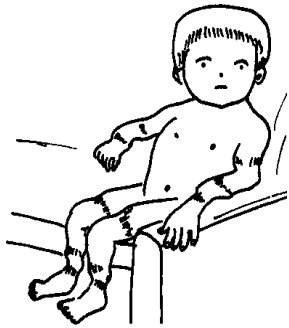
All gradations occur, from multiple fractures at birth to less severe forms where the child does not develop fractures until later in life. The sclerae, especially in the late-manifesting or 'tarda' cases, may be blue due to lack of opaque collagen with resulting translucency to the choroid. There may also be deafness due to otosclerosis and ligamentous laxity in the chain of ossicles.

As a result of these often multiple fractures, the limbs and trunk may be deformed and shortened. Fractures should be treated by the standard methods and usually heal satisfactorily. The remaining multiple deformities, however, especially of the femur, may require internal fixation including intramedullary nailing, to straighten them.

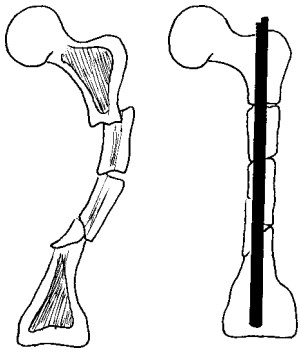
In severe cases calipers, spinal braces and other supports may be necessary to protect the brittle bones.

Generalised Abnormalities

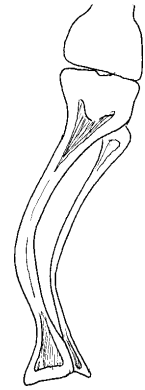
Osteogenesis Imperfecta



Child with osteogenous imperfecta showing limbs with multiple deformities



X-ray appearance of a deformed femur with multiple fractures



X-ray appearance of a deformed tibia

Arthrogryposis
This is due to collagen replacement of muscles surrounding joints and there are often multiple flexion contractures with thickened joint capsules and severe deformities which may lead to dislocation of the joints and very severe deformities.

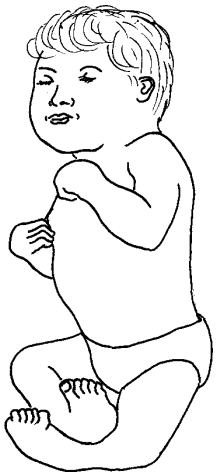
Still's disease
This is rheumatoid arthritis occurring in childhood and mainly affecting the peripheral joints. There are often systemic manifestations with an enlarged spleen and liver together with growth disturbances due to epiphyseal damage. The condition is discussed in Chapter 10.

Hurler's syndrome
This is a congenital condition with multiple deformities, mental disturbance and symmetrical dwarfism with limbs and trunk equally affected. It is due to a rare mucopolysaccharide disorder.

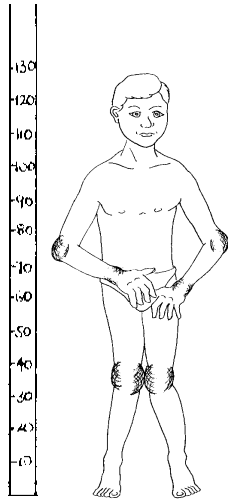
Diaphyseal aclasis
Multiple osteochondromata occur and these may occasionally develop into chondrosarcomata. This is discussed further in Chapter 8.

Other tumours
These are discussed in Chapter 8.

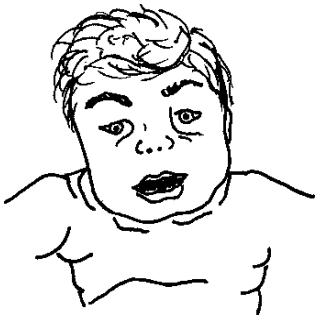
Generalised Abnormalities



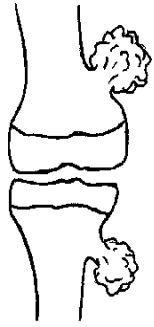
Arthrogryposis multiplex congenita



Still's disease



Hurler's syndrome



X-ray appearance of diaphyseal aclasis

Upper Limb Conditions

Limb deficiencies

Deficiencies of the limbs may vary from complete absence (amelia) to partial absence (phocomelia). Every gradation may occur and only one limb or all four limbs may be affected, or even absent.

