NOTES
MUSCULOSKELETAL CONGENITAL MALFORMATIONS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Soft tissue structure/bone growth/ development errors
- Present at birth (often)
- Isolated, or + associated anomalies

SIGNS & SYMPTOMS

- Disease-dependent malformations

DIAGNOSIS

DIAGNOSTIC IMAGING

E.g. X-ray, CT scan

OTHER DIAGNOSTICS

- Clinical evaluation

TREATMENT

SURGERY

- See individual disorders

OTHER INTERVENTIONS

- May resolve with age
- Conservative treatment (e.g. occupational therapy, splinting)

ARTHROGRYPOSIS

osms.it/arthrogryposis

PATHOLOGY & CAUSES

- Rare, non-progressive congenital disorder
  - Multiple joint contractures
  - AKA arthrogryposis multiplex congenita
- Decreased fetus movement in utero → fibrous connective, adipose tissue replaces muscle tissue → muscle shortening → joints fixed → affected joints unable to extend, flex
- Potential associated syndrome/disease (e.g. pulmonary hypoplasia, cryptorchidism, intestinal atresia, gastroschisis)
- Intelligence typically normal

TYPES

Amyoplasia

- Most common type; sporadic cases
- Affects most joints
- Four limbs involved symmetrically

Distal arthrogryposis

- Hands, feet (mainly)
- Potential specific gene defect association

Syndromic

- Primary neurological/muscle disease association

CAUSES

- Neurologic disorder (e.g. anterior horn disease)
  - 70–80% of cases
- Crowding *in utero*
  - Uterine malformation (e.g. intrauterine fibroids)
  - Multiple gestation pregnancy
- Oligohydramnios
  - Amniotic fluid volume low/abnormally distributed
- Maternal disorder (e.g. multiple sclerosis)
- Genetic disorder (e.g. spinal muscular atrophy type I)
- Muscle/connective tissue disorder (e.g. dystrophy, myopathy)

COMPLICATIONS

- Can't walk/delayed walking
- Psychosocial effects (e.g. shame, depression, social anxiety)

SIGNS & SYMPTOMS

- Congenital malformations present
- Typically affects all joints (potentially leg/arm joints only)
- Affected joints contracted (flexion/extension)
- Internal shoulder rotation
- Wrist, digit flexion
- Elbow, knee extension
- Hip dislocation
- Club feet
- Muscle weakness (especially amyoplasia)

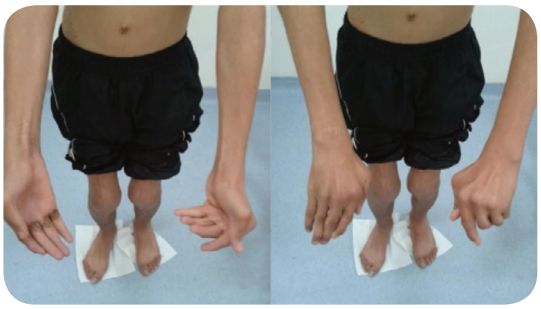

Figure 117.1 Contractures in the hands of an individual with arthrogryposis.

DIAGNOSIS

- Physical examination

DIAGNOSTIC IMAGING

Ultrasound

- 50% of diagnoses prenatal
  - Low mobility/abnormal fetus position

MRI

LAB RESULTS

- Test for cause (e.g. chromosomal microarray analysis for chromosomal abnormalities, muscle biopsy for myopathic disorders)

TREATMENT

SURGERY

- E.g. wrist surgery

OTHER INTERVENTIONS

- No curative method
  - Increase joint mobility, muscle strength, adaptive use pattern development
- Occupational therapy
  - Joint manipulation, casting
- Limb movement-enhancing devices
- Splinting

CLUBFOOT

osms.it/clubfoot

PATHOLOGY & CAUSES

- Common congenital malformation; one/both feet rotated
  - AKA talipes equinovarus
- Talus malformation
  - Feet, calf, peroneal muscles' medial side developed abnormally
- Isolated or can be associated with developmental dysplasia of hip, Larsen syndrome (+ other hip, knee, elbow malformations), spina bifida, arthrogryposis

TYPES

Congenital

- Affects bones, muscles, tendons, blood vessels

Syndromic

- Additional anatomic malformations and/or chromosomal/genetic abnormalities

Positional

- Fetal position (e.g. breech presentation), often restrictive uterine environment (e.g. oligohydramnios)

