NOTES
BONE & JOINT PATHOLOGY

GENERALLY, WHAT IS IT?

PATHOLOGY & CAUSES

- Non-neoplastic disorders affecting bones, joints
- Generally result in weakened bones; pathologic fractures, malformations
- Include disorders of normal bone structure due to
  - Impaired bone mineralization (rickets, osteomalacia)
  - Failure of bone resorption (osteopetrosis, osteosclerosis)
  - Disorders of bone formation (osteogenesis imperfecta)
  - Imbalance between bone formation, bone resorption (osteoporosis, Paget's disease)
  - Stress injury (Osgood–Schlatter disease)
  - Impaired vascularization (Legg–Calvé–Perthes disease)

SIGNS & SYMPTOMS

- May be asymptomatic
- Most common symptoms include
  - Bone pain, bone tenderness, pathologic fractures, bone malformations, nerve/tissue compression

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Lytic/blastic changes
- Bone fractures, malformations

Bone scan scintigraphy

- Extent, distribution of skeletal involvement

LAB RESULTS

- Etiology-dependent
  - Bone specific alkaline phosphatase, creatinine kinase, Ca^{2+}, serum 25(OH)D levels, etc.
  - *Biopsy:* microscopic changes

OTHER DIAGNOSTICS

- Clinical presentation

TREATMENT

- Causative treatment, palliative treatment (management of symptoms with no effect on course of the disease)

MEDICATIONS

- Supplementation therapy (vitamin D)
- Bisphosphonate therapy

SURGERY

- Surgery

OTHER INTERVENTIONS

- Fracture management (braces, intramedullary rods, etc.)

FIBROUS DYSPLASIA OF BONE

osms.it/fibrous-dysplasia-of-bone

PATHOLOGY & CAUSES

- Rare disorder → normal bone tissue replaced by fibrous tissue
- → brittle, weak, fracture-prone bones

TYPES

- Monostotic
  - AKA (McCune–Albright syndrome)
  - Most common; involves one bone
- Polyostotic
  - Involves multiple bones

CAUSES

- Post-zygotic activating mutations of guanine nucleotide stimulatory protein (GNAS) gene, which encodes α subunit of the G_s coupled protein receptor → constitutive receptor activation → replacement of bone with fibrous tissue

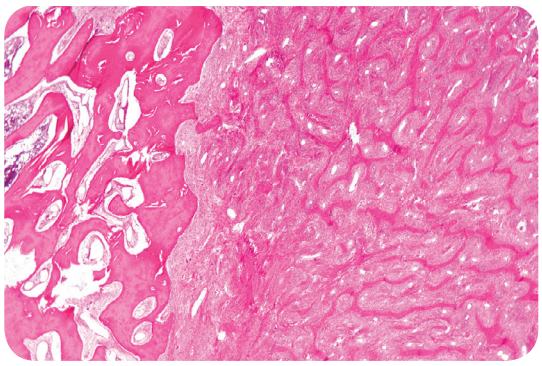

Figure 107.1 The histological appearance of bone in an individual with fibrous dysplasia of bone. Normal bone tissue is on the left. The affected bone is composed of thin disordered trabeculae. The bone marrow spaces are packed with fibrous tissue.

RISK FACTORS

- Sex
  - ↑ individuals who are biologically male
- Age
  - Symptoms usually occur in teen years

COMPLICATIONS

- Pathologic fractures
  - Repeated pathologic fractures can → "shepherd crook malformation" (varus angulation of the proximal femur)
- Severe scoliosis if spine affected

SIGNS & SYMPTOMS

- Mostly asymptomatic
- During teen years
  - Pain, swelling, pathologic fractures, malformations
- Most commonly affects proximal femur, tibia, ribs, skull
- Rare case of optic nerves/auditory canal compression
  - Vision/hearing loss

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Well-circumscribed lytic lesions in metaphysis/diaphysis with "ground glass" appearance
- Undulating pattern of cortical bone due to endosteal erosion
- *"Rind sign"*: thick, sclerotic bone layer surrounding lytic lesion
- Pathologic fracture → periosteal reaction

Total body scintigraphy

- Identify extent of bone lesions
  - Increased Tc^{99} uptake

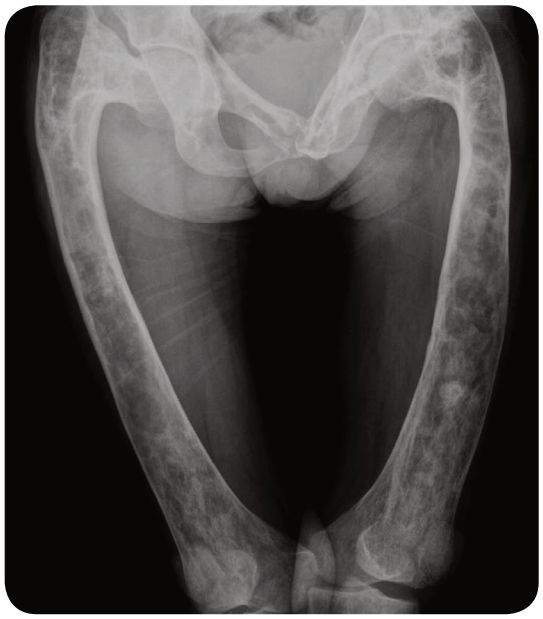

Figure 107.2 An X-ray image of the femurs demonstrating a shepherd's crook malformation. Both femurs are involved in this case of polyostotic fibrous dysplasia.