There may be absence of one or more bones on the medial and lateral aspects of the arm or leg and this may lead to deformities such as a Madelung's deformity (radial deviation of both hands) due to a short or absent radius or varus of both feet due to absent tibia. Often the digits on the side of the absent long bone are also deficient or absent.

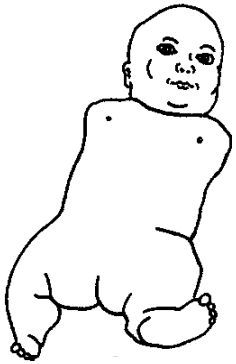
The cause of limb differences is usually drugs such as thalidomide in the first trimester of pregnancy. Other causes include irradiation, rubella, true genetic abnormalities or unknown aetiological factors.

Macromelia and macrodactyly

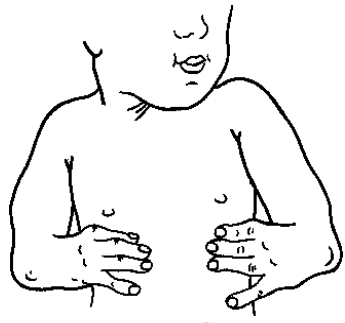
Overgrowth of the limb may also occur. It may be due to a true genetic abnormality when individual digits may have overgrown (usually three digits), or may involve the whole limb. In the latter case, congenital lymphangiectasis may be responsible.

Upper Limb Conditions

Drugs and irradiation



Amelia



Madelung's deformity

Idiopathic conditions

Macrodactyly

Macromelia

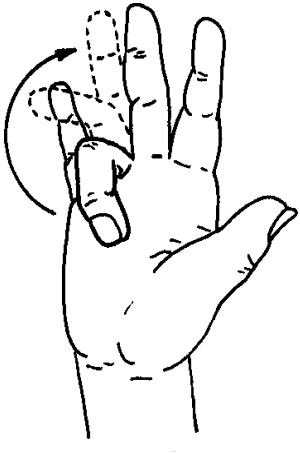
Trigger thumb and little finger
A contracture of the fibrous flexor sheath of the thumb or little finger may occur. This is usually symmetrical and affects either both thumbs or both little fingers. The digits may be held in fixed flexion or they may permit 'triggering' or 'snapping' into extension due to a secondary nodule on the tendon suddenly being released from the constricted flexor sheath like a tight cork in a bottle.

Syndactyly
Syndactyly is fusion of one or more digits. It may affect all the digits together, or, more commonly, pairs of digits such as index and middle finger together or with the ring and little finger fused. The limbs are usually, but not always, symmetrically affected.

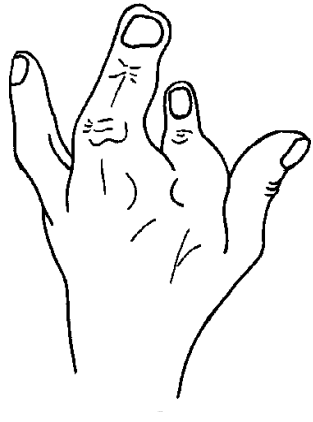
Lobster claw hand
This is different in that there is lack of fusion with usually only two digits creating a claw-like deformity as illustrated.

Absent or extra digits
Digits may be absent or an extra digit may be present as a small appendage. In the latter case amputation is usually indicated.

Upper Limb Conditions

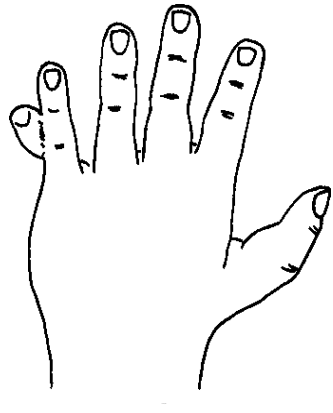


Trigger finger



Syndactyly

Lobster claw hand



Extra digit

Other developmental upper limb deformities

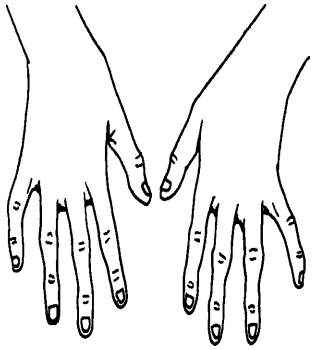
These are discussed elsewhere and may be part of a generalised developmental abnormality. They include the short upper limbs and spade-like 'chubby' fingers in achondroplasia, the long spider-like fingers with loose joints in Marfan's syndrome, as well as cranioclеidodysostosis, a condition where there is absence of membrane bones of the skull and clavicles. Also included are multiple osteochondromata of diaphyseal aclasis, and various types of bony fusion such as a synostosis between radius and ulna, or between the ulna and the humerus.