CAUSES

- Idiopathic (most cases)
- Structural anomalies
- Chromosomal/genetic abnormalities

RISK FACTORS

- Biologically male (2:1 male:female)
- Early amniocentesis
- Genetic factors
- Family history
- Multiple gestation pregnancy
- Oligohydramnios
- Uterine abnormality
- Fetal neuromuscular disorders

COMPLICATIONS

- Walking difficulty/inability
- Post-treatment recurrence
- Psychosocial effects

SIGNS & SYMPTOMS

- Bilateral (50% of cases)
- Three components
  - Hindfoot equinus
  - Midfoot varus
  - Forefoot adduction
- Individuals walk on feet sides (typically)
- Affected foot potentially smaller

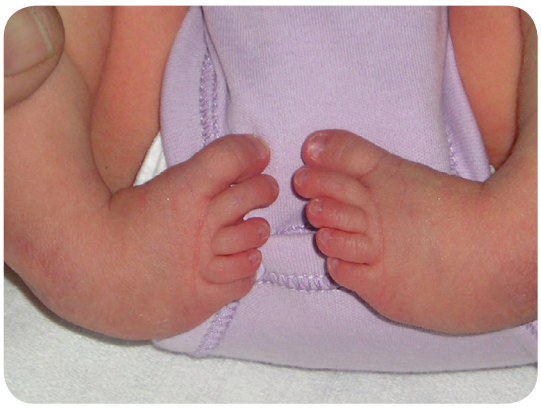

Figure 117.2 A neonate with club feet.

DIAGNOSIS

- Clinical diagnosis at birth

DIAGNOSTIC IMAGING

Ultrasound

- Prenatal
  - Abnormal foot positioning

LAB RESULTS

- Amniocentesis
  - Karyotype detects chromosomal abnormality (e.g. aneuploidy)

OTHER INTERVENTIONS

- Conservative therapy
  - Bracing, proper foot positioning, casting (Ponseti method)

TREATMENT

SURGERY

- Sometimes required (e.g. Achilles tenotomy → release tightness)

CONGENITAL HIP DYSPLASIA

osms.it/congenital-hip-dysplasia

PATHOLOGY & CAUSES

- Congenital malformation; abnormal acetabulum, proximal femur development → hip joint mechanical instability
  - AKA developmental hip dysplasia/ congenital hip dislocation
  - Presents at birth/childhood
- Joint ligament laxity/abnormal *utero* positioning → abnormal development, contact between acetabulum, femoral head
- Possible conditions associated (e.g. Ehlers–Danlos, spina bifida)

TYPES

Dislocation

- Femoral head completely outside acetabulum

Subluxation

- Femoral head partially outside acetabulum

Dislocatable

- Femoral head within acetabulum at rest, examination maneuvers dislocate easily (unstable hip joint)

Subluxatable

- Femoral head loose within acetabulum, examination partially dislocates (mildly unstable hip joint)

Reducible

- Femoral head outside acetabulum at rest, maneuvers can locate within acetabulum

Dysplasia

- Abnormally-shaped hip joint (usually shallow acetabulum)

RISK FACTORS

- Breech presentation
- Other anomalies present (e.g. congenital torticollis, congenital foot malformation)
- Family history
- Swaddling practices
- Biologically female
- First-born infant
- Oligohydramnios
- Limited fetal mobility

COMPLICATIONS

- Affected leg shorter, painful hip joint
- Osteoarthritis
- Decreased motion range → restricted hip joint adduction, flexion
- Asymmetric gait
- Low back pain
- Femoral head necrosis
- Psychosocial effects

SIGNS & SYMPTOMS

- Severity-, age-dependent
- Usually unilateral, left hip ↑ affected (20% of cases bilateral)
- Hip instability
  - *Ortolani maneuver*: infant supine, hips/knees flexed 90°; hip abducted, pulled anteriorly → dislocated femoral head slides back into acetabulum → palpable/audible clunk
  - *Barlow maneuver*: infant supine, hips/knees flexed 90°; hip abducted, pushed anteriorly → femoral head slides out of acetabulum → clunk
- Asymmetric thigh, groin skin creases
- Galeazzi sign
  - Knee height difference when infant supine (hips flexed, knees bent, feet on examining table) posterior displacement in dysplastic hip → affected side's knee lower
- Adductor spasm → limited hip abduction
- Pain (uncommon)