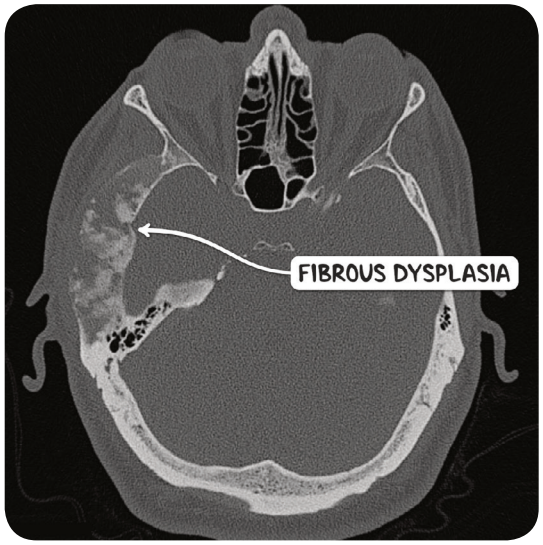

Figure 107.3 A CT scan of the head in the axial plane demonstrating fibrous dysplasia of the squamous temporal bone in a case of monoostotic fibrous dysplasia.

LAB RESULTS

Biopsy

- Thin, irregular bony trabeculae; described as "Chinese figures"

OTHER DIAGNOSTICS

Clinical presentation

- Albright syndrome may present along with endocrine abnormalities; e.g. "*café-au-lait*" spots, often on neck

TREATMENT

- Palliative; disease incurable
- *Asymptomatic:* observation
- *Symptomatic:* medications, surgery

MEDICATIONS

- Bisphosphonate therapy
  - Inhibit osteoclast activity, prevent bone loss, decrease bone pain

SURGERY

- Curettage; bone grafting; stabilization with plates, screws
- Rarely effective; high rate of recurrence

LEGG–CALVÉ–PERTHES DISEASE

osms.it/legg-calve-perthes_disease

PATHOLOGY & CAUSES

- Hip disorder characterized by osteonecrosis of proximal femur head due to compromised blood supply
- ↓ blood supply to proximal femoral head → osteonecrosis → infiltration of new blood vessels and resorption of necrotic bone → bone mass loss, growth cessation,weakening of bone
- Cause of blood supply disruption (ligamentum teres femoris or medial circumflex femoral artery) unknown

RISK FACTORS

- Age
  - Usually affects children 3–12 years
- Sex
  - ↑ individuals who are biologically male
- Heredity
- Endocrinologic abnormalities
- Hemodynamic disorders
- Trauma
- Steroid use

COMPLICATIONS

- Loss of bone mass can → pathologic fractures, malformations (e.g. coxa magna)
- ↑ risk of osteoarthritis in adulthood

SIGNS & SYMPTOMS

- Intermittent/chronic throbbing hip pain
- Referred knee/groin pain exacerbated by movement, especially internal hip rotation,
- Soreness, altered gait
- Reduced range of motion

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Initial scans often normal; nonspecific effusion of joint (widening of joint space) may be present
- As disease progresses
  - Flattening (coxa plana), fragmentation, demineralization of femoral head with subchondral lucency; proximal femoral neck malformation (coxa magna)

Bone scintigraphy

- Confirmation and evaluation
  - Extent of involvement
- Focal area of ↓ uptake in femoral head

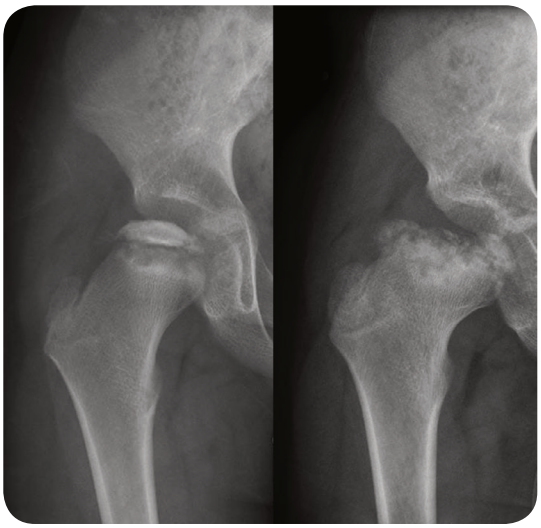

Figure 107.4 X-ray images of the right hip in a child with Legg–Calvé–Perthes disease. The diagnosis was made at six years (left) and by 8.5 years (right) the epiphysis has completely collapsed due to osteonecrosis.