General treatment

The aim, in treating these deformities, should be to maintain function rather than achieve a cosmetic result. Many patients, despite the deformities, have surprisingly good function and this must not be destroyed just to improve the appearance.

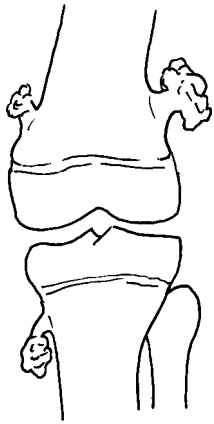
Artificial limbs specially tailored to the individual deformity have a real place, but many patients do not use these prostheses unless they are fitted in early childhood.

Treatment



Arachnodactyly

Craniocleidodysostosis



X-ray appearance of multiple osteochondromata

X-ray appearance of radio-ulnar synostosis

Spinal Conditions

These are discussed in more detail in Chapter 11. The four most significant deformities, however, are:

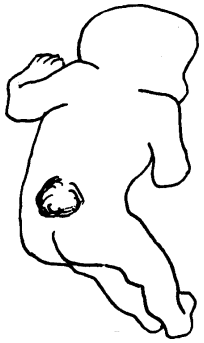
Spina bifida and meningomyelocele
This is a defect usually in the lower lumbar spine which may vary from being a small asymptomatic malfusion of the posterior parts of the vertebrae to a complete protrusion of the cord and the nerve roots, as in a meningomyelocele. In the latter case paralysis of the bladder and lower limbs and an associated hydrocephalus is common.

Scoliosis
This is a lateral curvature of the spine and its causes vary from a congenital hemivertebra to paralysis, as occurs in conditions such as poliomyelitis, but in many cases it is of unknown aetiology.

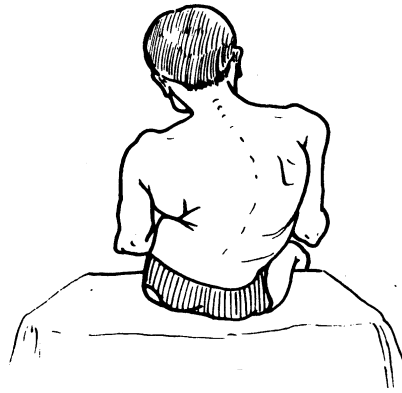
Unequal limb lengths will produce a scoliosis which is compensated for when the patient sits down or bends forward.

Kyphosis and kyphos
A kyphosis is a smooth forward curve of the spine, while a kyphos is an abrupt curve. A kyphosis may be due to Scheuermann's disease (an osteochondritis of the intervertebral disc spaces which mainly affects the thoracic vertebrae) or to paralysis. A kyphos is usually due to a fracture, infection or a secondary tumour of the spine.

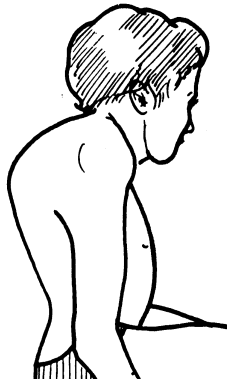
Spinal Conditions



Spina bifida



Scoliosis



Kyphosis

Kyphoscoliosis

Lower Limb Conditions

Hip deformities

Congenital dislocation of the hip

Congenital dislocation of the hip probably has a combined genetic and hormonal cause. It is much more common in some countries, such as Japan and Italy, and rare in others such as Africa. It is six times more common in girls than in boys and one third of cases are bilateral. The bilateral cases are probably mainly genetically determined.

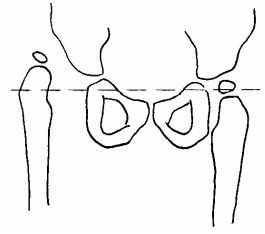
The main defects which appear in untreated cases are an upwardly dislocated hip, a poorly formed, sloping acetabulum (instead of the normal 60^0 shelf) and an ante-verted and poorly formed neck and head of femur (greater than the usual 35^0 anteversion).