DIAGNOSIS

- Clinical evaluation

DIAGNOSTIC IMAGING

Hip ultrasound, X-ray

- To detect abnormal acetabulum development, femoral head position

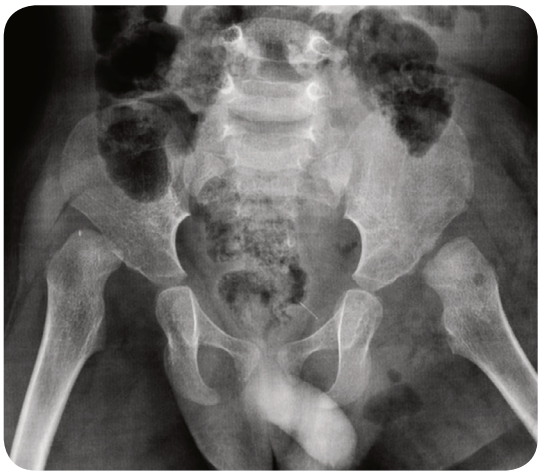

Figure 117.3 A plain radiograph of the pelvis of an infant with severe congenital hip dysplasia. There is complete dysplasia of both acetabula and superior dislocation of the femoral heads.

TREATMENT

- Early treatment critical
  - Obtain, maintain concentric hip reduction

SURGERY

- Closed/open hip reduction

OTHER INTERVENTIONS

- Abduction splinting
  - Device holds affected hip abducted, externally rotated (e.g. Pavlik harness)

CRANIOSYNOSTOSIS

osms.it/craniosynostosis

PATHOLOGY & CAUSES

- Premature calvarial suture closure → craniofacial malformation
- Abnormal dural attachments → tensile forces prevent bone growth → early suture fusion
- Abnormal skull growth
  - ↓ in perpendicular direction to fused suture
  - ↑ in parallel direction to accommodate brain growth
- Most cases isolated, sporadic; possibly genetic syndrome (e.g. Apert syndrome, Crouzon syndrome)

TYPES

- Classified by affected suture
  - Sagittal (most common)
  - Coronal
  - Metopic
  - Lambdoid
  - Multiple sutures

RISK FACTORS

- Multiple pregnancies
- Uterine abnormalities

COMPLICATIONS

- ↑ intracranial pressure
  - Vomiting, papilledema, headache
- ↓ brain growth
- Vision, hearing, speech, feeding impairments
- Neurodevelopmental delay
- Obstructive sleep apnea
- Abnormal head shape → psychosocial effects

SIGNS & SYMPTOMS

- *Phenotypes:* variable head shape, facial features (suture-dependent)
  - Sagittal suture fused → narrow, long skull (scaphocephaly/dolichocephaly)
  - Coronal/lambdoid sutures fused → diagonal skull malformation, asymmetric orbits (plagiocephaly)
  - Metopic suture fused → narrow, triangle-shaped forehead + prominent midline ridge (trigonocephaly)
  - Multiple sutures fused → Kleeblattschädel anomaly/microcephaly
  - Coronal sutures fused bilaterally → short, broad skull (brachycephaly)
  - Coronal suture fuses + any other suture → oxycephaly

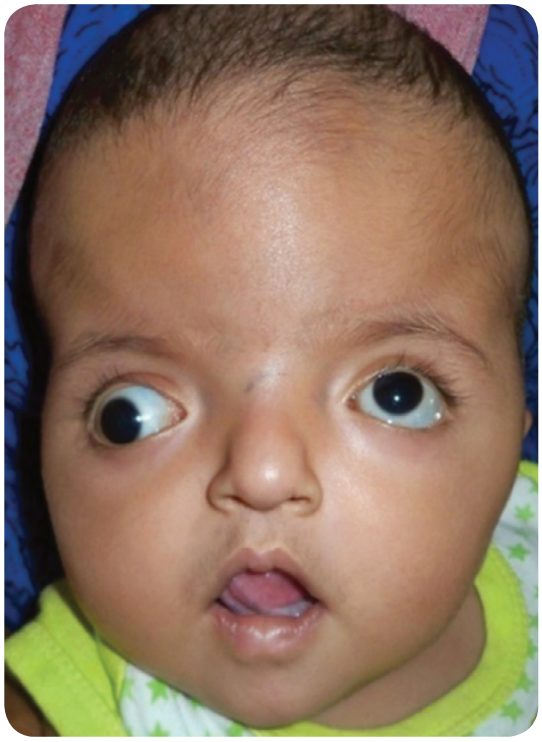

Figure 117.4 Facial features of an child with craniosynostosis in Apert syndrome.