MRI

- Confirmation and evaluation
  - Extent of involvement
- Hypointense bone marrow changes, subluxation of femoral head

TREATMENT

SURGERY

- External fixation to stabilize hip bone, relieve it from carrying body's weight
- Hip replacement is usually required > age 50

OTHER INTERVENTIONS

- Traction to remove mechanical pressure, reduce wear
- Braces, physiotherapy to restore range of motion
- Avoidance of contact sports/games, running, prolonged weight bearing
  - Swimming, cycling recommended to exercise hip muscles, restore range of motion

OSGOOD-SCHLATTER DISEASE

osms.it/osgood-schlatter_disease

PATHOLOGY & CAUSES

- Traction phenomenon characterized by stress inflammation, stress injury at point of insertion of patellar tendon (apophysitis) on proximal tibial tubercle
- AKA apophysitis of tibial tubercle

CAUSES

- Overuse during physical activity
  - Repetitive quadriceps contraction → traction on tibial tuberosity → micro-avulsion fractures of tibial tubercle, tendinous inflammation
- Severe cases may → complete tibial tubercle avulsion fracture (detachment of tibial tubercle)

RISK FACTORS

- Age
  - ↑ common in 11–14 year olds
- Activity level
  - ↑ common in physically active individuals

SIGNS & SYMPTOMS

- Swelling, pain at tibial tubercle
  - Exacerbated by trauma, activity; relieved by rest
- Limping; bony prominence of tibial tubercle
- Avulsion fracture → acute onset of pain
- Usually asymmetric, but often presents bilaterally

DIAGNOSIS

DIAGNOSTIC IMAGING

- Used only for atypical presentation (pain not related to activity, fever, rash etc.) to exclude other conditions (e.g. osteomyelitis)

X-ray

- Elevation of tibial tubercle
- Fragmentation of tibial tubercle
- Soft tissue swelling
- Calcification/thickening of patellar tendon

OTHER DIAGNOSTICS

- Clinical presentation

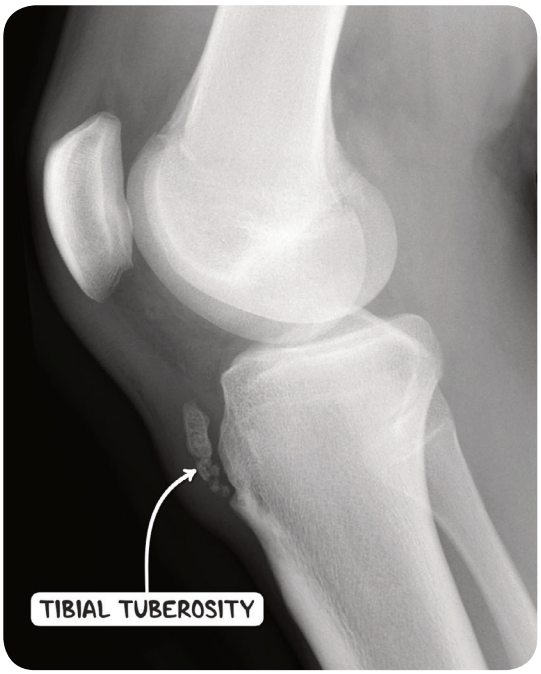

Figure 107.5 A lateral X-ray image of the knee in an individual with Osgood–Schlatter disease. There is fragmentation of the tibial tuberosity and overlying soft tissue swelling.

TREATMENT

MEDICATIONS

- Short term analgesics/NSAIDs use

SURGERY

- Ossicle resection, excision of tibial tuberosity
  - If everything fails, for individuals with closed growth plates

OTHER INTERVENTIONS

- Usually no treatment required
- Physical therapy

OSTEOGENESIS IMPERFECTA

osms.it/osteogenesis-imperfecta

PATHOLOGY & CAUSES

- Disease characterized by brittle bones prone to fractures due to impaired type I collagen synthesis
- Type I collagen
  - *Formation:* two α1 chains combine with one α2 chain → triple stranded type 1 procollagen → post-translational modification (folding, cross linking) → strong fibrils with enormous tensile strength
  - Important for structural integrity of bones, joints, eyes, ears, teeth and skin
- Affects primarily bones, but also other tissues containing type I collagen → structural abnormalities in affected tissues

TYPES

- Previously classified into nine subtypes based on family history, radiologic, clinical features
- Included is modified clinical classification → severity

Mild (type I)

- Mild bone fragility
- Variable fracture rate; minimal bone fractures before learning to walk
- Minimal malformation, normal stature
- Adults at higher risk for hearing loss, premature osteoporosis following menopause

Moderate to severe (types III–IX)

- Type III
  - Most severe type compatible with survival
- Moderate to severe rate of fractures
- Moderate malformations, short stature
- Children
  - Higher risk of hearing loss
- Adults
  - Earlier onset of hearing loss and premature osteoporosis than in mild form

Lethal form (type II)

- Most cases die *in utero*/within first year of life
- Severe fractures *in utero*
- Severe deformities; short stature
- Pulmonary hypoplasia → respiratory failure

CAUSES

- Autosomal dominant mutation of *COL1A1* or *COL1A2* (>90%), other genes encoding α1, α2 chains of type I collagen → misfolding of collagen proteins, loss of function → bone loss, fragility

SIGNS & SYMPTOMS

- Highly variable presentation
- Pathologic fractures with minimal/no trauma, malformations, short stature, scoliosis
- Skull malformations may cause compression, neurologic symptoms
- Blue discoloration/translucency of sclera due to decreased collagen, exposure of choroidal veins
- Hearing loss due to abnormalities in middle, inner ear ossicles
- Dentinogenesis imperfecta
  - Small, blue/translucent, worn down teeth
- Decreased structural integrity, hypermobility of ligaments, joints, skin
- Easy bruising

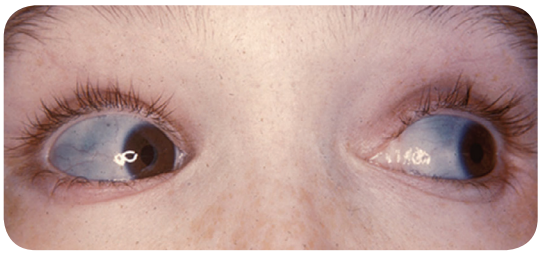

Figure 107.6 Blue sclera in an individual with osteogenesis imperfecta.