Every infant must be examined for dislocation or subluxation at birth. If CDH is present there will be limitation of abduction in flexion, usually with the ability to reduce the hip with a click (Ortolani's and Barlow's manoeuvre – see illustration). Telescoping of the hip, shortening of the thigh and increased folds in the thigh will be seen when the hip is dislocated. X-rays and ultrasound should be used to confirm the diagnosis and arthrography may be necessary.

Treatment varies from abduction pillows and a bulky nappy to keep the legs abducted in the first three months of life, to abduction splints and plaster. These must not over stress the hips lest avascular necrosis of the head of the femur occur. If the hips are completely stable at six months, careful observation is all that is necessary in most cases.

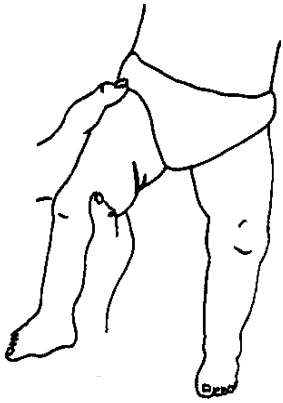
Lower Limb Conditions

Congenital dislocation of the hip



Asymmetrical skin folds

X-ray appearance of CDH



Limited hip abduction – Ortolani's test

Hip telescoping

In many cases, however, an infolding 'limbus' of capsule prevents concentric reduction, necessitating open reduction if splinting fails. A persistent anteversion of the head and neck of the femur may require external rotation osteotomy of the upper femur with plate fixation. Open reduction of the hip and excision of the limbus is sometimes necessary.

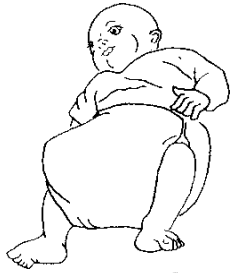
Between the ages of six months and six years, if these methods fail and the head is incompletely covered by an adequate acetabulum, various methods may be used to improve joint function and stability. These include innominate (Salter) osteotomy, or various other methods to increase the stability of the acetabulum including fashioning a shelf of bone or displacement of the ilium above the acetabulum. Up to the age of twelve years open reduction should also be considered but above this age the alternatives are no treatment, an osteotomy of the upper femur to give stability (Schantz) or in adult life a cementless total hip replacement.

Slipped capital femoral epiphysis

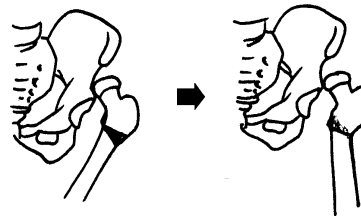
Slipping of the femoral epiphysis is usually seen between the ages of 10 and 15. This is caused by an imbalance between sex and growth hormones in the adolescent and may be precipitated by minor trauma. Early and very gentle reduction of the slipped epiphysis, together with early pinning of the hip, is the treatment of choice in most cases. Prophylactic pinning of the opposite side is also often required.

Lower Limb Conditions

Congenital dislocation of the hip

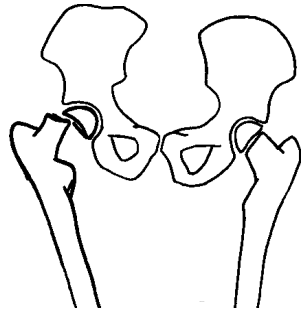


Abduction pillow



Schantz osteotomy

Slipped epiphysis



X-ray appearance of a slipped epiphysis

Perthes' disease

Perthes' disease is an osteochondritis of the femoral capital epiphysis and is due to disturbance of the blood supply to the femoral head. Children usually present between the ages of 5 and 10. The onset tends to be gradual and the patient complains of mild pain in the hip and walks with a limp. There is no constitutional upset, blood investigations are normal and examination of the hip shows slight limitation of all movements.

The cause is unknown but may be due to trauma at a critical stage of growth. A similar condition occurs in sickle cell anaemia.

Diagnosis is confirmed on X-ray which may not show any abnormalities in the earliest stages. Initially there is flattening and decrease in the depth of the epiphysis which becomes denser and fragmented. Nuclear scanning will show deficient uptake in the ossific nucleus of the head.