DIAGNOSIS

- Physical examination

DIAGNOSTIC IMAGING

X-ray, CT scan

- Identify fusion, malformation extent

LAB RESULTS

- Cephalometry → precisely measure head dimensions
- Genetic testing → identify mutations
- Funduscopy → detect papilledema

TREATMENT

SURGERY

- Reconstruct craniofacial structure

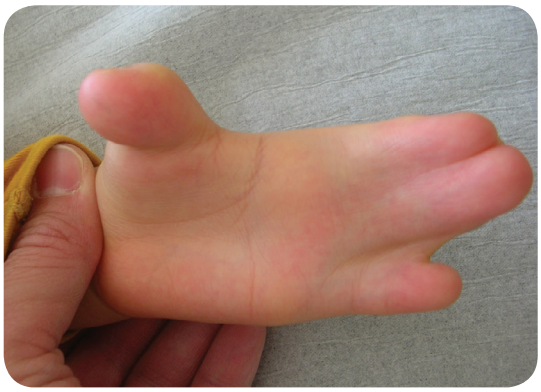

Figure 117.5 Syndactyly seen in an individual with Apert syndrome, which also causes craniosynostosis.

FLAT FEET

osms.it/flat-feet

PATHOLOGY & CAUSES

- Common malformation; moderate/complete foot arch flattening
  - AKA pes planus/fallen arches
  - Congenital/adult-acquired
- *Children:* abnormal foot muscle, tendon, bone development
- *Adults:* ↑ activity of proteolytic enzymes → break down muscle tendons → foot arch falls

TYPES

Rigid pes planus

- ↓ tarsal and subtalar joint range of motion + arch does not increase with toe raising

Flexible pes planus

- Physiologic or pathologic causes related to associated conditions (e.g. ligamentous laxity, foot muscle motor weakness, bony abnormalities, generalized syndromes)
- Type I
  - Most common type
  - Calcaneovalgus heel (depressed longitudinal arch that is associated with varying amounts of heel eversion)
  - Functional flat foot
- Type II
  - Hypermobile flat foot
  - Lax ligamentous and tight heel cords
- Type III
  - Clinical pes planus
  - Involves tibialis posterior tendon dysfunction
  - Often seen in dancers, ice skaters, athletes (e.g. basketball, tennis, soccer, ice hockey)

RISK FACTORS

- Loose connective tissue (e.g. Ehlers–Danlos syndrome)
- Neuromuscular conditions (e.g. cerebral palsy)
- Tarsal coalition (abnormal tarsal bone connection)
- Peroneal spasticity
- ↑ physical activity
- ↑ stress to foot
- Injury
- Increasing age (relatively common in biologically-female individuals > 40 years old)
- Obesity
- Rheumatoid arthritis
- Pregnancy (↑ elastin)

COMPLICATIONS

- Knee, hip, back pain
- Progress to high arches (adolescence)
- Abnormal gait → injuries
- Tendonitis

SIGNS & SYMPTOMS

- Normal foot arch absent (flat)
- Foot sole presses ground almost completely
- Abnormal gait
  - ↑ inward foot roll (overpronation)
- May involve foot, ankle, knee, hip, back pain

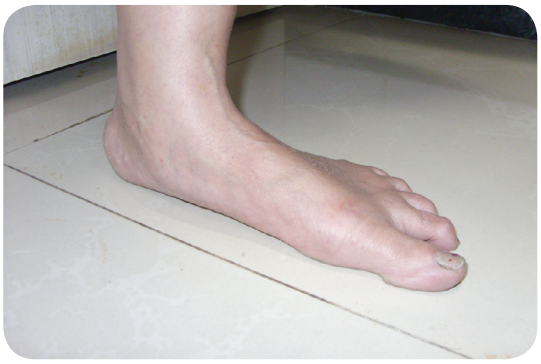

Figure 117.6 Complete collapse of the longitudinal arch has resulted in complete contact of the sole of the foot with the ground.