DIAGNOSIS

DIAGNOSTIC IMAGING

Prenatal ultrasound

- Lethal form
  - Severe micromelia (small, undeveloped extremities)
  - Decreased bone mineralization → skull compression with transducer pressure
  - Multiple bone fractures

Postnatal X-ray skeletal survey

- Mild form
  - Thinning of cortical bone
  - Wormian bones may be present (small, irregular bones between cranial sutures)
- Moderate to severe form
  - Cystic metaphyses
  - Severe osteoporosis
  - Popcorn calcification of metaphysis, epiphysis of long bones
  - Vertebral/rib fractures common
- Lethal form
  - Beaded ribs
  - Severe osteoporosis
  - Multiple fractures, malformations of long bones

LAB RESULTS

- ↑ serum alkaline phosphatase in blood
- ↑ Ca^{2+} in the urine

Biopsy

- Disorganized bone; decrease of cortical, trabecular width; cancellous bone volume
- Increased bone remodeling

Dermal fibroblast culture

- Abnormalities in quality/quantity of collagen synthesis

Prenatal DNA mutation analysis

- For at-risk pregnancies

OTHER DIAGNOSTICS

- Clinical presentation

TREATMENT

MEDICATIONS

- Bisphosphonate treatment for moderate to severe form (e.g. intravenous pamidronate)

SURGERY

- Surgical malformity correction
- Fracture management with intramedullary rods placement; telescoping rods for actively-growing individuals

OTHER INTERVENTIONS

- Physical therapy → prevent contractures, bone loss due to immobility

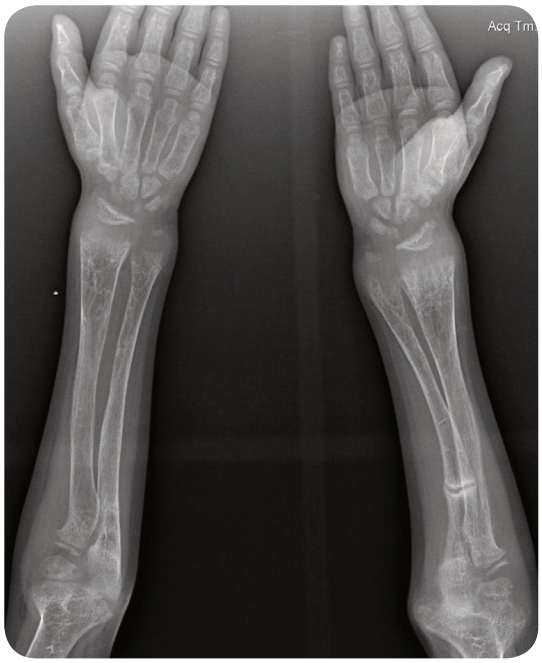

Figure 107.7 X-ray images of the arms of an individual with osteogenesis imperfecta. There is generalised osteoporosis as well as multiple fractures and malformations..

OSTEOMALACIA

osms.it/osteomalacia

PATHOLOGY & CAUSES

- Inadequate bone mineralization in adults due to lack of vitamin D, Ca^{2+}/PO^{3-}
- AKA rickets (in children)
- ↓ Ca^{2+} inhibits normal mineralization of newly formed osteoid during bone remodelling → weakening, softening of bones

CAUSES

- Vitamin D deficiency → insufficient intestinal absorption of Ca^{2+}
  - Most common cause
  - Insufficient sun exposure → UVB rays initiate vitamin D synthesis in the skin
  - Chronic kidney disease/ liver disease → lack of vitamin D activation
  - Insufficient intake
  - Malabsorption syndrome → insufficient intestinal absorption of Ca^{2+}, other minerals
  - Can occur as adverse effect of long term anticonvulsant use (e.g. phenytoin)
- X-linked hypophosphatemia

RISK FACTORS

- Limited sun exposure, use of strong sunscreens

- Dietary
  - Lactose intolerance, vegetarian diet
- Darker skin pigmentation

COMPLICATIONS

- May → secondary hyperparathyroidism
- Prolonged secondary hyperparathyroidism → Ca^{2+} resorption from bones → osteoporosis

SIGNS & SYMPTOMS

- Diffuse bone and joint pain
- Proximal muscle weakness
- Muscle spasms of hands, feet, tingling/numbness
- Bone fragility, increased risk of fractures with minimal trauma

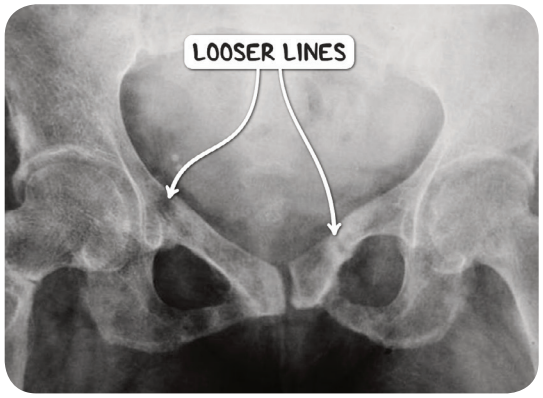

Figure 107.8 An X-ray image of the pelvis of an individual with osteomalacia. There are numerous pseudofractures, or Looser lines, in both the inferior and superior pubic rami.