The head gradually revascularises over a period of two years but the head and neck usually remain permanently flattened and deformed in severe cases.

Treatment should aim to contain the softened femoral head in the acetabulum and prevent full weight-bearing in the early stages. Initially the child should be admitted to hospital and treated with skin traction in abduction. As soon as the pain has settled this is replaced by an abduction walking frame with gradually increasing weight-bearing.

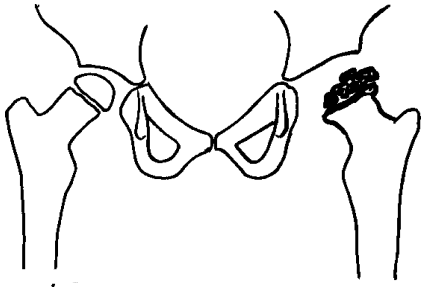
Severe cases are later complicated by osteoarthritis and may require total hip replacement.

Transient synovitis

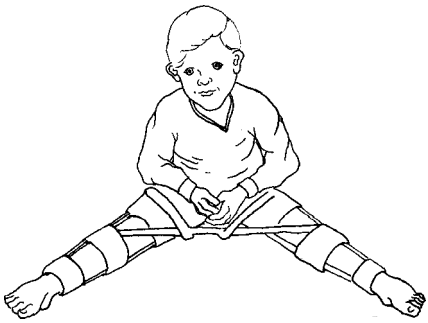
A transient synovitis of the hip is not uncommon in the first 10 years of life. The patient complains of severe hip pain with the hip flexed, initially abducted then

Lower Limb Conditions

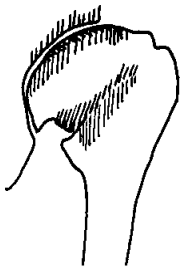
Perthes' Disease



X-ray appearance of unilateral Perthes' disease



Abduction splint



X-ray appearance of old Perthes' disease

adducted and *externally* rotated. X-rays and white cell count are initially normal with a normal or slightly raised ESR. The patient should be treated with Russell traction and bed rest as the differential diagnosis is a low grade infection such as tuberculosis or Perthes' disease.

Septic arthritis

An infective arthritis in childhood is a surgical emergency as it can lead to destruction of the head of the femur and osteomyelitis of the upper femoral shaft. In the first year of life it is called Tom Smith's disease.

The ESR and WBC count are raised, the child is usually pyrexic, ill and in severe pain. X-rays are normal in the early stages and joint aspiration with culture and drainage may be urgently required, together with intravenous antibiotics after blood for culture has been taken.

Coxa vara and valga and protrusio acetabuli

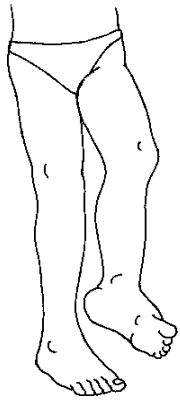
Coxa vara and valga refer to a decrease and increase respectively in the angle between the head and neck of the femur and its shaft. Protrusio acetabuli is an extension of the acetabular fossa into the true pelvis, which limits movement at the hip joint.

A mild degree of coxa vara and valga is common. A severe degree may be genetically determined and is usually bilateral. Asymmetrical coxa valga is often assoc-iated with a paralysed hip such as in poliomyelitis and spina bifida. Other causes include fractures of the neck of the femur.

A protrusio acetabuli, when the hip is deep in the acetabulum, may lead to later osteoarthritis, as may a very shallow acetabulum.

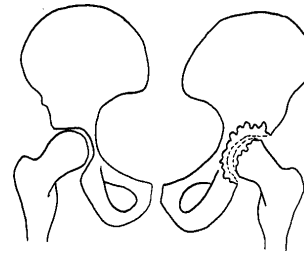
Hip Conditions

Transient synovitis



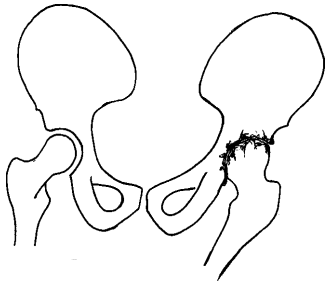
Flexed externally rotated and abducted

Infective arthritis



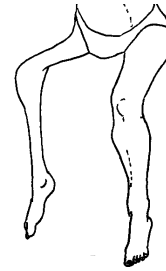
X-ray appearance of the destruction of the acetabulum and femoral head