DIAGNOSIS

- Clinical evaluation
- Wet footprint test
  - Individual wets feet, stands on paper → footprint with ↑ surface area

DIAGNOSTIC IMAGING

Feet X-ray

- Talonavicular coverage angle → abnormal lateral rotation
- ↓ calcaneus, inferior foot angle (calcaneal pitch)
- ↑ long talus axis, first metatarsal bone angle (Meary's angle)
- The anteater nose sign
  - Anterior tubular elongation of the superior calcaneus; approaches/overlaps the navicular indicated calcaneonavicular coalition

TREATMENT

- Sometimes unnecessary (arch may develop)

SURGERY

- Resection of abnormal bridge of bony, cartilaginous, or fibrous tissue (e.g. calcaneonavicular coalition)

OTHER INTERVENTIONS

- Conservative treatment
  - Supportive shoes
  - Orthotics (insoles stop inward roll)
  - Casting
  - Analgesics (e.g. NSAIDs)
  - Physical therapy

GENU VALGUM

osms.it/genu-valgum

PATHOLOGY & CAUSES

- *Knee malformation:* knees bend towards each other
  - Typically resolves by age nine
  - AKA "knock-knees"
  - Less common than genu varum

CAUSES

- Physiologic (age 2–5)
- Poor nutrition
- Obesity
- Lower extremity fracture
- Calcium deficiency
- Vitamin D deficiency
- Skeletal dysplasia
- Neoplasm
- Idiopathic

COMPLICATIONS

- Knee osteoarthritis
- Injuries
- Knee chondromalacia
- Psychosocial effects

SIGNS & SYMPTOMS

- Knee malformation
  - Knee joint's proximal portion bends inwards
  - Knee joint's distal portion bends outwards
- Can't touch knees, feet together
- Gait abnormalities
- Pain (uncommon)

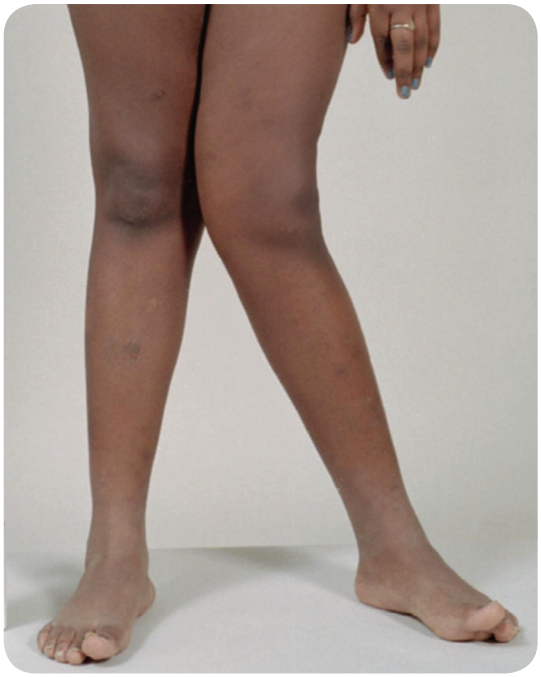

Figure 117.7 An individual with genu valgum of the left leg secondary to surgery and radiotherapy to treat a synovial sarcoma of the lateral distal femoral epiphysis as a child. The medial epiphysis continued to grow whilst growth of the lateral epiphysis was stunted.

DIAGNOSIS

- Clinical evaluation

DIAGNOSTIC IMAGING

X-ray

- Both legs (hips to feet) in standing position

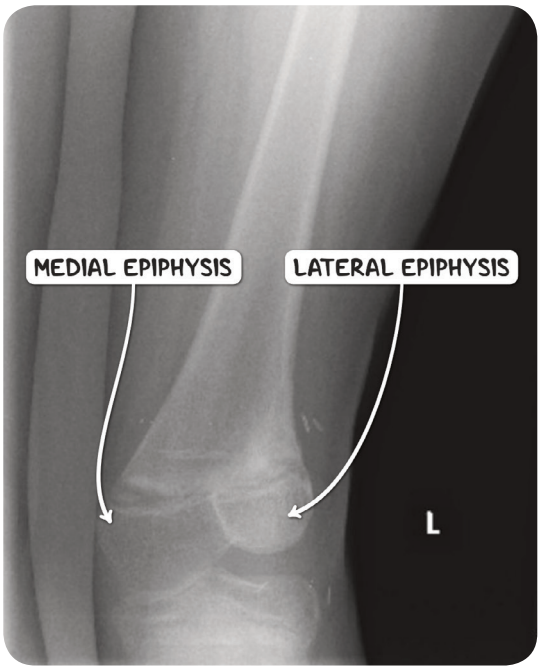

Figure 117.8 An X-ray image of the affected knee. The medial head is much larger than the lateral head.