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- ↓ bone mineral density, AKA osteopenia
- Loss of of trabecular bone, thinning of cortical bone
- "Looser lines"
  - Transverse lucencies resembling fractures, AKA pseudofractures
- In case of secondary hyperparathyroidism
  - Subperiosteal resorption of phalanges, bone cysts

LAB RESULTS

- ↓ Serum 25(OH)D levels
- ↓ Ca^{2+}, ↓ PO^{3-}
- ↑ Parathyroid hormone levels
- ↑ Alkaline phosphatase due to increased osteoblast activity

Bone biopsy with tetracycline labeling

- Rarely done
- ↑ unmineralized osteoid volume
- ↑ width of osteoid seams
- No sign of new bone mineralization

TREATMENT

MEDICATIONS

- Oral vitamin D supplementation
- Correction of Ca^{2+} intake

OTHER INTERVENTIONS

- Treat underlying cause

OSTEOPETROSIS

osms.it/osteopetrosis

PATHOLOGY & CAUSES

- Rare genetic disorder characterized by osteoclast dysfunction → hardening of the bone, AKA osteosclerosis
- Osteoclasts' failure to resorb bone → increased density, overgrowth of bones
- Despite increased density, bones have disordered architecture, lack flexibility; thus prone to fractures

TYPES

Autosomal recessive osteopetrosis, AKA infantile malignant type

- Caused by mutations in CA2 gene encoding carbon anhydrase
- Deficiency of carbonic anhydrase → inhibition of proton pumping → ↑ pH → osteoclasts fail to resorb bone because acidic environment required → imbalance between bone formation and bone resorption → excess bone formation

Autosomal dominant osteopetrosis, AKA adult benign type

- Caused by mutations in chloride channel 7 (*CLCN7*) gene; less severe type
- Associated with renal tubular acidosis → deficiency of carbonic anhydrase in kidney

SIGNS & SYMPTOMS

Autosomal recessive osteopetrosis

- Impaired growth, failure to thrive
- Osteomyelitis of mandible
- Dental abnormalities
- Visual/hearing impairment → sclerosis of skull bones, cranial nerve compression
- Hydrocephalus → obstruction of foramen magnum
- Hepatosplenomegaly and hypersplenism
  - Due to bone marrow replacement and anemia, resulting in extramedullary hematopoiesis
- Symptoms of anemia (e.g. weakness, fatigue, pallor)
- Symptoms of thrombocytopenia (e.g. bruising, hemorrhage)
- Symptoms of leukopenia (e.g. recurrent infections)

Autosomal dominant osteopetrosis

- Can be asymptomatic; most commonly affects spine, pelvis, base of skull
- Vision loss, hearing loss due to sclerosis of skull bones, cranial nerve compression
- Pathologic fractures
- Osteoarthritis

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Increased thickness, density of bones
- "Bone within bone" appearance
  - Classical for autosomal dominant osteopetrosis
- Sclerotic rings in iliac bones
- Widened costochondral junctions
- Radiolucent metaphyseal bands
- Sandwich vertebrae
  - Peripheral bony sclerosis with central lucency of vertebral body

LAB RESULTS

- Hypocalcemia (due to ↓ reabsorption of Ca^{2+})
- ↑ PTH (secondary hypoparathyroidism)
- ↑ acid phosphatase
  - Released from defective osteoclasts
- Creatinine kinase (CK-BB)
- Released from defective osteoclasts

TREATMENT

MEDICATIONS

- Ca^{2+}, PO^{3-}, vitamin D supplementation
- Osteomyelitis, other infections → antibiotics
- Anemia → erythropoietin, corticosteroids
- Leukopenia → gamma interferon

SURGERY

- Mend fractures
- Bone marrow transplantation

OTHER THERAPIES

- Not curable; treatment is supportive
- Fractures → braces
- Treat dental abnormalities

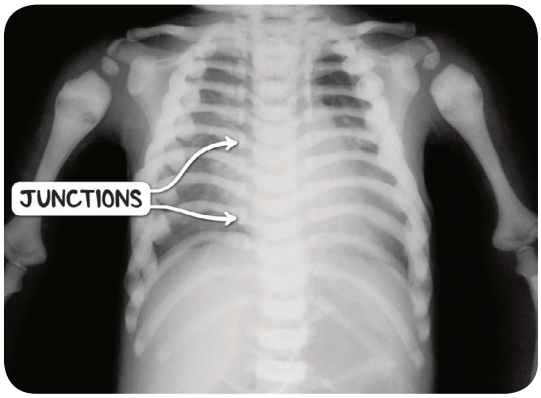

Figure 107.10 A plain chest radiograph of a child with osteopetrosis. There is characteristic widening of the costochondral junctions.