Coxa vara



X-ray appearance of steoarthritis

Coxa valga



Associated with paralysis

Knee conditions

Genu varum and genu valgum

Knock knees and bow legs are common in childhood and are often familial. If unassociated with poliomyelitis, injury or bone disease, they usually improve without treatment after the age of 3. The maximum acceptable separation of the medial malleoli is 7.5cm (3 inches) at 3 years. Occasionally corrective osteotomy is required at the age of 12–14 or stapling epiphysiodesis of the tibial and femoral epiphyses at about 10 years to stop epiphyseal growth on the contralateral side.

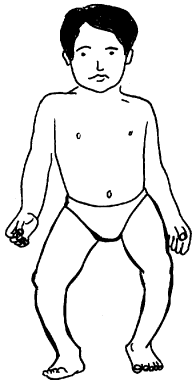
Congenital genu recurvatum

This is usually due to increased intra-amniotic pressure and excessive oestrogens at the time of birth. Immobilisation in a *padded* plaster, in as much flexion as possible for three weeks will usually effect a cure. Occasionally this condition is associated with arthrogryposis or with fibrotic and tight quadriceps and these conditions are difficult to treat and will usually require operation.

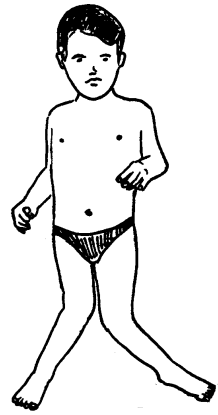
Osteochondritis

A painful knee, particularly between the ages of 10–15 years may be due to an osteochondritis of the femoral condyles and is classically seen on the lateral side of the medial femoral condyle. There is often a softened circular segment of cartilage which may become detached, together with its underlying bone, to form a loose body in the knee joint. If rest and support fail to secure its attachment and revascularisation at an early stage of the condition, operation with drilling or pinning may be required and occasionally excision of a loose fragment.

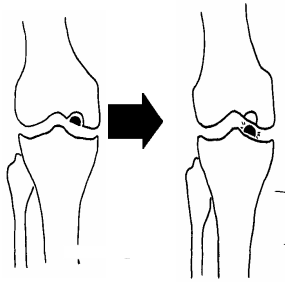
Knee Conditions



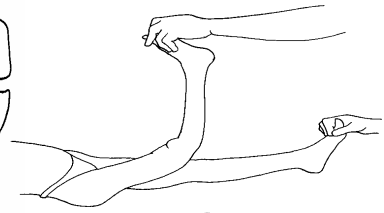
Genu varum
('bow legs')



Genu valgum
('knock
knees')



X-ray
appearance of
osteochondritis



Genu recurvatum

Ankle and foot conditions

Talipes equino varus

This is usually a congenital deformity present at birth, sometimes with a family history and may be unilateral or bilateral. The foot is pointed downwards and inwards, and has normal sensation and initially normal power.

Occasionally it is associated with spina bifida when both sensation and power may be diminished, and the spine must be inspected in all cases. Other neurological conditions, such as poliomyelitis and arthrogryposis, may also cause a similar deformity.

Treatment should be started as soon as possible with passive stretching and strapping, followed by plaster of Paris or splints after manipulation. Subsequent treatment may necessitate soft tissue correction and later a bony operation.

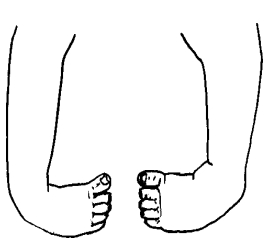
Talipes calcaneo valgus

This is the opposite deformity to equino varus and the foot is dorsiflexed and everted. It is usually caused by intrauterine pressure on the foetus and most cases are easily corrected by passive stretching, strapping and plaster. Occasionally there is a true genetic abnormality due to a congenital vertical talus or spina bifida and this is more difficult to treat. Poliomyelitis may cause a calcaneo valgus deformity due to muscle imbalance.