TREATMENT

- Treatment of underlying causes (e.g. vitamin D deficiency)

SURGERY

- If malformation persists after age 10
  - Medial distal femoral epiphysis stapling
  - Total knee replacement

OTHER INTERVENTIONS

- Orthotic devices
- Bracing

GENU VARUM

osms.it/genu-varum

PATHOLOGY & CAUSES

- *Most common knee malformation:* knees bow
  - AKA bow-legs

CAUSES

- Physiologic (birth to 18 months)
- Vitamin D deficiency (e.g. rickets)
- Poor nutrition
- Other musculoskeletal conditions (e.g. skeletal dysplasia)
- Infection/tumors/lower extremity fracture → abnormal leg growth
- Blount disease

COMPLICATIONS

- Knee osteoarthritis
- Psychosocial effects

SIGNS & SYMPTOMS

- Knee malformation
  - Knee's distal portion bends inwards
  - Proximal portion bends outwards (like archer's bow)
- Usually bilateral

DIAGNOSIS

- Clinical evaluation

DIAGNOSTIC IMAGING

X-ray

- Both legs (hips to feet) in standing position

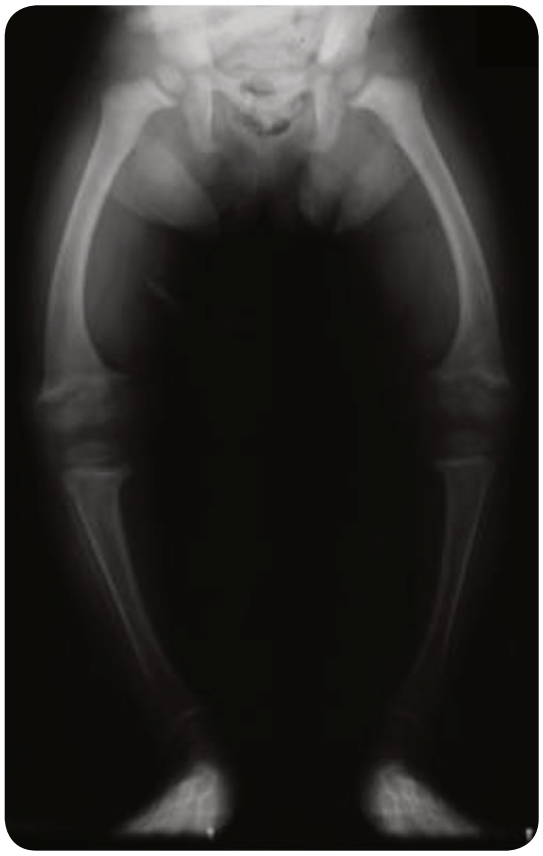

Figure 117.9 An X-ray image of a child with rickets displaying genu varum.

TREATMENT

- Treatment of underlying causes (e.g. vitamin D deficiency)

SURGERY

- If malformation persists

OTHER INTERVENTIONS

- Splinting
- Bracing

PECTUS EXCAVATUM

osms.it/pectus-excavatum

PATHOLOGY & CAUSES

- *Congenital thoracic wall malformation:* chest appears caved-in
  - Most common anterior chest wall disorder
  - AKA funnel chest
- Abnormal sternum, rib cage growth
- Unknown cause
  - *Possibly:* increased intrauterine pressure, increased sternum traction, abnormal cartilage development
- Usually sporadic

RISK FACTORS

- Biologically male (3–5:1 male:female)
- Family history
- Connective tissue disorders (e.g. Marfan syndrome, Ehlers–Danlos)
- Neuromuscular diseases
- Genetic conditions (e.g. Noonan syndrome)
- Rickets
- Congenital diaphragmatic hernia