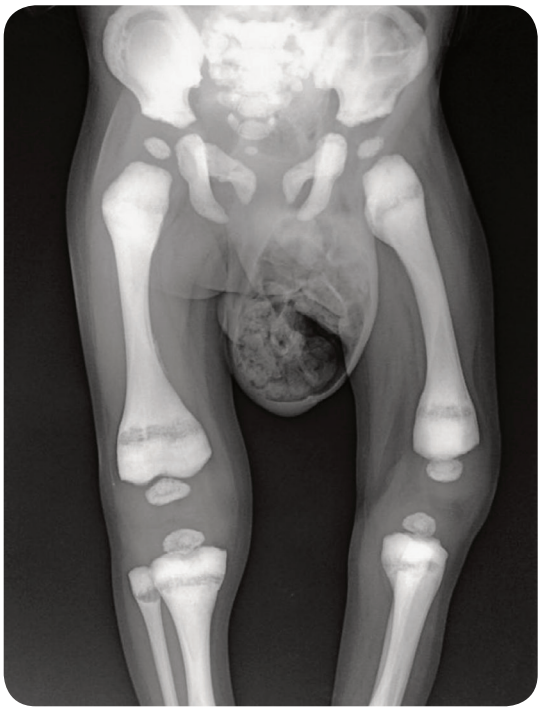

Figure 107.9 An X-ray image of a child with osteopetrosis. There is loss of bony cortico-medullary differentiation and there are lucent metaphyseal bands. There is an incidental large scrotal hernia.

OSTEOPOROSIS

osms.it/osteoporosis

PATHOLOGY & CAUSES

- Characterized by imbalance between bone formation, bone resorption → decreased bone density, pathologic fractures
- Caused by increased bone loss/decreased bone mass

RISK FACTORS

- Postmenopause (↓ estrogen)
- Alcohol consumption, smoking
- Immobility, malnutrition/malabsorption (↓ Ca^{2+})

- Hypogonadal states
  - Turner's syndrome, hyperprolactinemia, Klinefelter syndrome, hypothalamic amenorrhea, primary/secondary hypogonadism
- Endocrine disorders
  - Cushing's syndrome, hyperthyroidism, hyperparathyroidism, diabetes mellitus, acromegaly
- Inherited disorders
  - Osteogenesis imperfecta, Marfan's syndrome, hemochromatosis
- Rheumatologic disorders
  - Rheumatoid arthritis, ankylosing spondylitis, systemic lupus erythematosus
- Medications
  - Corticosteroids, antiepileptics, anticoagulants, L-thyroxine

SIGNS & SYMPTOMS

- Asymptomatic until fracture occurs
- Pathologic fractures; most commonly vertebral column, ribs, hips, wrists
- Compression fractures of vertebral column
  - *Signs:* loss of height, hunched posture, kyphosis
  - *Symptoms:* sudden back pain, radicular pain, spinal cord compression, cauda equina syndrome
- Chronic pain; unlikely without fracture

DIAGNOSIS

DIAGNOSTIC IMAGING

Dual-energy X-ray absorptiometry (DEXA scan)

- ↓ bone mineral density (BMD) ≥ 2.5 SD below the young-adult mean

LAB RESULTS

- Identification of potential secondary causes
  - Complete blood cell count
  - Ca^{2+}, PO^{3-}, creatinine, 25-hydroxyvitamin D levels
  - Thyroid-stimulating hormone (TSH)
  - 24-hour urine for calcium and creatinine

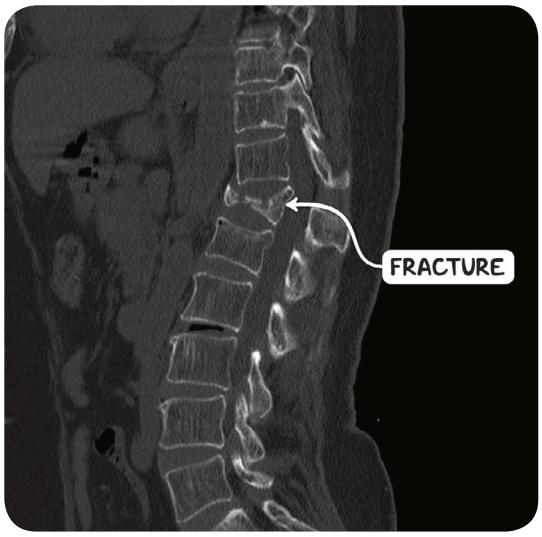

Figure 107.11 An MRI scan of the spine in the sagittal plane demonstrating a compression fracture of T12 secondary to osteoporosis.