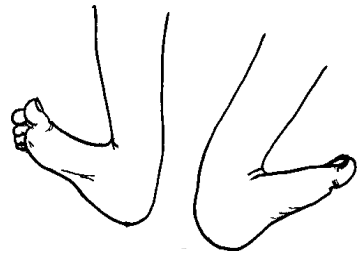
Pes cavus

This is a clawing of the longitudinal arch of the foot often associated with clawing of the toes. Mild cases are sometimes idiopathic and familial. Other cases include neurological conditions such as spina bifida, peroneal muscular atrophy, Friedreich's ataxia and poliomyelitis, as may vascular insufficiency.

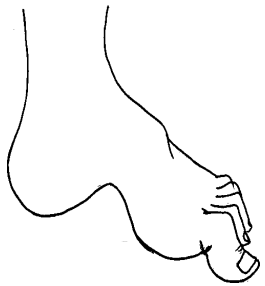
Ankle and Foot Conditions



Talipes equino varus



Talipes calcaneo valgus



Pes cavus ('clawfoot')

Pes planus ('flatfoot')

In severe cases callosities may develop under the forefoot and toes and may necessitate special footwear and occasionally soft tissue or bony correction.

Pes Planus

Flat feet is a common condition with the medial border of the foot in contact with the ground and the foot everted. There is often a family history. It may also be associated with a short tendo calcaneus with the valgus flat foot compensating for limitation of dorsiflexion.

The foot is usually mobile and the arch is restored when the patient stands on the toes. It may occasionally be rigid, and the cause may be a congenital calcaneo-navicular or other subtaloid bony bar leading to peroneal spasm and a spastic flat foot.

Mobile flat feet seldom require treatment except for an occasional small raise on the inner side of the heel or an arch support. Spastic flat feet sometimes require a subtaloid arthrodesis.

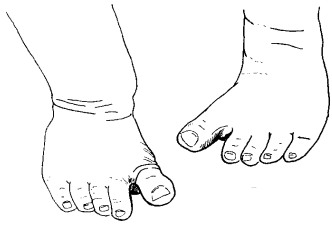
Metatarsus Adductus

This is a congenital deformity with the forefoot adducted and the child walking with an intoeing gait. It may be limited to the first metatarsal (metatarsus primus varus) in which case the big toe may be pivoted laterally (hallux valgus). Most cases do not require treatment apart from appropriate footwear, passive stretching, and sometimes a small raise on the outer side of the shoe.

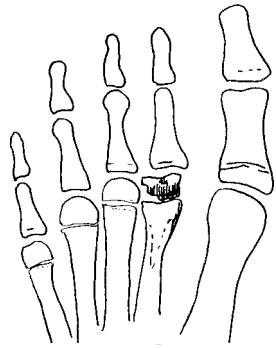
Osteochrondritis or Avascular Necrosis

This may affect the navicular (Köhlers disease) or head of the second metatarsal (Freiberg's disease) and both are probably due to trauma, with interruption of the blood supply resulting in avascular changes. Gradual revascularisation, with residual deformity and little disability usually oc-

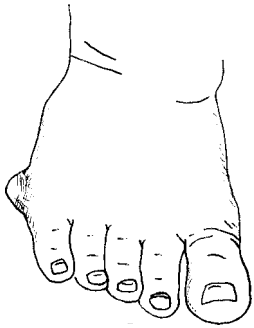
Ankle and Foot Conditions



Metatarsus adductus



X-ray appearance of Freiberg's disease – osteochondritis of second metatarsal head



Exostosis fifth metatarsal head

Syndactyly and overiding fifth toe

curs with conservative treatment with rest and perhaps supports.

Exostoses
These are often associated with an overlying bursa and may be due to irritation by footwear. They include the back of the calcaneus, the base or head of the 5th metatarsal (bunionette), and dorsum of the metatarsals in cavus feet. The most common site is the medial side of the 1st metatarsal head associated with a hallux valgus.

Accessory bones
These are due to a congenital deformity and are usually asymptomatic. They include the os trigonum (behind the talus) and os tibialis externus (the medial side of the navicular). They do not require treatment but are sometimes confused with an old fracture.

Syndactyly
This may be variable in extent and implies partial fusion of the web of one or more toes. It is often familial and seldom requires treatment.

Other toe deformities
The 5th toe may override or underride the 4th toe. Occasionally the other toes are clawed in the proximal or distal interphalangeal joint. In the latter case there may be an associated neurological abnormality (see above). Severe deformities occasionally require operative correction, but most cases can be treated by appropriate padding and shoes.