COMPLICATIONS

- Cardiorespiratory function impairments
- Psychosocial effects

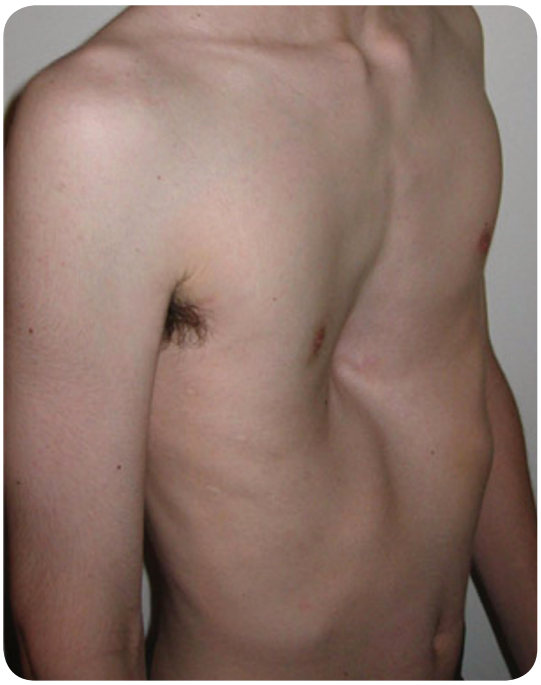

Figure 117.10 An individual with pectus excavatum.

SIGNS & SYMPTOMS

- Physical
  - *Chest malformation:* sternum's lower end depressed, lower ribs may protrude, narrowed chest wall diameter
  - Displaced heartbeat
  - Heart murmurs
  - Diminished lung sounds
  - Exercise intolerance
- Potential chest/back pain
- Respiratory symptoms
  - Shortness of breath, tachypnea

DIAGNOSIS

- Clinical evaluation

DIAGNOSTIC IMAGING

Chest CT scan

- Determine severity; assess lung, heart effects

LAB RESULTS

- Pulmonary function tests
  - Normal forced vital capacity
  - Total lung capacity, residual volume may be abnormal

OTHER DIAGNOSTICS

- Cardiology exams (e.g. electrocardiogram, echocardiography)
  - Abnormalities if heart compression, rotation
- Exercise testing
  - Impairment severity correlates with defect's degree

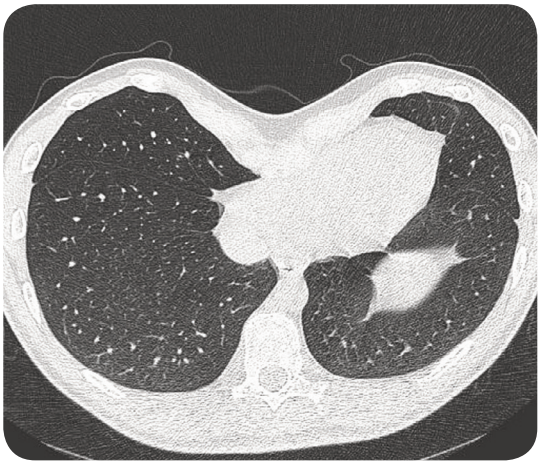

Figure 117.11 A CT scan of the chest in the axial plane demonstrating pectus excavatum.

TREATMENT

- Some cases resolve spontaneously (usually worsens in adolescence)

SURGERY

- Moderate/severe pectus excavatum

PIGEON TOE

osms.it/pigeon-toe

PATHOLOGY & CAUSES

- *Common developmental variation:* toe inward rotation
  - AKA in-toeing
- Typically resolves spontaneously
- Results from intrauterine molding

CAUSES

- Metatarsus adductus (most common in infants < one year old)
- Internal tibial rotation (most common between age 1–4)
- Increased femoral anteversion (most common in children > three years old)

COMPLICATIONS

- Long-term functional problems (rare)

SIGNS & SYMPTOMS

- Abnormal toe rotation when walking/standing
  - *Metatarsus adductus:* inward forefoot rotation
  - *Tibial torsion:* inward shin bone twisting
  - *Femoral anteversion:* inward femur twisting
- Non-flexible/flexible (if malformation can be hand-straightened)

DIAGNOSIS

- Clinical evaluation

DIAGNOSTIC IMAGING

X-ray

- Assess severity

TREATMENT

TREATMENT

- Observation
- Surgery rarely recommended