TREATMENT

MEDICATIONS

- Ca^{2+}, vitamin D supplementation
- Oral bisphosphonates (alendronate or risedronate)
- Selective estrogen receptor modulators (raloxifene)
- Parathyroid hormone/parathyroid hormone-related protein analog for severe osteoporosis

SURGERY

- Prompt surgery in case of hip fracture

OTHER THERAPIES

- Fracture management
- Lifestyle changes
  - Exercise; smoking, alcohol cessation

OSTEOSCLEROSIS

osms.it/osteosclerosis

PATHOLOGY & CAUSES

- Abnormal diffuse/patchy hardening of bone, increased bone density due to impaired bone resorption

TYPES

Acquired
- Paget's disease
- Osteogenic bone metastasis (e.g. prostate, breast cancer)
- Myelofibrosis
- Chronic osteomyelitis
- Hypervitaminosis D
- Hypoparathyroidism
- Schnitzler syndrome

Inherited
- Osteopetrosis
- Pyknodysostosis
- Osteopoikilosis

SIGNS & SYMPTOMS

- May be asymptomatic
- Generalised bone pain
- Pathologic fractures

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray
- ↑ Bone mineral density
- Increased bone thickness, density

TREATMENT

MEDICATIONS

- Ca^{2+}, PO^{3-}, vitamin D supplementation

OTHER INTERVENTIONS

- Fracture management

PAGET'S DISEASE OF BONE

osms.it/pagets-disease-of-bone

PATHOLOGY & CAUSES

- Characterized by localized, disordered bone remodeling
  - Excessive bone resorption → disorganized compensatory bone formation
- Three phases of pathogenesis
  - *Lytic phase:* osteoclastic hyperactivity → increased rate of localized bone resorption; bone remodeling increased up to 20x
  - *Mixed lytic-blastic phase:* compensatory osteoblastic hyperactivity → accelerated bone formation
  - *Sclerotic phase:* results in thick, sclerotic, disorganized bone ("woven bone") prone to fracture; new bone infiltrated by blood vessels (e.g. hypervascular state)

CAUSES

- Unclear; possible causes include
  - Slow virus infection (e.g.

paramyxoviridae) of osteoclasts
    - Mutations of *SQSTM1*, *RANK* genes involved in osteoclasts' function regulation

COMPLICATIONS

- Osteoarthritis
    - May distort alignment of bone, associated joint → higher mechanical force; rapid wear, degeneration
- Heart failure
    - Rarely, advanced Paget's disease → excessive demand on heart due to increased hypervascularity, arteriovenous (AV) shunts in affected bone
- Neurologic impairments
    - Neural tissue compression
- Rarely, malignant transformation (osteosarcoma)

SIGNS & SYMPTOMS

- Involves one or more bones; not generalized
- Most commonly affects pelvis, femur, lumbar vertebrae, skull
- Bone pain → microfractures, periosteal changes
- Pathologic fractures
- Bony malformations
    - Enlarged skull, AKA "increasing hat size"; spinal kyphosis; bowing of long bones
- Increased localized temperature → hypervascularity
- Arthritis of associated joints
- Hearing impairment → sclerosis of the skull bones, cranial nerve compression
- Decreased range of motion

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- *Osteoporosis circumscripta*
    - Well-defined osteolytic lesions of skull in early course

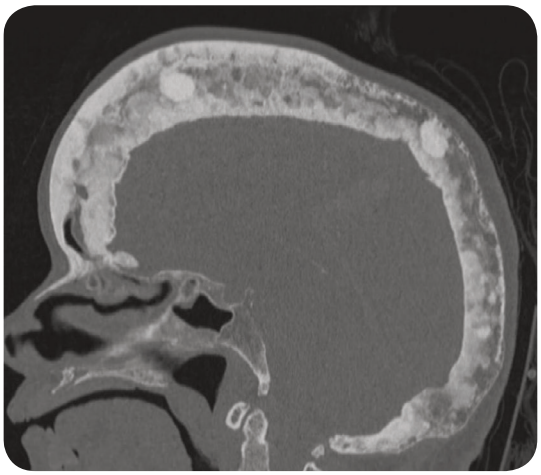

Figure 107.12 A CT scan of the head in the sagittal plane. The skull has the typical cotton wool appearance of Paget's disease of the bone.

- "Cotton wool appearance"
    - Mixed lytic/sclerotic lesions
- Looser lines
    - Transverse lucencies resembling fractures; AKA pseudofractures
- Squaring of vertebrae seen on lateral X-ray
- Tam O'Shanter sign
    - Enlarged overriding frontal bone
- "Candle flame sign"
    - Well V-shaped osteolytic lesion; characteristic of lytic phase

Bone scan scintigraphy

- Focal increased radionuclide uptake

LABORATORY RESULTS

Biopsy

- Mosaic pattern of lamellar bone
- Large, numerous osteoclasts with up to 100 nuclei (normal is 5–10)
- Affected bone marrow filled with highly vascular stroma

Blood test

- ↑ alkaline phosphatase

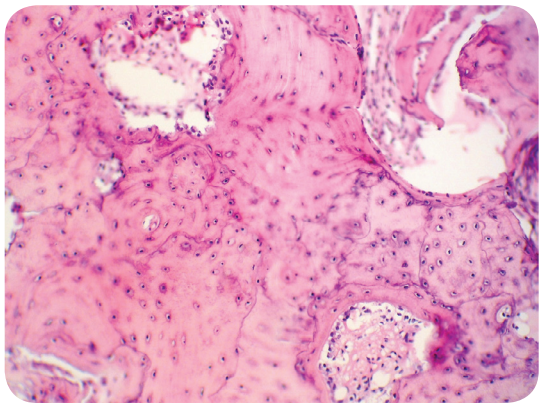

Figure 107.13 The histological appearance of bone in the Paget's disease. Bone formation is increased and highly disordered.

TREATMENT

MEDICATIONS

- Bisphosphonates (zoledronate, risedronate, pamidronate), calcitonin

SURGERY

- Correction of fractures, malformations

OTHER INTERVENTIONS

- Physical therapy

RICKETS

osms.it/rickets

PATHOLOGY & CAUSES

- Inadequate mineralization of cartilage in children's growth plates due to lack of vitamin D, Ca^{2+}, or PO^{3-}
- ↓ Ca^{2+} inhibits normal mineralization of epiphyseal growth plates → accumulation of unmineralized osteoid → softening of the bones, impaired growth and malformations

CAUSES

- *Most common:* vitamin D deficiency → insufficient intestinal absorption of Ca^{2+}
- Insufficient sun exposure → UVB rays initiate vitamin D synthesis in skin
- Chronic kidney disease or liver disease → lack of vitamin D activation
- Insufficient intake
  - Malabsorption syndrome → insufficient intestinal absorption of Ca^{2+} and other minerals
  - Maternal deficiencies → congenital rickets
- X-linked hypophosphatemia

RISK FACTORS

- Little sun exposure
- Darker pigmented skin
- Breastfeeding without vitamin D supplementation

COMPLICATIONS

- Bone fractures
- Secondary hyperparathyroidism
- Increased infection risk

SIGNS & SYMPTOMS

- Bone tenderness, pain
- Thinned, soft skull bones, AKA craniotabes; delayed closure of fontanelles
- Bowed legs or knock knees (genu varum or valgus)
- Frontal bossing (enlarged, prominent frontal bone)
- Widening of ankles, wrists; bowing of distal radius, ulna
- Pigeon chest malformation → Harrison's groove along thorax's lower border
- Muscle spasms, numbness
- Hypoplasia of dental enamel

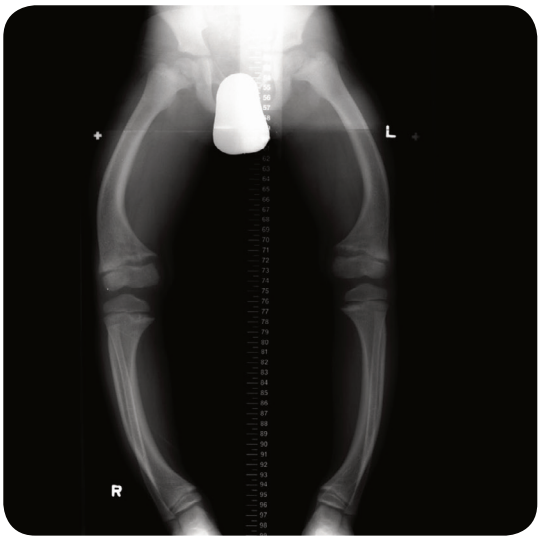

Figure 107.14 An X-ray image of both lower limbs demonstrating bowing in an individual with rickets.

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- ↓ bone mineral density, AKA osteopenia
- Bowed legs
- Widening of epiphyseal growth plate
- Thinning of cortical bone
- Metaphyseal cupping
- Looser lines
  - Transverse lucencies resembling fractures, AKA pseudofractures
- In secondary hyperparathyroidism
  - Subperiosteal resorption of phalanges, bone cysts

LAB RESULTS

Blood tests

- ↓ Serum 25(OH)D levels
- ↓ Ca^{2+}, ↓ PO^{3-}
- ↑ Alkaline phosphatase, ↑ parathyroid hormone levels

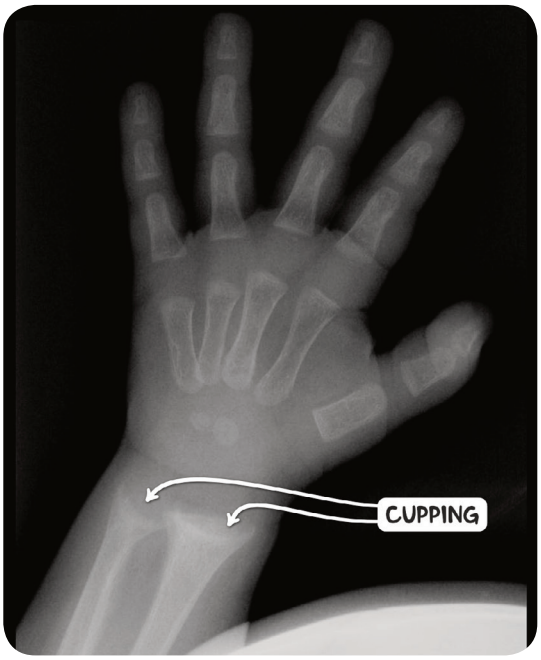

Figure 107.15 An X-ray image of the wrist demonstrating metaphyseal cupping, also known as metaphyseal flaring, in an individual with rickets.

TREATMENT

MEDICATIONS

- Oral vitamin D supplementation
- Correction of calcium intake

OTHER INTERVENTIONS

- Treat underlying causes